GSA2018_APCC69069

A and D genomes spatial separation at somatic metaphase in tetraploid cotton: evidence for genomic disposition in a polyploid plant

APCC6: Chromosome architecture,
epigenetics and cell division

APCC6 Abstract - Oral

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Chromosomal dispositions were analyzed on the metaphase plate of tetraploid cotton (AADD). At metaphase, the two subgenomes, A and D, were separated in a radial pattern in which the small D subgenome chromosomes tended to concentrate at the center and the large A subgenome chromosomes were scattered about the periphery on the metaphase plate. Although the ordered chromosome arrangement was disturbed in an artificial hexaploid (AADDGG), the separation pattern could be recovered after the majority of the additional genome (GG) chromosomes were removed by backcrossing the artificial hexaploid with the tetraploid cotton (AADD). A similar genome separation phenomenon was also found in synthesized tetraploid cotton (AAGG). These results indicate that the genome separation pattern could be established immediately after tetraploid cotton formation and could be stably inherited in tetraploid cotton. Given the evidence of parental genome separation in other plants and animals, we speculated that genome separation might be a normal phenomenon in diploid and polyploid species. These finding will shed light on the chromosome conformation in plant cells.

GSA2018_APCC63028

A fate of sex chromosomes and its partners in sex-limited yellow cocoon (Sy) strain of Bombyx mori

APCC6: Specialised chromosomes (sex chromosomes and B chromosomes)

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The silkworm, Bombyx mori, is domesticated lepidopteran insect (moths and butterflies) which has been scientifically and practically used for a long time. Sex-limited yellow cocoon (Sy) strain was induced by γ-irradiation in order to discriminate male cocoons from females. The W chromosome has been believed to be translocated a long fragment of chromosome 2 carrying yellow blood (Y) gene locus. Our BAC-FISH analysis of the Sy female revealed the W mutation consists of not only chromosome 2 but also chromosome 24 with reciprocal translocation. The female sex chromosomes and the partners (designated as W1, W2, Z, S2S24, Chr2 and Chr24) forms a hexavalent during meiosis. Because of normal karyotype in Sy males the sex chromosome constitution (major type, 2n=56) can be considered to be kept by the hexavalent formation and its segregation. We noticed a few male individuals of the Sy descendant whose growth are delayed with smaller body size in the final achievement. Real-time quantitative PCR (qPCR) demonstrated that 30 among 33 males have only single copies in wide range of chromosome 2 genes tested (approx. 1.5Mb to the end). We also found 3 males were fully single copy carrier of the chromosome 2 genes tested. One of the males (Syr6) was crossed with a p50 (+p/+p) female. The phenotype of hybrid larvae (p50Syr6) segregated to +p : pS = 1 : 1 in the both sexes. Further cross of the +p males to p53 (pY/pY) females segregate to +pY and pY phenotypes with 1 to 1 ratio. Similar qPCR analyses revealed that former +p and the latter pY individuals have single copies of the chromosome 2 genes tested. Hence we suppose that the Syr6 and the descendants are autosomal monosomy that is the second case in animals after 97-years-ago discovery in Drosophila melanogaster.

GSA2018_APCC653143

A new sponge model for regeneration? Sycon capricorn (Calcarea, Calcaronea) regeneration characteristics

GSA: Development and cellular genetics

GSA2018 Abstract - Oral

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Sponges (phylum Porifera) are morphologically simple animals and represent one of the earliest branching extant metazoan lineages. The unique body plan and phylogenetic position establish sponges as an important clade to provide insights into the evolutionary history of metazoan developmental processes. Despite their simple body plan, sponges feature a diverse array of developmental mechanisms. These are regulated by a complex genetic toolkit including homologues of genes with key roles in bilaterian development. Strikingly, sponges are able to deploy this genetic toolkit to accomplish remarkable regeneration after injury or reaggregation of dissociated tissues. However, although detailed descriptions of these processes exist in diverse sponge groups, our knowledge of the underlying molecular mechanisms is currently very limited. Here, we present a comprehensive morphological and histological characterisation of regeneration in the calcareous sponge Sycon capricorn (class Calcarea). S. capricorn displays radial symmetry and a syconoid body plan, a central atrial cavity surrounded by radial chambers housing the sponge “collar cells” - the choanocytes. Regeneration of tissue sections from S. capricorn body column is completed after approximately 5 days and is distinguished by high morphogenetic capacity and axial polarity preservation. This process is morphallactic and accompanied by extensive transdifferentiation of cells (primarily choanocytes). Together with ongoing studies of Sycon development as well as genomic and transcriptomic resources, this framework enables high-resolution single-cell analysis of regeneration in S. capricorn. We aim to establish S. capricorn as a calcisponge model for regeneration research to identify involved cells and signalling pathways and enable more in-depth comparative analysis within metazoans. The high regenerative capacity and relative ease of experimental manipulation make S. capricorn an attractive model to study sponge regeneration providing more insight into the evolution of this important metazoan trait.

GSA2018_APCC668114

A performance evaluation of targeted eDNA monitoring and eDNA metabarcoding to determine fish distributions.

GSA: Ecological genetics

GSA2018 Abstract - Oral

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The use of DNA derived from environmental samples, commonly referred to as environmental DNA or eDNA, is a powerful tool for monitoring the distribution of species. Environmental DNA based monitoring can use either a target approach, through the use of species-specific primers, or entire species communities can be characterized through the amplification of barcoding regions with universal primers and High-Throughput Sequencing of amplicon libraries (i.e. eDNA metabarcoding). While both approaches have been used extensively to monitor the distribution of aquatic species, a formal comparison of their performance is lacking. We will present the results of an extensive eDNA-based monitoring survey conducted in the Blakney Creek catchment (NSW). This catchment contains a population of the endangered Southern pygmy perch (SPP) but the long-term persistence of this population is threatened by the presence and continued spread of two invasive fish species, redfin perch (Perca fluviatilis) and common carp (Cyprinus carpio). Both targeted eDNA monitoring and eDNA metabarcoding was used to determine the distribution of the invasive redfin perch within the system and to evaluate the detection sensitivity of both monitoring approaches. Furthermore, seasonal sampling data was obtained to determine the influence of seasonality on both eDNA-based survey methods.

GSA2018_APCC686146

A pipeline to characterise structural variation in Indigenous Australian genomes

GSA: Bioinformatics and genomics

GSA2018 Abstract - Oral

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Structural variation, large-scale structural differences in genomic DNA, contribute greater genomic diversity at a nucleotide level than any other type of genetic variation, and are linked to a range of Mendelian diseases and complex traits. As such, accurate identification of structural variants from whole-genome sequencing (WGS) data will provide useful information in understanding human genetic diversity and its role in disease. Recently, next-generation sequencing has allowed for improved identification and characterisation of structural variation within human genomes. Compared to long-read sequencing, output from short-read sequencing technologies are less error-prone, more accurate and cost-effective. Because of these properties, short-read analyses are useful in detecting structural variation at a population-level scale. Traditionally, studies analysing short-reads to detect structural variants have used a single caller/program. However, the use of a single caller presents a lack of sensitivity in structural variation detection, as each caller has limitations and biases in integrating signals needed to accurately call specific types of structural variants. As such, pipelines integrating calls from multiple programs will provide a more robust and accurate structural variant callset for downstream analyses.Here, we present a pipeline that utilises output from multiple callers to detect structural variation in human datasets generated using Illumina short-read WGS. This pipeline incorporates the use of traditional callers such as LUMPY (Layer et al., 2014) and DELLY (Rausch et al., 2012), newer programs (SvABA; Wala et al., 2018), and a custom assembly-based pipeline to detect structural

variation in trios sequenced as part of Phase 3 of the 1000 Genomes Project. This pipeline, once sufficiently robust, will be used to detect structural variants within Indigenous Australian genomes at a population-level scale. Such analyses will contribute to the generation of a set of Indigenous Australian-specific reference genomes as part of the National Centre for Indigenous Genomics' Reference Genome Project.

GSA2018_APCC645124

Application of DArT seq derived SNPs for comparative genome analysis

GSA: Evolutionary genetics and comparative genomics

GSA2018 Abstract - Poster

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Bi-allelic Single Nucleotide Polymorphism (SNP) markers of variable length are useful for population genetic studies, where genetic variation is determined by a single nucleotide change in a certain locus among individuals within or between populations. In doing so, a large portion of the sequenced data (either side of the SNPs) remain unused. These data can contain information that is valuable beyond population genetics studies. In this study we used DArTseq (Diversity Array Technology) derived SNP markers developed from a non-model Australian native freshwater fish Golden perch (Macquaria ambigua) to demonstrate the use of such markers for comparative genome analysis.

The specific aim of the study was to identify gene-associated SNP markers and the associated homologies with evolutionary conserved genes. A total of 6,776 markers were concatenated to create a hypothetical genome (representing 3–10% of the actual genome), which was used to find sequence homologies with multiple fish species.

We identified sequence homologies for 18 evolutionary conserved genes (cd9b, plk2b, rhot1b, sh3pxd2aa, si:ch211-148f13.1, si:dkey-166d12.2, zgc:66447, atp8a2, clvs2, lyst, mkln1, mnd1, piga, pik3ca, plagl2, rnf6, sec63, ubr2) along with an ancestral syntenic block (euteleostomi Block_210) conserved across vertebrates. Repetitive sequences covered approximately 12.2% of the hypothetical genome where DNA transposon, LTR and non-LTR retrotransposons are most abundant. A hierarchical pattern of the number of sequence homologies with phylogenetically close species validated the approach for reproducibility. This new approach of using SNP markers for comparative genome analysis may provide insight into the genomes of non-model species in addition to population genetic information

GSA2018_APCC62729

Are neutral genetic diversity and divergence maintained in small populations of Drosophila melanogaster?

GSA: Evolutionary genetics and comparative genomics

GSA2018 Abstract - Oral

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It is known that diversity of neutral loci can be reduced due to adjacent loci under selection (i.e. “linked selection”). On the other hand, it has been hypothesized that linked selection might slow loss of neutral genetic diversity in small

populations. This phenomenon has been termed associative overdominance (AOD). However, whether AOD is a plausible explanation for the maintenance of genetic diversity in small populations is still an open question. An affirmative answer to this question would mean that conclusions based on genetic measures of genetic diversity and divergence could potentially be biased in the cases when they are most needed: the conservation of small, and usually endangered, natural populations. We investigated whether AOD could explain the lower than expected results of genetic divergence of a dispersal experiment. The experiment consisted of 36 pairs of populations of Drosophila melanogaster with known effective population size and dispersal rate. When comparing theoretical expectations against the experiment results, we found that measures of genetic divergence (Fst) were downwards biased on average by 65%. We used agent-based simulations to investigate whether the effective number of recombination events were negatively correlated with the degree of bias. We also investigated whether neutral loci located in regions with a higher number of exons showed larger bias. We used our results to assess the presumed mechanisms by which AOD might affect genetic diversity: the interplay between low recombination rates and selection in small populations. We argue that the interplay between low recombination rates and selection influences neutral genetic diversity. Our research highlights the importance of performing research that focuses on the adjustment of neutral theoretical models by the roles played by genome structure and genetic interactions, such as selection and recombination, which will ultimately allow us to better inform conservation efforts.

GSA2018_APCC658138

Assessing the benefits and risks of translocations in depauperate species: A case study of the conservation management of the critically endangered red-finned blue-eye, Scaturiginichthys vermeilipinnis

GSA: Ecological genetics

GSA2018 Abstract - Oral

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Translocations are increasingly being implemented to assist the conservation of declining native species and decrease their risk of extinction. Translocations must consider demographic, behavioural, ecological and genetic processes. As threatening processes continue to decrease population sizes and impose localised population extinctions, a species' genetic diversity is also likely to decline. Establishing new populations via translocations can help spread the risk of sporadic events leading to extinction in small, isolated populations, thereby increasing the overall survival probability of the species. But initiating new populations comes with two main challenges: i) the genetic diversity of translocated populations will be reduced compared to the source, and ii) removal of founder individuals will decrease genetic diversity in source populations, placing them at greater extinction risk. We present an empirical study on the conservation of red-finned blue-eye, Scaturiginichthys vermeilipinnis, which has recently undergone severe population extirpation, being reduced to a single remnant population. Several translocated populations have been established with founders sourced from this remnant population. Several generations after re-introduction, we show that translocated populations (both individually and combined) harbour reduced genetic

diversity compared to the remnant population. This is due to i) failure of the founders to capture all the genetic diversity present in the source population, and ii) a failure to retain the genetic diversity of the founders across generations through unbalanced breeding contributions. We studied the translocation trade-off between increasing the species probability of survival and decreasing the genetic diversity and derive general principles for the translocation of highly endangered populations. Finally we argue, prior to undertaking translocations, the trade-off between extinction risk and retention of genetic diversity needs to be determined to establish the optimal conservation strategy to increase the long-term persistence and evolutionary potential of a species.

GSA2018_APCC6946

Asynchronous development of sexual phenotypes causes temporary pseudohermaphroditism in female bearded dragons (Pogona vitticeps)

GSA: Development and cellular genetics

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For Australia's native Bearded Dragon (Pogona vitticeps), sex is complicated. They exhibit a genetic sex determination system (GSD) where females have a Z and ZW chromosome (ZWf) while males have two Z chromosomes (ZZ). However, if incubated at high temperatures (>32°C) genetic males reverse their sex and develop as phenotypic females (ZZf). When sex reversed females mate with genetic males, their offspring's sex is determined by temperature alone (temperature dependent sex determination, TSD), causing a change of sex determination system in one generation. While this was observed in adults, nothing was known about how this process occurred during embryonic development. By conducting incubation experiments that yielded all sexes (ZWf, ZZF and ZZ), we found that the pathway for female development was unaffected by the sex determination cue (temperature or sex chromosomes). Notably, all females developed hemipenes (male genitalia) before regressing them close to hatching. Histological examination of the gonads revealed differentiation occurs far earlier than complete hemipenis regression, which creates a prolonged period of temporary pseudohermaphroditism (TPH). Using scanning electron microscopy (SEM), we further discovered two novel hemipenal ultrastructures (hemipenal furrow and lattice) that are shared between males and females (ZWf and ZZf). As female hemipenis retention makes sexing embryos problematic, we developed a novel genotyping technique that allowed us to non-destructively obtain a sample of DNA for PCR based sex identification. It is generally assumed that the dimorphic development of genitalia begins shortly after gonad differentiation. However, the results of this study show this assumption does not hold true in this species. A close investigation of the limited literature published on female sexual development revealed there are two other species (Niveoscincus ocellatus and Barisia imbricata) also exhibit TPH. Combined these results reveal that female sexual development in squamates is considerably more complex than currently appreciated.

GSA2018_APCC64963

B chromosomes – a matter of chromosome drive

APCC6: Specialised chromosomes (sex chromosomes and B chromosomes)

APCC6 Abstract - Oral

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B chromosomes (Bs) are supernumerary chromosomes which are often preferentially inherited, deviating from usual Mendelian segregation. The balance between the so-called chromosome drive and the negative effects that the presence of Bs applies on the fitness of their host determines the frequency of Bs in a particular population. Advances in genome analysis and chromosome imaging improved our knowledge on the origin and drive of Bs. In contrast to the prevalent view that Bs do not harbour genes recent analysis revealed that Bs of sequenced species are rich in gene-derived sequences. I will summarize the latest findings on supernumerary chromosomes of rye (Secale cerelae) and the goatgrass Aegilops speltoides with a special focus on the origin and drive of B chromosomes. Novel CRISPR-Cas9 based chromatin imaging methods will be introduced to elucidate the spatio-temporal organization of the genome inside the nucleus. How novel analytical tools will expand our ability to uncover the biology of B chromosomes will be discussed.

GSA2018_APCC64265

Bizarro genomics: harnessing the chiral properties of DNA sequences for precision medicine

GSA: Bioinformatics and genomics

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Chirality is a geometric property describing any object that is inequivalent to a mirror image of itself. Due to its 5’-3’ directionality, a DNA sequence is distinct from an exact copy arranged in reverse sequence order, and is therefore chiral. A DNA sequence and its chiral partner sequence share many attributes, such as nucleotide composition and sequence entropy. Here we demonstrate that chiral sequence pairs also perform equivalently during bimolecular processes and bioinformatic analysis. We establish equivalence between chiral pairs of DNA sequences during many of the techniques that underpin genetic analysis, including PCR amplification, hybridization, whole genome, target-enriched and nanopore sequencing, sequence alignment and variant identification. Given these shared properties, we have developed synthetic chiral DNA reference standards that directly mirror clinically relevant regions of the human genome, disease-causing mutations and/or analytically challenging features. We show how these chiral sequences can act as commutable internal controls to measure and improve diagnostic performance during genome analysis, and improve variant interpretation for precision medicine.

GSA2018_APCC662163

Case study: Next-generation Phenotyping for Mucopolysaccharidosis with automated image analysis

GSA: Bioinformatics and genomics

GSA2018 Abstract - Oral

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Introduction Reducing the time-to-diagnosis for patients with rare genetic syndromes is a goal for many, especially where there are therapies available or active clinical trials. Some of these syndromes are known to have a facial phenotype, which is highly informative, and even discriminative, for clinical geneticists diagnosing patients. The use of computerised systems as clinical reference tools is therefore becoming increasingly important. In this study we sought to measure the effectiveness of a next-generation phenotyping technology that utilises Artificial Intelligence and Deep Learning algorithms to analyse 2D facial images of patients diagnosed with different types of Mucopolysaccharidosis (MPS). Method Images of consented and diagnosed patients with MPS were uploaded to Face2Gene, a suite of applications using next-generation phenotyping to detect facial patterns and match them with mathematical descriptors representing phenotypes of known genetic disorders hence providing a reference for clinicians diagnosing such patients. To compare the different MPS phenotypes, we used the Face2Gene RESEARCH application that generates a classification model using two or more cohorts of patients' photos. ROC curves and the respective areas under the curve were computed for comparisons. P-values were calculated based on 1000 random permutations. Results The analysis was performed by comparing each cohort of photos to one other cohort or to a combination of all other cohorts together. Results clearly show the ability of the algorithm to differentiate between the different types of MPS faces. p-values are ≤ 0.01 showing statistical significance and all AUC values obtained in the 21 comparisons conducted are > 0.74 with 19 experiments having an AUC ≥0.8, of which 11 have an AUC>0.9, with a low standard deviation. Conclusion The technology powering Face2Gene has the ability to recognize the phenotype of different types of Mucopolysaccharidosis and therefore could be used in the future for early screening of patients, leading to an earlier diagnosis and timely therapy.

GSA2018_APCC646125

Characterization of chromosome-specific and telomere repeats in Panax ginseng based on low-coverage whole-genome sequence reads

APCC6: Cytogenetics in the genomics era (Plants and Animals)

APCC6 Abstract - Poster

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Repetitive DNA elements are ubiquitous in plant genomes. Although repeats provide relevant information for cytogenetic, evolutionary, and genomic studies, identifying and characterizing their sequence and chromosomal distribution are not always easily achieved through conventional methods. However, a high-throughput identification of genomic repeats can be obtained with short reads from next-generation sequencing data. Here, we identified the telomeric and three chromosome-specific repeats in Panax ginseng using low-pass whole genome sequence data. The telomeric repeat sequence is same with the canonical angiosperm sequence, TTTAGGGn, and localized mostly in chromosome termini, except for an interstitial location in chromosome 10. A dinucleotide (GA) microsatellite, PgGA15, with total genome representation (GR) of more than 33 kb localized in the long arm of chromosome 20. An 11-bp minisatellite, Pgms1, with GR of more than 58 kb localized in the long arm of chromosome 1. Finally, a 201-bp satellite repeat, Pg201TR, with a GR of about 342 kb localized in the long arm of chromosome 8 and, at the NOR region of chromosome 14. Sequence and GISH analyses of Pg201TR suggest its evolutionary association with P. ginseng 45S rDNA intergenic spacer. These data provide chromosome-specific markers for cytogenetic studies of P. ginseng and a clue to the 45S rDNA evolution in Panax. This work was carried out with the support of the Cooperative Research Program for Agriculture Science & Technology Development (PJ013119), Rural Development Administration, Republic of Korea.

GSA2018_APCC67319

Chicken BACs and repeat element mapping suggest processes of chromosomal rearrangement and W chromosome differentiation in Siamese cobra compared to most other snake karyotypes

APCC6: Specialised chromosomes (sex chromosomes and B chromosomes)

APCC6 Abstract - Oral

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Contrary to the chromosome constitution of most snakes (2n=36), the cobra karyotype shows diploid chromosome number at 38. To investigate karyotype differences and the process of chromosomal rearrangement between cobras and most other snakes, chromosomics (cytogenetics and genomics) was conducted in Siamese cobra (Naja kaouthia). Siamese cobra chromosome 2 (NKA2) is subtelocentric, differing with those of most other snakes which are submetacentric. Chicken BAC probes, comprising the chicken Z chromosome (GGAZ) and GGA12, GGA13, and GGA18 were almost mapped to NKA2 in the same order as on most other snake chromosome 2, suggesting that pericentric inversion or centromere repositioning occurred in the cobra lineage. GGA2p and GGA27 were localized to NKAZ, and were similar to those of most other snakes, but not for NKAW. In addition, large C-positive blocks were observed on the entire arm of NKAW. Hybridization signals of telomeric repeat (TTAGGG)n and two microsatellite motifs (AAGG and AGAT) were also observed interstitially at the same position as large C-positive blocks on NKAWq, suggesting that the amplification of telomeric repeat and microsatellite motifs are strongly associated with differentiation and heterochromatinization of sex chromosomes in Siamese cobra. One chicken GGAZ BAC clone (CH261-129A16), comprising various microsatellite motifs and transposons was located in NKAWq. This suggests that repetitive sequences are shared between the sex chromosomes of chicken and cobra, and supports the hypothesis of an ancestral super-sex chromosome in amniotes. Degeneration of sex chromosomes was probably convergent between birds and snakes.

GSA2018_APCC641128

Chromosomal distribution of diversified chromatin modification marks in Asian orchids

APCC6: Cytogenetics in the genomics era (Plants and Animals)

APCC6 Abstract - Oral

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Orchidaceae with ≈25,000 species of 800 genera is one of the largest families among angiosperms. Out of which, Dendrobium, Cymbidium, Vanda, and Phalaenopsis originated from Asia are considered as highly economical valued orchids. The chromosome/nuclear structure, especially with respect to the dynamics of rapidly evolving chromatin have been studied at various cell divisions including mitosis, meiosis and pollen mitosis in orchids. We investigated chromosomal distribution patterns of chromatin modifications with special emphasis on histone methylation, acetylation, phosphorylation, and centromere-specific histone 3 (CENH3) marks in orchids using immuno-FISH. The chromosomal distribution of 5-mC suggests that the genomic DNA of orchids is highly methylated and CG dinucleotide makes methylated hotspots on entire chromosomes. Various orchid species revealed diversified pattern of DNA methylation profile. Furthermore, such epigenetic regulatory post-translational histone modification marks not only function individually but also capable to act in combination as a unique pattern. The dual modified histone H3S10phK14ac that eventually targets the peri-centromeric position of chromosomes may serve as a new cytological marker to identify centromere in plants especially in orchids where the cytological/chromosomal preparations are very difficult. The chromosomal distribution of eu-/heterochromatic-specific histone marks as well as phosphorylation of H3S10 and H3T3 histone during pollen mitosis I in orchids have also been investigated. Detailed analysis suggest that the phosphorylation of histone plays a critical role in chromosome condensation events throughout pollen mitosis I in plants including orchids. Spatial distribution of chromatin environment that exists between different cell types such as diploid/polyploids, vegetative/generative cells has also been discussed.

GSA2018_APCC617157

Chromosomal evolution in cichlid species flock from the Barombi Mbo lake

APCC6: Comparative, evolutionary and population cytogenetics

APCC6 Abstract - Poster

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The African cichlid fish radiations are the most diverse extant vertebrate radiations and provide a unique opportunity to study inferences on chromosomal evolution and cytotaxonomy. In this study we examined 11 morphologically distinct endemic cichlid species from Barombi Mbo, the largest crater lake along the Cameroon Volcanic Line in western Cameroon. These species apparently evolved via sympatric speciation from a single common ancestor, which colonized the lake approximately one million years ago. An extensive comparative cytogenetic analyses was performed in representatives of these species using both conventional (Giemsa staining, C-banding, Ag-NOR staining and CMA3/DAPI staining) and molecular (fluorescence in situ hybridization with 5S and 28S rDNA probes) methods. Despite the karyotype analyses showed no significant differences in chromosome number and karyotype morphology, we observed variability on the sub-chromosomal level. The first cytogenetic report for cichlid species flock from Barombi Mbo lake likely documents another extra ordinary case of chromosomes stasis in geologically significant timespan. Acknowledgements: Czech Science Foundation (project No.16-09784Y) and Basler Stiftung fuer biologische Forschunga Basler Stiftung fuer experimentelle Zoologie.

GSA2018_APCC65377

Chromosomal Location and Characterization of Long Terminal Repeat of Endogenous Retrovirus in the Olive Flounder

APCC6: Cytogenetics in the genomics era (Plants and Animals)

APCC6 Abstract - Poster

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Chromosomal Location and Characterization of Long Terminal Repeat of Endogenous Retrovirus in the Olive Flounder Hee-Eun Lee1,2, A-Ra Jo1,2, Hyun Hee Kim3,4, Hee-Jae Cha5, Heui-Soo Kim1, 2* 1 Department of Biological Sciences, College of Natural Sciences, Pusan National University, Busan 46241, Republic of Korea 2 Institute of System Biology, Pusan National University, Busan 46241, Republic of Korea 3 Departments of Life Science, Sahmyook University, Seoul 01795, Republic of Korea 4 Chromosome Research Institute, Sahmyook University, Seoul 01795, Republic of Korea 5Department of Parasitology and Genetics, College of Medicine, Kosin University, Busan 49267, Korea *Corresponding author: khs307@pusan.ac.kr Abstract Endogenous retrovirus (ERV) family has been identified with different copy numbers in various organisms. Long terminal repeat (LTR) of ERV has the capacity to exert regulatory influence as promoters and enhancers of cellular genes. Here, we identified OF-ERV9 derived from chromosome 9 of Olive flounder. The genomic structure of 5′LTR-gag-pro-pol-env-3′LTR showed truncated form. FISH data demonstrated their location on chromosome 4, 5, 6, 9, and 13. The 5′LTR elements of OF-ERV9 provided sites for binding of various transcription factors, indicating enhancer activity in cell lines. The OF-ERV9-LTR showed high sequence similarity with 3′ untranslated region (UTR) of functional genes (mkln1, slc37a3, LOC109642524, LOC109644478, LOC109626170, LOC109636349). Those genes also contain seed sequences (TGTTTTG) to bind LTR-derived microRNA (OF-miRNA307). The mkln1 gene expression was in contrast to OF-miR307 expression in various tissues of Oliver flounder. Taken together, our data could be of great use for understanding the molecular function of OF-ERV families in Olive flounder.

GSA2018_APCC673179

Chromosome and genome structure as a basis for assigning function to genes under-pinning wheat agronomic traits.

APCC6: Cytogenetics in the genomics era (Plants and Animals)

APCC6 Abstract - Oral

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Chromosome and genome structure as a basis for assigning function to genes under-pinning wheat agronomic traits. Rudi Appels, Hon Professor at University of Melbourne and LaTrobe University (AgriBio), Melbourne Victoria, Australia The centromere region is a classical cytological feature of chromosomes and can harbour genes of fundamental importance to wheat. The rapid advances in the structure analysis of the 21 chromosomes of wheat will be presented with a focus on the centromere of chromosome 7A in the chromosome per se as well as the telosomes 7AS and 7AL. The outputs from the research define a valuable relationship to the syntenic rice chromosome 8, the biology of genes located in the centromere and the location of the kinetochore protein CENH3. The study reveals the centromere to be flexible in structure while contributing a stable (low recombination) environment for a gene space that is essential for wheat productivity.

GSA2018_APCC66210

Chromosome architecture in human and gibbons: Evolutionary consideration for radial 3D arrangements of chromosome territories

APCC6: Chromosome architecture,
epigenetics and cell division

APCC6 Abstract - Oral

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Individual chromosomes are occupied as “chromosome territories (CTs)” within the nucleus. How are CTs spatially localized? What kind of factors can be involved to arrange their positioning? From studies for over two decades it has been revealed that the radial distribution of CTs from center to nuclear rim has the following characteristics. 1) It depends on the physical size and gene density of each CT; larger gene-poor CTs are located toward periphery and smaller gene-dense CTs are located into interior of the nucleus. 2) Evolutionarily syntenic chromosomal regions of CTs are inclined to localize the same radial positioning, but it is influenced by the pericentromeric heterochromatin. 3) Lamin asssociated domains (LADs) might be affected to be tethered near the nuclear rim. 4) Actin related protein 6 (Arp6), one of the ubiquitous components of chromatin remodeling complexes, has affected to global nuclear radial distribution of CTs. Arp6-knock out chicken DT40 cells have shown disturbed global nuclear architecture. In summary, spatial radial distribution of CTs can be affected strongly with physical properties whereas less affected with the status of gene expression or epigenetic factors. In the present study I focused on the gibbons showing the highest speed of chromosome evolution in mammals. Approximately over 90 evolutionary conserved breakpoints (ECBs) have been observed between human and gibbons. To investigate spatial organization in human and gibbons, radial 3D arrangements were analyzed by 3D-FISH with CTs, ECBs, and repetitive DNA sequences. The results showed Alu and LINE1 sequences were located near the nuclear center and periphery, respectively, however, centromeric satellite DNAs were detected as relatively stronger fluorescent signals near the center than those of periphery suggesting that radial 3D arrangements have reflected the different distributed number of repetitive elements between periphery and interior nuclear space. Organization of nuclear architecture from evolutionary views will be discussed.

GSA2018_APCC678160

Chromosome Instability of Stem Cell Lines on Applications in Regenerative Medicine

APCC6: Applied cytogenetics (e.g agriculture and biomedical)

APCC6 Abstract - Oral

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Abstract It has passed over a decade since the establishment of iPS cells (induced Pluripotent Stem cells). Studies on clinical applications of a variety of tissue stem cells in the field of regenerative medicine have exponentially increased. However, strict quality control of the tissue stem cells is the most importantly required task for clinical treatment in safe, so as the mechanism of re-programing and gene regulation in early embryonic development has not been fully understood yet. In the present report, I'd like to provide examples of evidence of chromosome instabilities in the established tissue stem cell lines, such as in cases of an ES cell line derived from crab-eating monkey, human iPS cell lines and cord blood derived mesenchymal stem cells. Taken together from the results of above mentioned data and knowledge from recent literatures, I hope to discuss with the usefulness of karyotyping especially in its early passage of the establishment in order to guarantee the qualities of the stem cells.

GSA2018_APCC612175

Chromosome-level genome assembly and reconstruction of evolutionary events in birds and other dinosaurs

APCC6: Cytogenetics in the genomics era (Plants and Animals)

GSA2018 Abstract - Oral

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Darren K. Griffin1*. Rebecca E. O'Connor1, Michael N. Romanov1, Joana Damas2, Marta Farré2, Henry Martell1, Lucas Kiazim1, Rebecca Jennings1, Anjali Mandawala3, Sunitha Joseph1, Katie E. Fowler3, Eden A. Slack2, Emily Allanson2, Malcolm Ferguson-Smith4, Paul M. Barrett5, Nicole Valenzuela6, Denis M. Larkin2 1 School of Biosciences, University of Kent, Canterbury, UK 2 Department of Comparative Biomedical Sciences, Royal Veterinary College, University of London, London, UK 3 School of Human and Life Sciences, Canterbury Christchurch University, Canterbury, Kent, UK 4 Department of Veterinary Medicine, Cambridge University, Cambridge, UK 5 Department of Earth Sciences, Natural History Museum, Cromwell Road, London SW7 5BD, UK 6 Department of Ecology, Evolution, and Organismal Biology, Iowa State University, USA The ultimate aim of a genome assembly is to create a contiguous length of sequence from the p- to q- terminus of each chromosome. Most assemblies are however highly fragmented, limiting their use in studies of gene mapping, phylogenomics and genomic organisation. To overcome these limitations, we developed a novel scaffold-to-chromosome anchoring method combining reference-assisted chromosome assembly (RACA) and fluorescence in situ hybridisation (FISH) to position scaffolds from de novo genomes onto chromosomes. Using RACA, scaffolds were ordered and orientated into 'predicted chromosome fragments' (PCFs) against a reference and outgroup genome. PCFs were verified using PCR prior to FISH mapping. A universal set of FISH probes developed through the selection of conserved regions were then used to map PCFs of peregrine falcon (Falco peregrinus), pigeon (Columba livia), ostrich (Struthio camelus), saker falcon (Falco cherrug) the budgerigar (Melopsittacus undulatus). Using this approach, we were able improve the N50 of genomes seven-fold. Results revealed that Interchromosomal breakpoint regions are limited to regions with low sequence conservation, shedding light on why most avian species have very stable karyotypes. Our combined FISH and bioinformatics approach represents a step-change in the mapping of genome assemblies, allowing comparative genomic research at a higher resolution than previously possible. The universal probe set, facilitates research into avian karyotype evolution and the role of chromosome rearrangements in adaptation and phenotypic diversity in birds. Indeed, they have been used on over 20 avian species plus non-avian reptiles (including turtles), shedding light into the evolution of dinosaur species. Non-avian dinosaurs remain subjects of intense biological enquiry while pervading popular culture and the creative arts. While organismal studies focus primarily on their morphology, relationships, likely behaviour, and ecology there have been few academic studies that have made extensive extrapolations about the nature of non-avian dinosaur genome structure prior to the emergence of modern birds. We have used multiple avian whole genome sequences assembled at a chromosomal level, to reconstruct the most likely gross genome organization of the overall genome structure of the diapsid ancestor and reconstruct the sequence of inter and intrachromosomal events that most likely occurred along the Archosauromorpha-Archosauria-Avemetatarsalia-Dinosauria-Theropoda-Maniraptora-Avialae lineage from the lepidosauromorph-archosauromorph divergence ~275 mya through to extant neornithine birds.

GSA2018_APCC637129

Chromosome-specific painting in rice using bulked oligonucleotides and its applications

APCC6: New and emerging chromosome technologies

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The chromosome-specific probe is a fundamental tool of chromosome painting and has been commonly applied in mammalian species. The technology, however, has not been widely applied in plants due to a lack of methodologies for probe development. Identification and labeling of a large number of oligonucleotides (oligos) specific to a single chromosome offers us an opportunity to establish chromosome-specific probes in plants. However, never before has whole chromosome painting been performed in rice. We developed a pooled chromosome 9-specific probe in rice, which contains 25,000 oligos based on the genome sequence of a japonica rice (Oryza sativa, AA, 2n=2x=24). Chromosome 9 was easily identified in both japonica and indica rice using this chromosome 9-painting probe. The probe was also successfully used to identify and characterize chromosome 9 in additional linesof O. sativa, a translocation line, two new aneuploids associated with chromosome 9 and a wild rice (Oryza eichingeri, CC, 2n=2x=24). The study reveals that a pool of oligos specific to a single chromosome is a useful tool for chromosome painting in rice.

GSA2018_APCC635174

Chromosomes in the Light of Evolution

APCC6: Comparative, evolutionary and population cytogenetics

APCC6 Abstract - Oral

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Chromosome rearrangements are a hallmark of genome evolution and essential for understanding the mechanisms of speciation and adaptation. Determining the types and chronology of chromosome rearrangements over evolutionary time scales has been a difficult problem due primarily to the lack of high quality, chromosome-scale genome assemblies that are necessary for reliable reconstruction of ancestral genomes. In addition, for genome-wide comparisons that require resolving large numbers of rearrangements of varying scale, determining ancestral chromosomal states is challenging both methodologically and computationally. We recently developed a new computational tool for reconstructing ancestral chromosomes at high resolution, called DESCHRAMBLER, which uses syntenic fragments constructed from whole-genome comparisons of both high quality chromosome-scale and fragmented assemblies in a phylogenetic probabilistic framework. We applied DESCHRAMBLER to sequenced genomes of 21 species that included representatives of 10 eutherian orders. Seven ancestral genomes leading to human were reconstructed, including the ancestor of all placental mammals. These reconstructions revealed a detailed history of chromosome rearrangements that occurred during ~105 million years of eutherian evolution. Our results provide an evolutionary basis for comparison of the genome organization of all eutherians that will facilitate greater understanding of the role of chromosome rearrangements in adaptation, speciation, and the etiology of inherited and spontaneously occurring diseases. With ongoing efforts to sequence vertebrate genomes from all classes, it will be possible to extend chromosome reconstructions deeper into evolutionary time. Sequence-based reconstruction of ancestral chromosomes thus provides a new opportunity to bridge the gap between traditional cytogenetics and genomics by illuminating the origins of ancestral synteny and the timing of chromosome rearrangements during evolution.

GSA2018_APCC647156

Chromosomes of sexual and unisexual Australian carp gudgeons (Hypseleotris, Gobioidei: Eleotridae)

APCC6: Comparative, evolutionary and population cytogenetics

APCC6 Abstract - Poster

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Carp gudgeons (Hypseleotris) belong to a group of Australian native freshwater fishes, which includes sexually reproducing species as well as hybridogenetic biotypes that reproduce by mating with coexisting sexual congeners. Here, we present the first cytogenetic analysis in four sexual species and one hybrid biotype of Hypseleotris using standard and molecular cytogenetic protocols (e,g. Giemsa staining, C-banding, Ag-NOR staining and CMA3/DAPI staining, FISH with 5S and 28S rDNA probes). Our data showed no differences in diploid chromosome number however, we observed variability in chromosome morphologies, and variability on the sub-chromosomal level, such as different accumulation of repetitive sequences. Our data provides the first insight into chromosome morphologies of this unique unisexual fish species complex and will provide basis to discover cytogenetic mechanisms in the evolution of unisexuality in these non-traditional model organisms. Acknowledgements: The study has been supported by the project No. MSM200451701 from the Czech Academy of Sciences and the project EXCELLENCE CZ.02.1.01/0.0/0.0/15_003/0000460 OP RDE and RVO: 67985904.

GSA2018_APCC630180

Chromosomes Unbalanced: aneuploidy, deletions, and duplications in human pluripotent stem cells.

APCC6: Applied cytogenetics (e.g agriculture and biomedical)

APCC6 Abstract - Oral

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Chromosomes Unbalanced: aneuploidy, deletions, and duplications in human pluripotent stem cells. Karen Dyer Montgomery, PhD, FACMG Karen Dyer Montgomery Genetic Consultants, LLC, Santa Fe, USA The potential use of human stem cell lines for therapy makes it essential to ensure that they have a normal genome. They often don't. Recurrent numerical and structural chromosome imbalances are acquired by pluripotent stem cells (hPSCs) during culture. Full and partial trisomies of chromosomes 8, 12, 17, 20 and X are common. Structural changes resulting in gains in critical regions of the long arm of chromosome 1 and chromosome 20 are not unusual. Abnormalities are similar between human embryonic stem cells and induced pluripotent stem cells. The changes are also consistent among varied derivation protocols, culture conditions, and geographic locations. Questions arise as to the cause and effect of this phenomenon. The tendency of hPSCs to become cytogenetically abnormal may have substantial impact on their use in regenerative medicine.

GSA2018_APCC666185

Classifying ENU induced mutations from spontaneous germline mutations in mouse with machine learning techniques

GSA: Bioinformatics and genomics

GSA2018 Abstract - Poster

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Understanding sequence diagnostic signatures associated with mutagenesis mechanisms can facilitate the development of more accurate models for identifying disease causing mutagens. In most genetic variation catalogs, variant data are derived from a mixture of mutagenic processes. This presents a challenge to assigning the mechanistic origins of an individual variant. Information regarding the mechanistic origin of point mutations is present in surrounding DNA sequence. These motifs can reflect the combination of chemical and biochemical influences of neighbouring bases on mutagenesis. We assess whether information from sequence neighborhood can be used to identify mutations resulting from the potent chemical mutagen, ENU. ENU is a synthetic chemical employed in mutagenesis studies, introducing novel point mutations to genomes. We developed a machine learning classifier to discriminate between ENU-induced and spontaneous point mutations in the mouse germline. Our classification results reveal that a combination of k-mer size and representation of second-order interactions among nucleotides was able to improve classification performance in comparison to the naive classifier approach.

GSA2018_APCC628113

Climate-driven mito-nuclear selection in Australian birds: developing a multidisciplinary approach

GSA: Evolutionary genetics and comparative genomics

GSA2018 Abstract - Oral

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Diversifying selection between populations that inhabit different environments can promote lineage divergence within species and drive speciation. The mitochondrial genome and nuclear genes with mitochondrial function can be a target for climate-driven selection because they co-encode essential proteins of the oxidative phosphorylation system or regulate system functioning. Understanding evolutionary drivers in wild systems requires integration of comparative and population genomics and studies of physiology and fitness. Multi-species investigation into intraspecific mitochondrial variation showed that in half of the widespread Australian songbirds climate significantly predicts distribution of mitochondrial variation beyond that predicted by geographic distances and geographic position. Analyses of mitogenomes yielded candidates for positive selection in some lineages of some species. In one of these, the Eastern Yellow Robin, north-south lineage divergence ~2 million years ago was followed by two adaptive mitochondrial introgressions. Now, inland and coastal populations occupy areas with different climates and harbour divergent mitolineages despite similarity for majority of nuclear genes. But these populations are differentiated at a small subset of nuclear genes that map to a ~15 megabase region on autosome 1A. This region is characterised by strong linkage disequilibrium and low genetic diversity and is enriched by genes with roles linked to mitochondrial functions (N-mt genes). Elevated differentiation and sequence divergence at this region suggest that N-mt genes might act as barrier loci resisting gene flow. Similar regions of differentiation on chromosome 1A are found among other songbirds, and may present a common mechanism facilitating climate adaptation. We are testing whether the two genomic forms of Eastern Yellow Robin are adapted to their local environments via traits connected with mitochondrial metabolism.

GSA2018_APCC61150

CmSat162 and CmSat189, new satellite DNAs in Cucumis melo L.

APCC6: Comparative, evolutionary and population cytogenetics

APCC6 Abstract - Oral

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Melon karyotyping still remains a challenge, due to not only the small chromosome sizes and poor stainability but also the limitation of cytological markers which commonly rely on ribosomal RNA genes (45S rDNA and 5S rDNA) and centromere repeat as the probes. Satellite DNAs (SatDNAs), occupying significant portion in Cucurbitaceae within their genomes, are making them valuable markers for chromosome identification. By utilizing melon genome database, we identified and found new SatDNAs in melon, namely CmSat162 and CmSat189, which were successfully hybridized to melon somatic cells, pachytene chromosomes and extended DNA fibers. CmSat162, a new centromeric SatDNA, was hybridized into 12 pairs of melon chromosomes and co-localized with Cmcent probe. In addition, CmSat189, located on specific region of each chromosome, allowed the discrimination and characterization of individual homologous chromosomes in melon. We also demonstrate that the transcription of these SatDNAs may have important function for maintaining centromere structures in melon. Our results suggest that CmSat162 and CmSat189 are useful for melon karyotyping. Additionally, those probes may contribute to integrate the physical, chromosomal and genetic maps for genome sequencing project.

GSA2018_APCC697153

Comparative cytogenetics in ancestral teleost fishes: karyotype characterisation from two species of Australian Saratoga, Scleropages leichardti and Scleropages jardini (Osteoglossidae)

APCC6: Comparative, evolutionary and population cytogenetics

APCC6 Abstract - Oral

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Despite substantial progress fish cytogenetics, there are still several gaps in our knowledge about genome evolution and organization in one of the early branching lineages of teleosts - the Osteoglossomorpha. Here, we present comparative cytogenetic study of its two Sahulan species Scleropages leichardti (Günther, 1864) and S. jardinii

(Saville-Kent, 1892). To characterise their karyotypes we applied both conventional (Giemsa staining, C-banding, Ag-NOR staining and CMA3/DAPI fluorescence) and molecular (FISH with rDNA and microsatellite probes, CGH) cytogenetics techniques. Karyotype analyses showed differences in diploid chromosome number between S. leichardti (2n = 44) and S. jardinii (2n = 48), with morphology and variability on the sub-chromosomal level, such as different accumulation of heterochromatin. Moreover, we uncovered heterogeneous composition of AT/GC rich regions on chromosomes of both species. Among actinopterygians, cytogenetically detectable AT/GC heterogeneity has so far only been documented in gars (Lepisosteiformes, another early branching teleost lineage). We argue that this genome feature is ancestral to all teleosts and has been lost in most recent lineages. Our data provided valuable insights into early karyotype evolution and genome compartmentalization in actinopterygians. Acknowledgements: The study has been supported by the project No. MSM200451701 from the Czech Academy of Sciences and by CAPES/Alexander von Humboldt Foundation (Process 88881.136128/2017-01) and the project EXCELLENCE CZ.02.1.01/0.0/0.0/15_003/0000460 OP RDE and RVO: 67985904.

GSA2018_APCC674155

Comparative cytogenetics in Australian rainbowfishes (Melanotaeniidae)

APCC6: Comparative, evolutionary and population cytogenetics

APCC6 Abstract - Oral

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Rainbowfishes (Melanotaeniidae) is the largest monophyletic group of freshwater fishes found in Australia and New Guinea, with 109 species currently described. Despite its high diversity, this group remains poorly studied cytogenetically, since karyotype data are available from only about 5% of species. In this study, we describe

karyotypes of three Australian rainbowfish species, Running River rainbowfish (Melanotaenia sp.), red rainbowfish Glossolepis incisus and Cairns rainbowfish Cairnsichthys rhombosomoides, representing two close related and one more divergent rainbowfish species. All three species has diploid 2n = 48. We also analysed heterochromatin distributions by C-banding, AT/GC organization of genome by CMA3 staining, location of rDNA sites and location of TTAGGG telomeric sequences by FISH. No sex chromosome heteromorphy was detected using standard and advanced cytogenetic (e.g. CGH). Our data suggest uniformity in Melanotaeniidae diploid number, however, we detected variable chromosome structures and variability in heterochromatin distributions and NOR location. Results of this study shed light into genome evolution within Rainbowfishes. Acknowledgements: The study has been supported by the project No. MSM200451701 from the Czech Academy of Sciences and the project EXCELLENCE CZ.02.1.01/0.0/0.0/15_003/0000460 OP RDE and RVO: 67985904.

GSA2018_APCC68713

Comparative genomics through the development of universal cross-species BAC sets

APCC6: Comparative, evolutionary and population cytogenetics

APCC6 Abstract - Oral

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Comparative genomics using targeted BAC based FISH (fluorescence in situ hybridization) probes, is generally restricted to closely related species due to extensive sequence divergence between distantly related species. The ability to identify precise regions of homology between species (for genome mapping, phylogenomics and genome organisation studies) and the ability to anchor genomic sequence data to chromosomes (for chromosome assembly) is therefore restricted. To overcome these difficulties, we developed a set of universal avian BAC probes, selected through the identification of evolutionary conserved regions. This BAC set was then used to upgrade the genomes of 5 avian species to a chromosome-level. Successful hybridisation of these probes to a further ~30 avian species revealed genome-wide patterns of chromosome stability and rearrangement between species. In addition, the probes successfully hybridized on non-avian reptile species (turtles and anole lizard) revealing a level of genome conservation extending far beyond birds. Further, we applied the approach developed for avian probes to the

selection of BACs from the cattle and human genome with the aim of generating a universal mammalian BAC set. Selection criteria were validated by testing probes on species at key nodes of the phylogenetic tree. Hybridisations were achieved on species as diverse as horse (Equus ferus), dolphin (Tursiops aduncus), bat (Lophostoma silvicolum) and lemur (Eulemur macaco). These preliminary results illustrate that our combined FISH-bioinformatics approach is also applicable to mammals. Development of a universal BAC set therefore permits cross-species sequence anchoring and comparative genomic research at a higher resolution than previously possible, providing new insight into the nature of genomic evolution and genomic stability.

GSA2018_APCC6747

Comparative phylogeography: How dispersal rates influence beta diversity of species

GSA: Ecological genetics

GSA2018 Abstract - Oral

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Species dispersal rates vary depend on intrinsic movement rates and species habitat preference and are predicted to influence rates of speciation and extinction and hence spatial patterns of diversity. Using data from geckos and scincid lizards across the diverse and heterogeneous savanna-like Top End region of Australia, we explore how dispersal rates over medium to long term shape patterns of species beta diversity. We apply phylogeographic and landscape genetic methods to mtDNA and extensive SNP datasets to contrast dispersal rates across multiple rock specialists and generalist species of geckos and skinks to test the predictions that specialist species are more dispersal limited than the more widespread, habitat generalist species, and that indices of dispersal limitation predict beta diversity of species. We first identified major populations within widespread species, applying bGMYC to geographically extensive mtDNA data. We then generated SNP (DArT) data for geographically dispersed individuals within each major lineage for analyses of Isolation By Distance (IBD) and to test for spatial expansion. We also verified the degree of admixture in individuals. Our results showed that, for widespread species, geckos have stronger phylogeographic structure when compared to skinks. This is possibly a result of low dispersal and high local persistence in geckos. For widespread species of geckos, all species have deep phylogeographic structure. Nonetheless, habitat specialization seems to influence the population structure of skinks and geckos, where specialist species in both groups presents more structure compared to less structure and more evidence of range expansion in generalists. Overall, we suggest that intermediate dispersal rates are important to maintaining meta-population viability while allowing divergence and hence endemism. In turn, this promotes geographic structuring of species diversity and high beta diversity in this diverse tropical biome.

GSA2018_APCC63522

Comparison of age-related sex differences and telomere length of Siamese Cobra (Naja kaouthia) with remarkable amplification of telomeric repeats on the W sex chromosome

GSA: Evolutionary genetics and comparative genomics

GSA2018 Abstract - Oral

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Telomeres comprise tandem repeats of non-coding DNA that protect the ends of chromosomes from degradation during cell division. Telomeres have been considered in senescence and mortality because they tend to shorten with growth, sex, and age. Here, age-specific telomere length in female and male Siamese cobra (Naja kaouthia) was investigated for possible relation to morphological condition. Measurements of relative telomere length (RTL) were performed on erythrocytes of 80 cobras (29 females and 51 males, age range 3 weeks – 11 years) using quantitative real-time polymerase chain reaction (qPCR). Total length (TTL) and snout-vent length (SVL) of cobras were also examined. Results showed association between age and SVL (female, R2=0.30 and male, R2=0.31), and between age and TTL (R2= 0.28 for both female and male). Two simple regression models between age and RTL were then constructed in accordance with two age groups (A: ≤ 4 years and B: > 4 years). Interestingly, in Group A, increase of telomere length was observed in females (p .05, R2=0.91) but not in males (p=0.62, R2=0.03). By contrast, in Group B, telomere length decreased with age in both sexes (female, R2=0.38 and male, R2=0.21) suggesting that increasing age above 4 years resulted in cobra telomere shortening. No evidence for quadratic or higher-order equations was found for the relationship between RTL and SVL or TTL. Observation with chromosome mapping of telomeric repeat (TTAGGG)n, indicated that the female W chromosome showed degeneration and remarkable (TTAGGG)n amplification, although interstitial telomeric sites were found on autosomes in either sex. This might relate to the increase of telomere length in females. Telomere dynamics may affect the aging process differently in females and males given their divergent reproductive strategies. Further research is required regarding sex chromosome constitution, in particular, the degenerate W chromosome and links to female fitness in snakes

GSA2018_APCC675144

Connecting speciation with macroevolution using phylogenomics

GSA: Evolutionary genetics and comparative genomics

GSA2018 Abstract - Oral

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With ready access to sequence data from 1000's of loci, we can have the potential to obtain a much higher resolution understanding of speciation processes, and how different speciation dynamics translate to macroevolutionary outcomes. We have developed exon-capture systems that yield genome-scale sequence data, with dense sampling of intraspecific (phylogeographic) lineages through to large clades of species. This approach, combined with phenotypic analyses reveals a high incidence of truly cryptic diversification, which can span millions of years as envisaged under the "Protracted Speciation Model" (PSM). As envisaged by the PSM, we also observe evidence of lineage fusion and/or introgression leading to strong cytonuclear discordance. These observations have several implications for macroevolutionary analyses. First, sampling independently evolving lineages below and above the level of taxonomically recognized species can provide richer insights into diversification processes. Second, with such dense sampling of closely related lineages, care must be taken to use appropriate (ie. Multi Species Coalescent) methods to properly estimate the branch lengths on which many macroevolutionary inferences are based. Third, evidence for often extended speciation processes and/or reticulation should challenge use of macroevolutionary models that assume instantaneous and irreversible speciation

GSA2018_APCC628178

Conserved sex-linked markers in a skink with population divergence in sex determination

GSA: Ecological genetics

GSA2018 Abstract - Oral

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Abstract: Sex determination systems are exceptionally diverse and have undergone multiple and independent evolutionary transitions among species, particularly reptiles. However, the mechanisms underlying these transitions have not been established. Here, we tested for differences in sex-linked markers in the only known reptile that is polymorphic for sex determination system, the spotted snow skink, Niveoscincus ocellatus, to quantify the genomic differences that have accompanied this transition. In a highland population, sex is determined genetically, whereas in a lowland population, offspring sex ratio is influenced by temperature. We found a similar number of sex-linked loci in each population, including shared loci, with genotypes consistent with male heterogamety (XY).However, population-specific linkage disequilibrium suggests greater differentiation of sex chromosomes in the highland population. Our results suggest that transitions between sex determination systems can be facilitated by subtle genetic differences.

GSA2018_APCC616109

Continental aridification correlates with the parallel acquisition of burrowing behaviour in Australian giant cockroaches (Blaberidae: Geoscapheinae)

GSA: Evolutionary genetics and comparative genomics

GSA2018 Abstract - Oral

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The ways in which abiotic factors contribute to parallel evolution – the evolution of similar, derived phenotypes in independent, closely related lineages – remain understudied. Australian blaberid cockroaches of the subfamilies Panesthiinae (‘wood feeders’) and Geoscapheinae (‘soil burrowers’) are two closely related groups that provide a striking example of parallel evolution of burrowing behaviour. The ancestral wood-feeding panesthiines migrated from Asia ~20 million years ago before soil burrowing was independently acquired multiple times in the derived geoscapheines. We investigated whether specific abiotic factors were associated with the parallel evolution of soil burrowing behaviour, and whether divergence events of geoscapheines from panesthiine ancestors are consistent with niche conservatism or diversification. These analyses were paired with statistical tests exploring the extent of morphological convergence in the Geoscapheinae on forelegs adapted for burrowing. We then generated environmental niche models for members of both subfamilies using presence-only data and abiotic variables related to temperature, precipitation, and soil composition.An existing phylogenetic framework was used to reconstruct ancestral niches and compare environmental niche models, and tests for niche overlap and phylogenetic signal were also performed. We found that soil burrowing behaviour is consistently correlated with thirteen abiotic factors associated with aridity, including a wider range of temperatures and lower precipitation levels. Evidence for convergence in leg morphology and body dimensions across soil burrowers was found. Overall, we find that the onset of aridity in Australia – and therefore the shrinking of available mesic habitats – likely contributed to the repeated and independent acquisition of soil burrowing behaviour in geoscapheine cockroaches.

GSA2018_APCC61988

Contribution of micro- and nano-technology to chromosome science

APCC6: New and emerging
chromosome technologies

APCC6 Abstract - Oral

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Contribution of micro- and nano-technology to chromosome science.Kiichi FukuiGraduate School of Pharmaceutical Sciences, Osaka University, Suita, Osaka, Japan Chromosome is discovered in the middle of the 19th century and has intensively been studied both in structural and functional aspects through these three centuries. One prominent fact revealed so far is that the basic structure of the chromosomes is common though the higher organisms, such as plants and animals, although they took quite different evolutional pathways. It has been shown that the chromosome has a very elastic physical property like a chewing gum and a very versatile nature for rearrangements as revealed in cancer cells. Also it has already been shown that there are two important factors on chromosome structure, chromosome scaffold and chromatin fiber, although how they interact is not known. To give the reasons to these points, we need to know the structure of the chromosome. Micro- and nano-technology are now effective tools for chromosome research by their recent rapid development. They can be used both for functional and structural studies of chromosomes, although their direct advantages have been shown mainly in structural aspects. In this presentation, utilization of the three most promising micro- and nano-technologies: super-resolution microscopy (in particular, three-dimensional structured illumination microscopy, 3D-SIM), focused ion beam/scanning electron microscopy (FIB/SEM), and transmission electron microscope tomography (TEM tomography) will be introduced giving the results so far obtained by out group. 3D-SIM and TEM tomography revealed double stranded nature of the chromosome scaffold. FIB/SEM depicted the size of the basic chromatin fibers within a human chromosome. Further applications of micro- and nano-technologies in the fields of chromosome science will be discussed.

GSA2018_APCC62918

Counting with DNA in environmental metabarcoding studies: are sequence counts useful?

GSA: Ecological genetics

GSA2018 Abstract - Oral

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Many biodiversity studies make use of metabarcoding, an approach which combines high-throughput sequencing (HTS) with DNA barcoding to characterise organisms in complex mixtures (e.g. in eDNA or bulk samples). The approach is also used to study animal diet by analysing food DNA in faecal samples. One features of HTS is that it provides counts of DNA sequences and therefore has the potential to provide not only a qualitative list, but also a quantitative assessment of what DNA is present. But what do these counts mean and do they reflect biomass of metazoans in a sample? Is it reasonable to use read proportions to retrieve semi-quantitative information, or should we work strictly with presence/absence datasets? Here, we explore how sequence counts are used in metabarcoding studies and discuss results from our research on animal diet (seals and seabirds) and zooplankton communities. We point out that summaries based on frequency of occurrence data have their own biases and argue that in some situations the quantitative interpretation of count data can be justified. We also outline our use of correction factors to account for taxa-specific recovery biases and the inclusion of internal standards to enable quantitative comparisons between samples.

GSA2018_APCC62651

Cross species gene mapping identify evolutionary conserved synteny of proto-squamate sex chromosomes

APCC6: Comparative, evolutionary and population cytogenetics

APCC6 Abstract - Oral

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Agamid lizards are squamate reptiles which show diverse sex determination systems. As the pivotal agamid model, the bearded dragon (Pogona vitticeps, PVI) exhibits ZZ/ZW micro-sex chromosomes, whereas the sex chromosome system and sex linked genes of several agamids remain unknown. To investigate bearded dragon sex chromosome homology, cross-species BAC-FISH mapping was performed in Calotes versicolor (CVE) and Amphibolurus muricatus (AMU) using three BAC clones containing genes from PVI ZW sex chromosomes. Results showed that two BAC clones 3L7 and 150H19 were localized to regions adjacent to NOR of CVE2q and one pair of CVE microchromosomes, respectively, while all BACs were mapped to the same microchromosomes of AMU. Notably, 3L7 was also located on the region adjacent to NOR AMU2q. This suggests that the presence of microchromosomes in CVE and AMU resulted from chromosomal fission adjacent to NOR near the telomeric site of chromosome 2, and subsequent fusion with a proto microchromosome. These rearrangements collectively support our previous reports on mapping in other dragon lizards that the bearded dragon ZW pair shared common ancestry with squamate reptile chromosome 2, which now shows partial homology with the chicken Z chromosome. This supports our previous hypothesis of existence of an ancestral super-sex chromosome in amniotes.

GSA2018_APCC612131

Cytogenetic analysis of Australian Grassland Earless Dragon Tympanocryptis pinguicolla Mitchell, 1948

APCC6: Specialised chromosomes (sex chromosomes and B chromosomes)

APCC6 Abstract - Oral

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Agamid lizards (Squamata: Agamidae) are karytoptically heterogeneous. This family has 480 species under six subfamilies and are distributed in Asia (including Indo-Australian Archipelago), Australasia and Africa. In Australia, there are 89 species within 14 genera from subfamily Amphibolurinae are currently described. This group is also well known for having diverse mode of sex determination mechanism including genotypic (GSD), Temperature dependent sex determination (TSD) and GSD with temperature overrides. These lizards, particularly, the Australian species have shown evolutionary lability in sex determination mechanisms with a number of recent transitions within GSDs and between GSD and TSDs. Approximately, one quarter of the agamid species have been karyotyped and heteromorphic sex chromosomes have only been identified in 9 species. Most dragons have microchromosomes as sex chromosomes, which are crytptic, while only one species reported to have macro chromosomes as sex chromosomes and all species have female heterogamety (ZZ/ZW). Grassland Earless Dragon Tympanocryptis pinguicolla is an Australian Agamid lizard that is a grassland specialist species and considered as an Endangered species in Australia. Phylogenetically and cytologically, it is a sister species of well-characterized Pogona vitticeps. The species has been karyotyped but sex determination mechanism and sex chromosome are still unknown. Here, we applied advanced molecular cytgenetics techniques and cross species chromosome painting to identify sex chromosomes in this species. Preliminary data suggests that this species possess a female heterogamety with a pair of micro chromosome

being their sex chromosomes as in P. vitticeps with certain degrees of homology.

GSA2018_APCC64370

Cytological Effect of Histone Modification by Iron Stress in Rice

APCC6: Chromosome architecture,
epigenetics and cell division

APCC6 Abstract - Poster

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Cytological Effect of Histone Modification by Iron Stress in Rice Aqwin Polosoro1,2, Wening Enggarini2 and Nobuko Ohmido*1 1Graduate School of Human Development and Environment, Kobe University, Japan2 Indonesian Center for Agricultural Biotechnology and Genetic Resources Research and Development, IAARD, Indonesia. *Corresponding author email address: ohmido@kobe-u.ac.jp Iron is one of the vital elements in almost all biochemical and cellular process in animal and especially in the plant. However, high iron content in the field is harmful on almost plant organism and development due to free iron catalyzes the formation of reactive oxygen species (ROS) through the Fenton reaction, generating hydroxyl radicals which lead to oxidative stress and causing a severe effect on lipids, proteins, and DNAs. As a result, plant response will be followed by activation and inactivation of the related genes, which is controlled by histone modification at the chromosomal level. This research was conducted to examine rice histone modification of dimethyl-histone H3(Lys9) (H3K9Me2) which is related to heterochromatin under the acute iron condition. The immunostaining experiment was performed toward whole root tissue of several rice varieties which are tolerant indica, moderate japonica, and sensitive indica. All varieties showed the similar histone modification pattern during the treatment. The immunostaining under iron stress condition confirmed the enrichment of H3K9Me2 in the whole region of the root, especially in the meristematic region which might contribute to the delay of root growth. Keywords: H3K9Me2, rice, iron toxicity stress, histone modification, retrotransposons

GSA2018_APCC672141

DArTseq genotyping for bacterial identification: testing of complexity reduced methods

GSA: Other

GSA2018 Abstract - Oral

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A study is being conducted to test DArTseq genotyping technology for use in bacterial identification and strain typing. This technology involves sequencing of complexity reduced genomes derived by digestion with pairs of restriction enzymes and subsequent PCR amplification. The amplified restriction fragments are then sequenced. The aim of this study is to test different pairs of restriction enzymes to determine the number of sequenced fragments generated from the different combinations.A total of 33 previously identified bacterial isolates provided by the Department of Microbiology in the Canberra Hospital were genotyped using DArTseq. Restriction endonuclease pairs used were: PstI – HpaII, PstI – MseI and MseI – HpaII. Samples were sequenced to give approximately 80,000 short reads per sample and fragment lengths between 40 to 69 bp. Sequences obtained were used for BLAST against the NCBI nt database collection. A set of bioinformatics tools were developed to process data and obtain the identity of the closest

matching strain from amongst the NCBI nt database. Once the closest matching fully sequenced genome was identified, BLAST was performed against the identified genome. Sequencing results from 33 samples combined, produced the following number of fragments for PstI-HpaII, PstI-MseI and MseI-HpaII: 5860, 5271 and 14746 tags respectively. The uniquely aligned restriction fragments for Enterococcus faecium yielded 374, 660 and 1888 fragments respectively; similarly for Staphylococcus aureus yields were 108, 527 and 1781 fragments respectively.Methods gave a different number of sequenced tags for BLAST alignment, even at the same total sequencing depth; however, they resulted in the same identification results at species level and sometimes differed at the strain level. The method which yielded the largest number of fragments may potentially offer finer resolution for identification purposes as well as further information which may be applicable to strain typing due to the higher genome coverage obtained.

GSA2018_APCC66015

De novo transcriptome sequencing and gene expression profiling with/without B-chromosome plants of Lilium amabile

APCC6: Specialised chromosomes (sex chromosomes and B chromosomes)

APCC6 Abstract - Oral

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B-chromosomes are small piece of chromosomes that are present in excess of normal or requisite chromosomal complements. Several species in the genus Lilium are known to carry B chromosomes. Lilium amabile, a native Lilium species in Korea, showed complex chromosome complements including aneuploids and B chromosomes in their somatic cells. B-chromosome are often preferentially inherited and derived in many ways at pre-meiotic, meiotic, or post-meiotic divisions, but the molecular mechanisms were rarely available. We carried out a transcriptome sequencing and de novo assembly of L. amabile in plant being normal diploids (2n=24) and having B-chromosome plants (2n=24+1B). A total of 77,087 genes were assembled and 59,339 were expressed in both samples. The differentially expressed genes of B-chromosome plants versus control diploids (FDR 0.05) was 8,048, of which 4,157 were up-regulated and 3,891 were down-regulated in B-chromosome containing plants. Gene ontology and pathway enrichment analysis indicated that a majority of genes have similar functions. Comparative transcriptome analsis found 2,484 uniquely expressed genes in B-chromosome containing plants. Our transcriptome results enhance our understanding of the transcriptional regulations of L. amabile in response to having B-chromosome and provide a valuable resource for the future investigation.

GSA2018_APCC673176

Defining the Molecular Targets of Evolution

GSA: Evolutionary genetics and comparative genomics

GSA2018 Abstract - Oral

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With significant advances in next-generation sequencing technologies we now have the genomes of hundreds vertebrate species but understanding how the differences and similarities within these genomes control species diversity is largely unknown. The Tasmanian tiger or thylacine (Thylacinus cynocephalus) was the largest carnivorous Australian marsupial to exist into the modern era. Their phenotypic resemblance to the eutherian canids is considered the most striking example of convergent evolution in mammals. This convergence is even more astounding when you consider that they last shared a common ancestor over 160 million years ago. Sadly, the last known thylacine died in captivity in 1936. We sequenced the thylacine genome from a preserved pouch young specimen and used it to examine the genetic basis of its convergence with canids. We performed comparative genomic analyses and demonstrated that in spite of their extraordinary phenotypic convergence, the genes and pathways targeted by positive selection differ markedly between the thylacine and canines. Despite not seeing any enrichment in protein coding homoplasy between the canids and the thylacine, some interesting genes were identified. We have extended these analyses to look at convergence within the noncoding portion of the genome and can identify convergent evolution in regions consistent with driving the craniofacial convergence observed between these species. Convergent evolution coupled with comparative genomics provides a powerful tool to examine how evolution works at the DNA level.

GSA2018_APCC635167

Disentangling chromosomal speciation in rock-wallabies

APCC6: Cytogenetics in the genomics era (Plants and Animals)

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Chromosomal rearrangements are known to generate divergence between populations and play a role in speciation. Our understanding of the effects of such structural variation in genic divergence is still in its infancy. Here we combine cytogenetic and genomic approaches to explore the effects of chromosomal rearrangements on the evolutionary history of a chromosomally diverse Australian marsupial genus Petrogale. In particular, we focus on five species from Queensland which diverged in the Pleistocene and show evidence of introgression across the genome. Using genomic data, we examine how this structural variation relates to genic divergence and aim to identify regions of the genome that are divergent and which could be responsible for reproductive isolation. We apply a targeted exon capture approach, SNP data and whole genome sequencing, together with knowledge from chromosome mapping to explore the processes driving diversification in this group. Combining cytogenetic and genomic approaches is essential to understanding the mechanisms driving speciation in this chromosomally diverse system. Our results will shed light on the role of structural variation in driving divergence.

GSA2018_APCC6993

Dissecting the chromosomal composition of mutagen-induced micronuclei in Brachypodium distachyon using multicolour FISH

GSA: Other

GSA2018 Abstract - Oral

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Numerous chemical and physical agents can affect the structure of DNA and cause double-strand breaks, which may result in chromosome aberrations and the formation of micronuclei, which eventually leads to DNA loss. Nowadays, many studies on their genotoxicity are based on the scoring of micronuclei in plants. The application of a micronucleus test combined with fluorescence in situ hybridisation (FISH) permits the involvement of specific chromosomes or chromosome fragments in micronuclei formation to be determined. However, due to the dearth of FISH probes that can specifically target individual chromosomes in plants, until now the distribution of chromosome aberrations was usually based on using repetitive DNA such as centromeric, telomeric and rDNA sequences. Chromosome painting (CP), which permits the selective visualisation of entire chromosomes or their specific segments during both cell division and interphase, is one of the most advanced FISH-based approaches. In plants, CP is limited to several small-genome species, including the model grass Brachypodium distachyon (Brachypodium). Here, we used multicolour FISH and CP to characterise the composition and to better understand the origin of the micronuclei that are induced in Brachypodium root-tip meristematic cells, which are caused by the application of chemical (maleic hydrazide) and physical (X-ray) mutagens. We show that this approach permits effective qualitative and quantitative analyses of the micronuclei and we attempted to use it to detect possible ‘hot spots’ for DNA breaks. We demonstrate that CP provides a significant improvement in the sensitivity of the ‘standard’ micronucleus test, which is commonly used in testing genotoxicity using plant cells. It appears that the application of mutagenic treatments to Brachypodium combined with its well-developed cytomolecular resources makes it a very promising, if not unique, model system for studying mutagenesis among all monocotyledonous plants. This study was supported by the National Science Centre Poland (grant no. DEC-2012-04-A-NZ3-00572, DEC-2015-18-M-NZ2-00394).

GSA2018_APCC63861

Divergence of PBI-DdeI centromeric satellite DNA between henophidian and caenophidian snakes with rapid independent amplification and evolution following the library model

APCC6: Comparative, evolutionary and population cytogenetics

APCC6 Abstract - Oral

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PBI-DdeI centromeric satellite DNA (stDNA) was found in Burmese Python (Python bivittatus) and conserved in diverse Python species with different chromosomal location and copy number. Filter hybridization and molecular cloning were performed on 40 snake species (representing 10 families) to extensively investigate levels of conservation and organization of PBI-DdeI in snake lineage. An intense positive hybridization signal was only observed in P. bivittatus. PCR detection with specific PBI-DdeI primers resulted in a ladder-like pattern of DNA bands typical of stDNAs in 15 snake species. This pattern was based on repetition of the 186–209 bp monomer unit and 98 new monomer units sequences were obtained. Phylogenetic analysis showed three PBI-DdeI subfamilies, with most PBI-DdeI sequences grouped in their respective species, except for Ahaetulla prasina. PBI-DdeI sequences of henophidian and caenophidian snakes were positionally mixed in all subfamilies, suggesting that PBI-DdeI centromeric satellite DNA was acquired in the ancestral snake genome. The sequences then evolved gradually through nucleotide substitution and homogenized to become concomitantly fixed at the species level. Measurements of stDNA copy number with quantitative real-time PCR revealed different PBI-DdeI copy numbers in each species with high copy numbers found in P. bivittatus, Ophiophagus hannah and A. prasina genomes, suggesting that diverse PBI-DdeI subfamilies coexist in the genomes of related species with rapid independent amplification among species following the library model. PBI-DdeI was localized to the centromeric region of several chromosomes in Naja kaouthia, differing in the chromosomal location of PBI-DdeI in P. bivittatus. These results collectively indicate that PBI-DdeI was dispersed in the ancestral snake genome and subsequently amplified on different chromosomes independently in each species.

GSA2018_APCC660107

Diverse or depleted? Genetic differentiation across an expanding range in invasive Australian cane toads

GSA: Ecological genetics

GSA2018 Abstract - Oral

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Invasive species typically undergo a genetic bottleneck following introduction, yet they are defined by their ability to establish and spread in novel environments. This is surprising because rapid evolution, which is thought to require high genetic variation, is likely essential for invaders to expand their ranges across heterogeneous environments. Thus, it is possible that invaders may evolve through other means, such as through heritable epigenetic changes. To explore this possibility, it is vital that we characterise genetic diversity and polymorphism in the introduced range. Cane toads underwent serial translocation from South America to Puerto Rico to Hawaii before introduction to Queensland, Australia in 1935. They have since expanded their range across several heterogeneous environments in northern Australia, and have exhibited phenotypic changes across space. Previous measurements of genetic diversity at individual loci in the cane toad suggest that it is low; however, the only previous genome-wide survey was conducted using RADSeq, which is primarily positioned in intergenic regions. We used RNA-Seq data to analyse single nucleotide polymorphisms (SNPs) from 111 toads collected across invasive ranges in Hawaii and Australia to characterise genetic diversity in these populations. We found extremely low levels of genetic diversity, likely the result of genetic bottlenecks during introduction. The ability of cane toads to change phenotypically across environments in the absence of genetic differentiation supports the possibility that epigenetic factors may underlie evolutionary changes.

GSA2018_APCC646112

DNA methylation patterns in two marsupial lineages with diverging patterns of karyotype evolution, Dasyurids and Petrogales (rock-wallabies)

APCC6: Comparative, evolutionary and population cytogenetics

APCC6 Abstract - Oral

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The two most speciose families of marsupials have very different rates of chromosomal evolution. All dasyurids (carnivorous/insectivorous marsupials) examined to date have almost identical karyotypes, though this has not prevented speciation in the family (over 70 species recognised). In contrast, macropods (wallabies and kangaroos) are typified by a high level of karyotype divergence. In particular, the genus Petrogale (rock wallabies) shows remarkable levels of chromosome rearrangement, most often involving Robertsonian fusions; throughout the 22 recognised species of rock-wallabies, only two share a similar karyotype. Our previous work had demonstrated marked differences in the DNA methylation patterns between a representative of each family, the Tasmanian devil and tammar wallaby. Devil chromosomes are characterised by hypermethylation of the terminal region of chromosomes, with low levels of methylation throughout the chromosomes. In contrast tammar wallabies have extremely hypermethylated centromeres, and moderate to high levels of methylation throughout the chromosome bodies; some chromosomes had signs of elevated methylation at terminal regions. One notable chromosomal difference between these marsupial families is an unusual telomere length dimorphism in dasyurids. Since cytosine methylation of subtelomeric regions has been shown to regulate telomere length and stability, we posited that the hypermethylation of telomere regions and the telomere length dimorphism on devil chromosomes contribute to the stability of the dasyurid karyotype. To test this hypothesis, we examined broad patterns of DNA methylation and telomeric staining in three dasyurid species, and four Petrogale species, at the chromosome level. All species tested share a common pattern of chromosome terminal region hypermethylation, with generally low levels of DNA methylation throughout the rest of the chromosome body. This pattern is in spite of chromosomal rearrangements in the Petrogale species. Hypermethylation of telomeric regions is therefore not associated with telomere length dimorphism, but is a more common feature of marsupial chromosomes.

GSA2018_APCC63438

Dramatic sterility in male hybrid catfish (Clarias gariepinus x C. macrocephalus) with failure of homologous chromosome pairing in meiosis I and different karyotypes of parental species

APCC6: Applied cytogenetics (e.g agriculture and biomedical)

APCC6 Abstract - Poster

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Sterile hybrid animals exhibit spermatogenic disruptions, with decreased number and/or malformation of mature sperms. Hybrid catfish (2n=55) are important cultured fish derived from male African catfish (Clarias gariepinus; 2n=56) and female bighead catfish (C. macrocephalus) (2n=54). They are sterile with gametogenic failure in male. Despite the generality of this phenomenon, the spermatogenic phenotype has not been well described and comprehensive understanding of the genetic basis of the disruption is still elusive. Observation of the meiotic configuration in ten male hybrids showed that meiosis succeeded in the early pachytene stage but failed to progress beyond the diplotene-diakinesis stage in primary spermatocytes, suggesting the presence of a number of degenerated spermatocytes. Histological examination recorded no postmeiotic cells and spermatozoa in hybrid testes. Many testicular cells undergoing apoptosis were present, as shown by immunohistochemistry for activated caspase 3 and TUNEL assays. These results collectively suggest that sterility in male hybrids is caused by spermatogenic disruptions during meiosis I, resulting from the failure of homologous chromosome pairing with chromosomal incompatibility between the parental genomes in addition to their diverse karyotypes.

GSA2018_APCC62596

Endogenous De Novo Centromere Formation in rice centromeric regions

APCC6: Chromosome architecture, epigenetics and cell division

APCC6 Abstract - Poster

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Centromere is essential for the faithful segregation of chromosomes in eukaryotes. Although centromere typically contains tandem repeat DNA sequences, its function doesn't rely on these DNA modifs. If canonical centromeric DNA sequences are lost, a few regions without the typical centromeric repeats constitute a de novo-formed centromere, termed neocentromere, which found in human patients, wheat-barley, oat-maize addition lines and maize. In rice (Oryza sativa), centromeric tandem repeats are known as CentO, which combine centromeric proteins, including the centromeric histone H3 (CENH3). CENH3 is the hallmarks of functional centromere identification. In this studies, we found a indica rice line Del 8 which is the total loss of the CentO sequences but the presences of CENH3 in the centromere of chromosome 8 (Cen8) region, suggesting the endogenous de novo centromere is formed. In order to make clear the location of the neocentromere and the structure characteristics of new centromeric genomic DNA, we performed CENH3-ChIP-seq and found that neocentromere is located at a-43kb-long region on the long arm of chromosome 8 in Del 8, indicating canonical histone H3 was replaced with CENH3 in this region. Combined with RNA-seq, three high transcriptional active genes and three low transcriptional genes are contained from original centromere to neocentromeric region, however, only three low transcriptional genes located in neocentromeric region. These results imply the neocentromere expansion tend to choose the region which contains no genes or inactive genes and skip the region which contains active genes. These results reveal the characteristics of neocentromeric formation, and provide a powerful support for the study of eukaryotic centromere evolution.

GSA2018_APCC655104

Environment and space drive patterns of horizontal gene transfer in soil bacteria

GSA: Evolutionary genetics and comparative genomics

GSA2018 Abstract - Poster

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Horizontal gene transfer (HGT) is increasingly recognized as a major evolutionary force driving adaptation in prokaryotes. However, the environmental factors that drive the opportunity and relative success of HGT events among natural bacterial populations have yet to be empirically evaluated among closely related strains at the landscape level. Here, we quantified HGT among closely related strains of a plant-associated bacteria, Bradyrhizobium japonicum, using whole genome data and evaluated the predictive power of two major factors frequently hypothesized to affect signals of HGT in natural populations, spatial proximity and ecological similarity. By evaluating HGT among strains sampled in a spatially explicit design across a complex environmental landscape spanning large environmental gradients (e.g. climate, soil and plant characteristics), while accounting for phylogenetic history, our analyses revealed two major results. Firstly, that HGT was more likely to occur when bacterial isolates were sampled in the same or nearby locations supporting the hypothesis that spatial proximity increases the opportunity for HGT. Secondly, that HGT was much more likely between bacterial isolates collected from different locations with similar environmental conditions, despite these locations (in some cases) being hundreds of kilometers apart. This pattern supports the prediction that HGT between dissimilar environments is less likely to be observed because there is a higher probability of receiving maladaptive genes, leading to HGT events that are not selectively maintained in the environment. The results of our analyses provide empirical evidence for the important role of HGT in bacterial adaptation, and the potential for bacteria to adapt to environmental change.

GSA2018_APCC62182

Estimating evolutionary rates and timescales using phylogenomics

GSA: Bioinformatics and genomics

GSA2018 Abstract - Oral

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Genomic data present unprecedented opportunities for resolving the branching patterns and timescale of the Tree of Life. In particular, analyses of genome-scale data sets allow us to improve and refine our knowledge of evolutionary timeframes. The timing of evolutionary events can be inferred using molecular clocks, which are now being adapted to handle the avalanche of data provided by genome projects. In this talk, I will describe our recent efforts to estimate the evolutionary timescales of flowering plants and of marsupial families. I will also describe our ongoing work on estimating rates of evolution at various biological scales across the Tree of Life.

GSA2018_APCC628116

Evolution of a terrestrial ectoparasite on a Trans-Tasman marine host: the penguin and the tick

GSA: Evolutionary genetics and
comparative genomics

GSA2018 Abstract - Oral

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Parasites are often incapable of significant active dispersal, instead relying on hosts to facilitate movement and transmission. Seabirds are highly effective at long-distance dispersal, and their fidelity to large multi-species colonies exposes them to terrestrial ectoparasites such as the tick (Acari). The common seabird tick (Ixodes uriae) is one of the most widespread species in the world, suggesting the ectoparasites are moving freely among seabird colonies via aerial dispersal. Unlike most seabirds, however, penguins disperse primarily underwater. This presents a unique challenge for penguin-associated ticks, which are terrestrial yet must deal with the effects of seawater and pressure to disperse with their hosts. Here, we used next-generation sequencing to investigate phylogeographic structure and infer connectivity in little penguin (Eudyptula spp.) ticks (Ixodes spp.) on both fine (within a colony) and broad (among colonies across its range in Australia and New Zealand) scales. Physiological experiments were also carried out to test the physical limits of little penguin ticks in relation to the conditions they would face during marine dispersal, in order to ascertain whether underwater dispersal presents an effective barrier to tick movement. Results suggest that ticks may be able to withstand seawater emersion, that the extent of penguin social behaviour dictates fine-scale parasite population structure, and that trans-Tasman movements of little penguin ticks has occurred but remains too limited to maintain ongoing gene flow. We conclude that parasitic adaptations to harsh terrestrial conditions may confer survival in seemingly impossible environments.

GSA2018_APCC61087

Evolution of rDNA Mappings and ITS Phylogeny in Sweetpotato and its Wild Relatives

APCC6: Cytogenetics in the genomics era (Plants and Animals)

APCC6 Abstract - Poster

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The sweetpotato (Ipomoea batatas) is ranked as the seventh most important food crop in the world. However, the origin and genomic organization of sweetpotato remain unclear. Ribosomal DNA (rDNA) and internal transcribed spacer (ITS) sequences could provide useful information for better understanding the evolutionary and phylogenetic relationships within the genus. In the current study, the number and distribution of 45S rDNA and 5S rDNA were studied in eighteen accessions of series Batatas, including twelve diploids, one tetraploid I. tabascana, one hexaploid I. trifida and four I. batatas accessions. ITS sequences were used as well to construct phylogenetic trees. From the results of rDNA localization, the number and distributions of 45S rDNA varied both interspecifically and intraspecifically. The number of 45S rDNA varied from four to twelve among diploid wild species, sixteen and twenty-four signals were detected in the tetraploid I. tabascana and hexaploid I. trifida respectively, and it also varied from twenty to twenty-two among the cultivars. Only polyploids and one diploid named I. tiliacea had the interstitial 45S sites. In contrast, the number of 5S rDNA was completely consistent with ploidy level. Two, four, six sites were detected in the diploid, tetraploid and hexaploid species respectively. Phylogenetic tree based on the results of ITS sequences showed that I. grandifolia was the most distantly related to I. batatas, while the remaining thirteen species were divided into two groups. The first group included the polyploids and five diploids. The second group contained six species with a genome previously named the ‘A genome’. The data obtained in the current study indicate that hexaploid I. trifida may be a segregate of I. batatas. Of those species sharing the ‘A genome’, I. tiliacea may contribute to the genomic organization of the sweetpotato, and the ‘A genome’ may share the origin of I. batatas.

GSA2018_APCC629164

Evolutionary process of ZW sex chromosomes in Hokou gecko, Gekko hokouensis

APCC6: Specialised chromosomes (sex chromosomes and B chromosomes)

APCC6 Abstract - Poster

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Sex-determining systems are variable in reptiles. Birds and snakes have genetic sex determination (GSD) whereas all crocodilians and tuataras exhibit temperature-dependent sex determination (TSD). In turtles and lizards, both GSD (XY and ZW) and TSD species are recognized. Thus transitions between sex determination systems have occurred in many lineages of turtles and squamates and sex chromosomes have also arisen multiple times together with those transition. Gekkota is one of clades where high variation of sex-determining systems is observed. The sex chromosome of Hokou gecko (Gekko hokouensis) is of particular interest because the ZW pair is homologous with the avian ZW system (Kawai et al. 2009). Gekko japonicas, closely related for G. hokouensis, and a cryptic species of G. hokouensis exhibit TSD. These results suggest an idea that the sex chromosomes of G. hokouensis and Aves were independently derived from the same autosome by convergent evolution. In our previous study, by FISH mapping of cDNA clones, a pericentric inversion was identified between the acrocentric Z chromosome and the subtelocentric W chromosome and DMRT1, a sex determination gene in vertebrates, was located to the region where the inversion occurred. Here we compared sequences between the Z and W homologs of DMRT1. As the result, a SNP was identified in the coding sequence. We also sequenced the Z and W sequences, and searched differentiated sequence between Z and W homologs, and W-specific sequences to investigate evolutionary process of the G. hokouensis sex chromosome.

GSA2018_APCC655177

Evolutionary transitions in the early bilaterian karyotype

APCC6: Comparative, evolutionary and population cytogenetics

APCC6 Abstract - Oral

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Comparisons of diverse animal genomes have shown that a network of gene linkages is deeply conserved, albeit with considerable changes in gene order, a property referred to as conserved synteny or macro-synteny. Using data from several ongoing studies across metazoans, I will present the impact of chromosomal-scale assemblies on our evolutionary inferences of the invertebrate chromosomal complement. In particular, we use these deeply conserved syntenies to infer the evolutionary dynamics of proto-vertebrate genomes by comparing extant vertebrate sequences with a chromosome-scale assembly of amphioxus, an invertebrate chordate whose lineage diverged prior to the emergence of vertebrates. We find large-scale correspondence of the extant amphioxus chromosomes to the inferred ancestral linkage groups. We can resolve two successive genome-wide duplications in the lineage leading to jawed vertebrates, separated by a period of extensive chromosome fusions and rearrangement. We furthermore identify asymmetric retention of genes across vertebrate karyotypes, indicative of allotetraploidy, or chromosome doubling after hybridization. Finally, I will compare this evolutionary history to other chromosomal-scale dynamics observed in invertebrate lineages, in particular cephalopods.

GSA2018_APCC6909

Examining rapid evolution of insecticide resistance using exon capture on historical and contemporary pest moth samples

GSA: Ecological genetics

GSA2018 Abstract - Oral

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The cotton bollworm (Helicoverpa armigera) is a major pest of cotton and other agricultural crops, costing billions of dollars in management and yield losses globally. A key aspect of its success as a 'megapest' is the moths' ability to rapidly evolve insecticide resistance. My research uses DNA from Australian moth samples collected > 100 years ago, comparing genomic sequences from these historical insecticide-free pest genomes to contemporary populations of H. armigera that are currently exposed to insecticides. Examining the genetic make-up of a rapid adaptive response to an environmental change provides a template for testing questions about the presence or absence of pre-adapted resistance genes and for understanding the mechanisms behind successful pest status. In this talk, I will describe my experimental system and discuss some of the preliminary results from my research, including a focus on library quality/metrics and bioinformatic considerations.

GSA2018_APCC68955

Existence of BovB non-LTR LINE retrotransposons in snake lineages suggests potential for horizontal gene transfer and sex bias in copy number.

APCC6: Comparative, evolutionary and population cytogenetics

APCC6 Abstract - Oral

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Transposons are present as dynamic elements in all eukaryotic genomes. Their ability to ‘jump’ around the genome and amplify offers potential for significant genomic change resulting in segment duplication, chromosomal rearrangement and sex chromosome differentiation. Long interspersed nuclear elements (LINEs) accumulate in snakes on Z or W sex chromosomes in several lineages. Partial sequences of the reverse transcriptase gene of BovB LINE non-LTR retrotransposon were sequenced and characterized from males and females of 24 snake species to better understand LINE dynamics and organization in snake genomes. Phylogenetic analysis showed four BovB LINE subfamilies with most BovB element sequences grouped among respective species. Most caenophidian snakes were clustered together in the same subfamily, except for Acrochordus javanicus which was grouped with boid and xenopeltid snakes. Three of four subfamilies contained dispersed henophidian snakes and other squamate reptiles as outgroups. This phylogenetic topology differed from the tree based on functional nuclear and mitochondrial gene sequences, suggesting that horizontal transfer events occurred in snake lineages. Average Ka/Ks approached 1, suggesting that after retrotransposition the BovB element was influenced by pseudogene-like evolution. Quantitative real-time polymerase chain reaction (qPCR) was applied to measure BovB element copy number. Variation was demonstrated between sexes with males showing higher copy number than females in eight species (Cylindrophis ruffus, Python bivittatus, Daboia siamensis, Homolopsis buccata, Naja kaouthia, Naja siamensis, Ahaetulla prasina and Ptyas mucosa), whereas females proved higher than males in two species (Python regius, Coelognathus radiata). This suggests that sex-biased copy number of the BovB element occurred independently in snake lineages, possibly by preferred accumulation of transposons on Z or W sex chromosomes.

GSA2018_APCC62193

FISH Characterization of Ginseng Chromosomes Using Repeat Probes Identified from Whole Genome NGS Sequences

APCC6: Cytogenetics in the genomics era (Plants and Animals)

APCC6 Abstract - Oral

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Ginseng, Panax ginseng C. A. Meyer, has been used as a king medicinal herb in east Asia and has a large genome of more than 3.5 Gbp in 24 haploid chromosomes. Its genome structure, constitution, and phylogenetic history have just gained pace in recent years. Molecular cytogenetic research was carried out in ginseng using next generation sequencing (NGS) whole genome sequence (WGS) reads. Refined fluorescence in situ hybridization (FISH) karyotype was established with repeat probes including several long terminal repeat retrotransposon (LTR-RT, PgDel2, PgDel3, PgTat1, PgTat2, PgTork) and one tandem repeat (Pg167TR) probes. FISH results showed differential accumulation of Ty3/gypsy LTR-RT into subgenomes, suggesting a non-random preferential amplification of retrotransposons and an allotetraploid origin of P. ginseng. Pg167TR was the most abundant TR and showed unique distribution pattern among the 24 chromosomes which allowed to discriminate and characterize each individual chromosome. An extensive Pg167TR repeat accumulation outside functional PgCACTA transposon seemed to indicate non-functionalization of ancient PgCACTA element and a subsequent tandem repeat amplification. Sequence comparison categorized the units into 2 subclasses, PgTRa and PgTRb. Genomic distribution of PgTRb revealed hybridization to only 12 out of 24 chromosome pairs and these support an allotetraploid origin of the ginseng genome. High-throughput and more efficient genome-wide repeat mining was accomplished with low-coverage WGS reads using RepeatExplorer algorithm. The telomeric and two chromosome-specific repeats in ginseng genome were identified and pre-labelled oligonucleotide probes (PLOP) were developed for FISH analysis. The FISH procedure using PLOP was rapid and efficient and the probes were verified as excellent molecular cytogenetic markers to identify each chromosome. These results will be useful for breeding, genetic and physical map integration, sequence assembly validation, and comparative cytogenetic studies to unravel ginseng genomic history. This work was supported by the Cooperative Research Program for Agriculture Science & Technology Development (PJ013119), Rural Development Administration, Korea.

GSA2018_APCC695132

FISH mapping of rDNA and telomeric repeats in 12 Senna species

APCC6: Comparative, evolutionary and population cytogenetics

APCC6 Abstract - Poster

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Several species in the genus Senna under the family Fabaceae, had been intensively studied owing to its industrial and medicinal potential. However, there is limited chromosomal information essential for cytogenetics and genomics research in the genus. Fluorescence in situ hybridization (FISH) allows visualization of DNA sequences on the chromosomes. Here, we performed (FISH) to analyse comparative karyotypes according to the distribution of 5S and 45S rDNAs, as well as Arabidopsis-type telomeric repeat (TTTAGGG)n on mitotic metaphase chromosomes in 12

Senna species. All species showed the chromosome number of 2n = 28 except for S. tora with 2n = 26. Only one pair of 5S rDNA was detected in all species, while the number of 45S rDNA signal varied according to the species; four pairs in S. didymobotrya, S. spectabilis, and S. viarum, three pairs in S. covesii, two pairs in S. candolleana, and one pair in S. bauhinioides, S. floribunda, S. multiglandulosa, S. occidentalis and S. tora. A hemizygous distribution of 45S rDNA was observed in S. leandrii and S. sulfurea. Telomeric signals were observed on the terminal region of all the chromosomes. In S. tora, an intense and thick telomeric signals appeared on interstitial regions of all chromosomes. This phenomenon was also observed in S. occidentalis but in fewer number of chromosomes and of weaker signals. Interspecific FISH karyotype variation among the 12 Senna species reveals the genome dynamics in Senna. These preliminary karyotype data will be useful for further cytogenetic mapping of other major repeats to understand the genome structure and history of the genus Senna.

GSA2018_APCC67079

FISH-Aimed Chromosome Analysis of HERV Element and Its Genomic Features in Primates

APCC6: Comparative, evolutionary and population cytogenetics

APCC6 Abstract - Oral

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The human endogenous retroviruses (HERVs) have been subjected to many amplification and transposition events resulting in a widespread distribution of complete or partial retroviral sequences throughout the primate genome. Most HERV families have been inserted into the primate genome and subjected to amplification on several occasions between the divergence of hominoids and Old World monkeys 30-45 million years ago. Compared to another HERV families (HERV-I, HERV-R, and HERV-W), the HERV-K family including solitary LTR elements represents abundant copy numbers in humans and great apes. Using UCSC genome browser and Repbase, the copy number of HERV-W family are calculated, indicating that 153 HERV-W elements and 209 solitary sequences are detected. They are distributed in whole chromosomes except for chromosomes 16 and 22, which are consistent with the data of Southern blot and PCR analyses. They can influence gene transcription and biological function through various mechanisms. Long terminal repeats (LTRs) of human endogenous retroviruses (HERVs) have been shown to influence the expression of neighboring genes. HERV insertion event during primate evolution could be genetic marker for the study of phylogeny and evolution. Phylogenetic analysis of HERV elements provides important information regarding the evolutionary history of the retroelements and various events on the primate genome. Accumulated changes of the HERV elements in gene regulation are likely to be functional factors for the process of diversification, speciation and evolution consequences.

GSA2018_APCC698137

Formation of micronucleus and aborted pollen in xBrassicoraphanus, an intergeneric allotetraploid between Brassica rapa and Raphanus sativus

APCC6: Applied cytogenetics (e.g agriculture and biomedical)

APCC6 Abstract - Oral

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Interspecific or intergeneric hybridization can give rise to genetic diversity, but the resulting hybrids often suffer from diverse genetic and developmental defects. Notable among them are chromosomal instability and infertility, mainly caused by genome incompatibility between distantly related genomes. xBrassicoraphanus is a synthetic allotetraploid (2n = 4x = 38) derived from an intergeneric cross between Chinese cabbage (Brassica rapa; 2n = 20) and radish (Raphanus sativus; 2n = 18). Four cultivars of xBrassicoraphanus BB1, BB4, BB6, and B50 were all descended from a genetically fixed individual, but display varying degrees of phenotypic diversity and pollen viability. For instance, the BB1 cultivar is the most stable in terms of phenotypic and genetic uniformity, whereas BB4 is relative unstable with a high frequency of inviable pollen formation. Microscopic analysis revealed that more than half of BB4 pollens aborted with irregular morphology, while producing significantly large numbers of polyads during microsporogenesis, approximately 6.98% of which contained micronuclei. We also observed mispairing and abnormal segregation of meiotic chromosomes during microsporogenesis in BB4. Our findings suggest that chromosome instability is persistent in the genome of hybrid offspring, accompanied with the formation of micronuclei and aborted pollen. Thus, the divergent genomes in the hybrid species still require a comprehensive stabilization process after hybridization, presumably involving gradual genome reconstruction and/or epigenetic changes to produce stable and fertile individuals over successive generations.

GSA2018_APCC679170

From Genomes to Telomeres

APCC6: Chromosome architecture,
epigenetics and cell division

APCC6 Abstract - Oral

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Telomeres are nucleoprotein structures that function to cap the ends of linear chromosomes, preventing them from being recognised as DNA double strand breaks. In normal somatic cells, telomeres shorten with each round of cell division, eventually resulting in cell cycle arrest. The replicative immortality of human cancer cells is achieved by activation of a telomere maintenance mechanism (TMM) to extend telomeric DNA and counteract telomere attrition. To achieve this, cancer cells utilise either the enzyme telomerase, or the Alternative Lengthening of Telomeres (ALT) pathway. These distinct molecular pathways are incompletely understood with respect to activation and propagation, as well as their associations with clinical outcomes. We have identified significant differences in the telomere repeat composition of tumours that use ALT compared to tumours that do not. We have used these differences to stratify the TMM of tumours using telomere reads extracted from whole genome sequencing datasets. This has enabled us to investigate the genetic signatures associated with telomerase and ALT activation in cancers. Importantly, this classification approach is applicable across all tumour types. Analysis of pathway mutations that were under-represented in ALT tumours, across 1,075 tumour samples, revealed that the autophagy, cell cycle control of chromosomal replication, and transcriptional regulatory network in embryonic stem cells pathways are involved in the survival of ALT tumours. We are currently conducting functional studies to determine how these pathways contribute to ALT activation and propagation. Overall, our approach demonstrates that telomere sequence content can be used to stratify TMM in cancers, and has identified novel molecular mechanisms involved in these pathways.

GSA2018_APCC63960

From Servant to Master: the Ribosome’s instructive role in hematopoietic cell fate determination

GSA: Development and cellular genetics

GSA2018 Abstract - Oral

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Transcriptional control of gene expression is viewed as the key instructor of stem cell fate. In contrast, dogma has relegated ribosome biogenesis and mRNA translation as a necessary, but passive, contributor to cell and tissue specification during development. Intuitively, impaired ribosome function should (and usually does) cause reduced tissue growth and stunted development. Inexplicably, cancer arises more frequently in patients with ribosomal protein (RP) loss, particularly in the blood lineage. Our recently developed Drosophila hematopoietic models may provide a rationale for the prevalence of leukemia in patients with reduced RP levels. Following tissue specific depletion of the RPs (s19 and s24) most frequently mutated in the ribosomopathy Diamond Blackfan Anemia (DBA), we observe stem cell fate defects, overproliferation and massive overgrowth of the blood compartment. Interestingly, knockdown of RPs19 and RPs24 differentially altered stem and progenitor cell differentiation, which suggests depletion of particular RPs from ribosomes might actively disrupt cell fate by modulating the classes of transcripts translated. Indeed, Mass Spec data from the RP deficient lymph glands revealed increased protein abundance for several factors previously implicated in driving developmental programs of growth and differentiation, including master transcriptional regulators and chromatin remodeling machinery. As RNA-Seq revealed mRNA transcript levels for these factors were unchanged, we hypothesise that their defective translation underlies the stem cell fate defects and blood compartment overgrowth. A combination of TEM and Bi-FC analysis revealed that RP depletion did not significantly reduce mature ribosomes in the cytoplasm, suggesting that altered protein abundance is not due to competition for ribosomes. We are currently using mass spec to determine if RPs19 or s24 depletion alters ribosomal composition in Drosophila hematopoietic cell lines, and whether mRNA species encoding stem cell fate proteins are differentially translated using RNA-seq of polysome fractions.

GSA2018_APCC63030

Genes are Information, so information theory is pervading genetics

GSA: Other

GSA2018 Abstract - Oral

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Geneticists are by now familiar with Bioinformatics, which uses massive computer power to manage our massive datasets. They are less familiar with information theory, which concerns the analysis of how the computers themselves function – ie how information can be transmitted or manipulated. Information theory's founder, Claude Shannon, based it on gas theory, but he focused on electronic information. However, Shannon recognised that the theory applies to any kind of information, and he even made an early attempt to apply it to genes - hampered by not knowing that genes were DNA. Around the same time, geneticists such as Kimura were beginning to use the same gas theory, to produce the equations that fill our genetics textbooks today, which forecast heterozygosity, numbers of allelic types, etc under a wide range of conditions. Surprisingly, Shannon's method was taken up enthusiastically not by geneticists, but in ecology, where it is used only as a raw measurement of diversity, with limited ability to make testable predictions. Two important things have now changed. First, it has been recognised that ecology and evolution share four basic processes – dispersal, random change, adaptation, and generation of novelty (speciation, mutation, and recombination). Secondly, the last decade has seen a burgeoning of uses of Shannon information for molecular diversity, including predictive equations, at all levels from sequences and expression in cells through to macroevolution. As a result, we are set to see analyses integrated at all these levels, with the bonus that information/entropy provides a general predictive approach, not just for genetics and ecology, but for other studies such as the physical environment, and may even extend to assist with evolutionary programming in AI. For further detail, see: Sherwin et al. 2017. TREE doi.org/10.1016/j.tree.2017.09.012

GSA2018_APCC688189

Genetic identification and the Southern Ocean CPR survey: a useful combination?

GSA: Ecological genetics

GSA2018 Abstract - Poster

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The Continuous Plankton Recorder (CPR) has been used to characterise zooplankton biodiversity along transects covering hundreds of thousands of kilometres in the Southern Ocean CPR survey. We investigated the potential to use DNA metabarcoding (species identification from DNA mixtures using high-throughput DNA sequencing) as a tool for rapid collection of taxonomic data from CPR samples. In our study, zooplankton were collected on CPR silks along two transects and identified using standard microscopic methods and by sequencing a mitochondrial COI marker. DNA increased the number of metazoan species identified and provided high resolution taxonomy of groups problematic in conventional surveys (e.g. larval echinoderms and hydrozoans). Metabarcoding also generally produced more detections than microscopy, but this sensitivity may make cross-contamination during sampling a problem. In some samples, the prevalence of DNA from large plankton such as krill masked the presence of smaller species. Overall, the genetic data represents a substantial shift in perspective, making direct integration into current long-term time-series challenging. We discuss a number of hurdles that exist for progressing this powerful DNA metabarcoding approach from the current snapshot studies to the requirements of a long-term monitoring program. Given the continually increasing efficiency of DNA metabarcoding, it is almost certain this approach combined with underway sampling will play an important role in characterising the status and trends of key species in future Southern Ocean monitoring programs.

GSA2018_APCC629162

Genome of Sycon capricorn, a model to investigate evolution of animal body plans

GSA: Development and cellular genetics

GSA2018 Abstract - Oral

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The stunning morphological diversity within the animal kingdom is likely a result of extensive radiation of gene regulatory networks specifying animal body plans. This diversity has long inspired researchers to investigate the evolution of animal development; yet, we know relatively little about the evolutionary origins of key animal body features such as determination of multiple cell types, axial polarity, body symmetry, and germ layer specification. Insight into the evolution of animal body plans can be gained through the study of early branching animal lineages. Sponges, often argued to be the oldest extant animal clade, bridge the gap between our most recent unicellular ancestors and the more familiar “true” multicellular animals (eumetazoans, including cnidarians and bilaterians). Intriguingly, despite their relatively simple body plans, the majority of the key eumetazoan developmental gene regulatory network components are found in sponge genomes. These include the canonical Wnt pathway, which is involved in myriad developmental processes including cell fate decision-making, establishment of cell and tissue polarity, and patterning of the main body axis among eumetazoans. I will describe a new sponge model system for the study of developmental gene regulatory networks. Sycon capricorn, a calcaronean sponge found along the eastern Australian coast, is well-suited as a model species due to its local abundance, remarkable regenerative capacity, and a growing suite of molecular data resources, including draft genome. I have used a hybrid genome assembly approach incorporating massive short-read sequencing with cutting edge MinION long read sequencing. I will present the results of genome annotation using RNAseq data, which will be used as a reference for an upcoming ChIP-Seq study in S. capricorn, aimed at identification of the canonical Wnt signalling targets in this sponge.

GSA2018_APCC697159

Genome size, chromosome number and distribution patterns of rDNAs in some Onobrychis species

APCC6: Comparative, evolutionary and population cytogenetics

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The genus Onobrychis is a member of the Fabaceae family with approximatelly 170 perennial and annual species. Some species of the genus are important fodder crops. Previous cytogenetic studies in genus Onobrychis are limited to chromosome counts and construction of karyotypes based on classical staining methods. New molecular cytogenetics techniques have not yet been applied to investigate Onobrychis genomes. The objective of this study was to explore usage of flow cytometer and in situ hybridisation (FISH) methods on Onobrychis species for the first time. The present study reports genome size of 200 Onobrychis accessions belong to approximatelly 25 different species and distribution patterns of rDNAs in some of the species. Based on the results of the study, mean 2C DNA content of the Onobrychis species varied between 2.73 pg and 0.70 pg as the differences among species were statistically significant. Species generated two clearly distinquishable groups according to mean 2C DNA content. Two

different essential chromosome number such as n=7 and n=8 were counted in the study as the majority of the accessions were polyploid with 2n=28 chromosomes. Based on the results of the FISH analysis, diploid species had distinctive distribution patterns of rDNA loci, whereas polyploids analysed in the study revealed similar rDNA organisation. Acknowledgements: This study was supported by a TUBITAK grant (TOVAG-215O526).

GSA2018_APCC61068

Genomic Evolution of Bird Sex Chromosomes

APCC6: Specialised chromosomes (sex chromosomes and B chromosomes)

APCC6 Abstract - Oral

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Birds are the most speciose and arguably the most successful terrestrial vertebrate with tremendous phenotypic diversities that facilitate their inhabitation throughout the planet. They can be divided into two clades: Paleognathae (tinamous and flightless ratites), and Neoganathae with over half of the latter, and also of all bird species as passerines. All birds have independently evolved ZW sex chromosomes, in contrast to the XY of mammals. I will present here progress from our recent genomic studies of sex chromosomes of 15 paleognaths and 11 passerines. We found almost all ratites have long pseudoautosomal regions (PAR) that span over 2/3 the entire sex chromosome length and have homologous recombination. Interestingly, a similar pattern has also been found in half of the tinamous species with a much higher mutation rate than the ratites. Due to the ancestral rapid speciation, PAR and also suppression of recombination evolved independently within individual lineages/species, presenting a great diversity of PAR lengths and W-linked gene repertoire. On the other hand, all the studied passerines have short PARs and shared stratified regions of recombination suppression between sex chromosomes. Given the extensive sexual selection in species like bird-of-paradise, it is surprising to find that majorities of studied passerines have retained substantial (>70) W-linked genes that are conserved across species. These results illuminate diversified mode of sex chromosome evolution of birds, and also the distinctive mutational and selective forces that underlie the evolution of ZW and XY systems.

GSA2018_APCC66297

Genomic tests for ‘blocked’ dispersal

GSA: Ecological genetics

GSA2018 Abstract - Oral

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Dispersal is a fundamental process affecting species’ distributions and population connectivity, yet we know that dispersal does not always (indeed, often fails to) result in colonisation and establishment. In such cases, dispersal places organisms in a position to leap on new opportunities provided by environmental change – dispersers are poised to colonise territory liberated by disturbances, or when environmental conditions improve. In this talk I will provide examples from recent research in the Antarctic and Australasia in which dispersal that has apparently failed to result in colonisation was detected using genomic analyses. Such research allows us to understand potential colonisation and invasion pathways, and thus to predict how ecosystems might respond to various types of environmental change. For example, I will present genomic evidence that Antarctica is not – as has long been believed – biologically isolated; that organisms can, and do, cross the Southern Ocean to reach Antarctica, but are presumably currently unable to establish because of the region’s environmental extremes. With global warming, however, we should expect to see successful establishment of numerous non-Antarctic species, even without human-mediated transport of organisms to the region.

GSA2018_APCC638145

Haplotype assembly using third generation sequencing

GSA: Genetics and medicine

GSA2018 Abstract - Poster

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Haplotype information is important for genetic analysis and has many applications in population genetics and biomedical research. Third generation sequencing data can provide long but error-prone reads. For example, PacBio and Minion reads are long enough to span over multiple heterozygous variants and are therefore ideal for phasing. However, the high error rate in these reads also makes SNP calling and haplotype assembly challenging. Most existing approaches for long-read based phasing still rely on SNP calling from the second generation data (e.g., Illumina reads) and do not scale up to handle long reads with relatively high coverage (e.g., above 50X). Moreover, most of these approaches also require a reference genome to infer the SNPs. We develop a pipeline that incorporates both reference and haplotype assembly. We first assemble all the long and error-prone reads into preliminary contigs where two haplotypes collapse on. We then align all raw reads to the above contigs and further infer reliable SNPs. Finally, we develop phasing algorithms to derive two haplotypes when there are enough heterozygous variants between them. Our phasing algorithm uses the divide-and-conquer idea and thus is able to scale up to handle third generation datasets with higher coverages. Experiment results also show that our phasing algorithm is also potential to reveal assembly errors in preliminary assemblies.

GSA2018_APCC62048

Historical biogeography and taxonomy of the anti-equatorial fish genus Microcanthus (Teleostei: Microcanthidae)

GSA: Evolutionary genetics and
comparative genomics

GSA2018 Abstract - Oral

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The fish genus Microcanthus consists of a single species, the stripey (Microcanthus strigatus), which has a large but disjunct anti-equatorial distribution across the Indo-West Pacific. Such patterns are seen in the distributions of several other fish taxa, whereby sister species are found to the north and south of the equator but not at the equator itself. However, the stripey is regarded as a single species despite comprising four geographically isolated populations. The relationships among these populations have not been confidently resolved. By analysing genetic data, I will estimate the degree of population structure, evolutionary timescale, and biogeographic history of the stripey. The patterns identified in these analyses will be compared with those in other fish taxa with anti-equatorial distributions. This will provide insights into the geological and oceanographic processes that have shaped the distributions of marine organisms. I am also performing the first integrative taxonomic analysis of Microcanthus strigatus using morphological characters and molecular data. These data will allow me to evaluate potential cryptic diversity within the species, opening the way for nomenclatural revisions where needed. The results of this project will potentially have implications for conservation and biodiversity management.

GSA2018_APCC642110

Homogenisation of telomere length in a transmissible cancer, devil facial tumour 2

GSA: Other

GSA2018 Abstract - Oral

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A decade after the appearance of the first devil facial tumour (DFT1), first reported in 1996, a suite of genetics work was done to understand the transmissible cancer. These revealed that though the cancer was slowly evolving and capable of changing in reaction to its 'environment', it had reached more a less a point of 'genomic stability'. In 2014, a new independent transmissible cancer was found in Tasmanian in devils, devil facial tumour 2 (DFT2). This has presented us with a chance to study the cancer in its 'early' stages, an opportunity missed with DFT1. We present work done investigating telomeric changes in DFT2. Telomeres, the ends of chromosomes, erode over time, and a lack of telomeric ends may cause massive genomic rearrangements through a breakage-fusion cycle, potentially leading to oncogenesis. There is evidence of this occurring between chromosomes 1 and X in DFT1, and likewise evidence of a chromosome fusion occurring between chromosomes 1 and 6 in DFT2. We know that Tasmanian devils, and dasyurids in general, have a telomeric length dimorphism; for each chromosome pair, the maternal chromosome has short, and the paternal long, telomeres. This dimorphism had disappeared in DFT1 by the time its telomeres were investigated, but is still present in DFT2. By looking at long term cell culture of DFT2 over 200 population doublings, we present evidence that the telomeres of DFT2 are likely to homogenise, as in DFT1. The long telomeres shorten over time, yet the short telomeres are maintained, likely by the telomerase enzyme. This indicates that telomere length is stabilising in DFT2, and is likely to end up homogeneous as in DFT1. Without telomeric erosion and instability, DFT2 is unlikely to undergo new chromosome fusions. These results give a glimpse as to how DFT1 could have lost the telomere length dimorphism.

GSA2018_APCC65820

Homogenization of satellite DNAs and chromosome size-correlated compartmentalization reveal state of diversity in squamate reptile genomes

APCC6: Comparative, evolutionary and population cytogenetics

APCC6 Abstract - Oral

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Squamate reptiles show very dynamic genome structure between lineages (macro- and microchromosomes and no microchromosome in the genome) which might be associated with their richness at around 8,000 species. One genomic region that is an indicator of the dynamic state comprises satellite DNAs (stDNAs). These are highly repeated sequences constituting large portions of the heterochromatic region of any genome. At least four stDNA subfamilies coexist in varanid genomes and mostly differ in nucleotide sequences and copy number. Comparing stDNA subfamilies and species revealed that similarities between each sequence unit within different species of the same stDNA subfamily were higher than those of other stDNA subfamilies belonging to the same species. This suggests that stDNA families lack homogenized species-specific nucleotide positions in varanid lineages. These results agreed with the library model of stDNA evolution, in which diverse stDNA families or subfamilies coexist in the genomes of related species, with disparate amplification among species. By contrast, monomers of a stDNA family exhibit higher sequence similarity than the same stDNA family of related species in snakes, suggesting that mutations in stDNA monomers are homogenized and concomitantly fixed at species level. Additionally, stDNA sequences in snakes show no genomic compartmentalization between micro- and microchromosomes as a consequence of homogenized chromosome size-correlation. Conversely, stDNA sequences are specifically located on macrochromosomes in varanids, suggesting chromosome size-correlated compartmentalization between macro- and microchromosomes. Evolutionary dynamics of stDNA (e.g. copy number, nucleotide sequence, location) can, therefore, provide insights into genome organization and evolution

GSA2018_APCC650196

Homology of sponge, cnidarian, and bilaterian body plans: was Haeckel right?

GSA: Development and cellular genetics

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The emergence of complex animal life forms is one of the major events in the history of life on Earth and it is often a topic of interest and intense disagreements. One of the biggest obstacles to understanding animal evolution is the perceived difficulty of evolving a complex human body from single-cell organisms. Ernst Haeckel, along with some other 19th century biologists believed that body plan organization of sponges (Porifera) is an intermediate step between protists and the simple multicellular animals, such as cnidarian polyps. According to this view, the innermost layers of animal bodies involved in digestion (the sponge choanoderm and the endodermally derived cnidarian gastrodermis or bilaterian gut) were homologous structures. While neglected for a century, this view recently received support from studies comparing expression of several developmental regulatory genes in sponges, cnidarians, and bilaterians. In this project, we will investigate and compare global gene expression profiles of sponge, cnidarian and bilaterian tissues, especially the choanoderm with endoderm, the pinacoderm with ectoderm, and putative sponge larval sensory cells with neuronal and sensory cells of cnidarians and bilaterians. In addition to well-established cnidarian and bilaterian models (including sea anemones and mammals), we will use an emerging sponge model, Sycon capricorn (Calcarea). To generate tissue-specific transcriptomes, we will use the recently developed single-cell sequencing technology, Drop-Seq. We will then map the obtained transcriptomes of the cell types using a combination of in-silico and laboratory approaches. Lastly, we will compare the identified cell types between the selected models to assess their homology. Testing this hypothesis will not only bring us closer to answering the question “where do we come from?”, but it will also generate extensive data that will provide a foundation and resource for understanding the molecular background of evolutionary and developmental origin of complex animal cells and tissues.

GSA2018_APCC664183

Identification of the sex-associated SNP markers of olive flounder (Paralichtys olivaceus)

GSA: Bioinformatics and genomics

GSA2018 Abstract - Poster

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The olive flounder Paralichthys olivaceus, is one of the most economically important marine aquaculture species, with a selective breeding program to increase aquaculture production in Korea. Production of monosex female stocks is desirable in commercial production since females grow faster and mature later than males. Understanding the sex

determination mechanism and developing sex-associated markers will shorten the time for the development of all-female production, thus decreasing the costs of farming. The sex determination mechanism is presently unknown in olive flounder. In this study, we found the sex-associated single nucleotide polymorphism (SNP) markers by next-generation sequencing together with bioinformatics analysis. There were 18,867 SNP loci between males and females. 3 SNP markers, that show significant differences in sexes, were identified, and all of these markers were located in chromosome 9. In order to validate sex-associated markers, artificial gynogenetic diploid, which are genetically females, was produced. A genotype analysis on these 3 SNP markers in males and females showed that female, gynogenetic diploid, and pseudomales are 100% homozygous and 90% in male individuals are heterozygous. The ratio of males are not 100% heterozygous because homozygous females, due to environmental factors controlling sex differentiation in females, are sex-reversed pseudomales. Meanwhile, around 1:1 sex ratio was observed in a total of 450 juvenile offspring from three other families. To culture all-female juvenile olive founders, we analyzed SNP genotypes in 100 males. The selected 10 pseudomales mated with genetically confirmed females, and SNP genotypes of their juveniles were then determined. As a result, these juveniles were 100% females genetically. The development of these sex-associated markers and results of their molecular sexing in different populations provide strong evidence for a sex determination system of female homogametry or male heterogametry (XX/XY) in olive flounder.

GSA2018_APCC6332

Identifying chromosomes and reconstructing their evolutionary rearrangements in Brachypodium using comparative chromosome barcoding

APCC6: Comparative, evolutionary and population cytogenetics

APCC6 Abstract - Poster

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Although Brachypodium is a small genus of temperate grasses with B. distachyon as a model organism, the entire genus represents a useful model system in various areas of plant molecular cytogenetics. The favourable cytological features of Brachypodium species and already existing resources permitted the first true chromosome painting in monocotyledonous plants. The Brachypodium genus consists of c. 20 species, which are quite diverse in their basic chromosome number, nuclear genome size and ploidy levels. This makes their exact phylogenetic relations complex and still far from being resolved. Recent synteny-based paleogenomic analyses have identified polyploidisation and dysploidy as the prime mechanisms that are responsible for the diversity in plant karyotypes and have indicated that nested chromosome fusions (NCF) are crucial in shaping the chromosome structure in grasses. Although this provided insight into the putative numbers of protochromosomes in the monocot progenitors and permitted the karyotypes of some present-day grasses, including those of B. distachyon to be connected with their hypothetical ancestral karyotypes, these studies did not involve any other Brachypodium representatives. Here, we present the comparative characteristics of various Brachypodium karyotypes using comparative multicolour FISH with chromosome-specific probes. In order to gain detailed insight into the structure and evolution of individual

chromosomes at the cytomolecular level, we conducted cross-species FISH mapping with a series of BAC clones that were derived from chromosomes Bd1-Bd5 of B. distachyon and ordered on the physical map. Using this approach, we demonstrated the presence of NCFs and other chromosome rearrangements across the genus, some of which were strictly genome-specific. Our studies, in particular when combined with the results of the ongoing whole genome sequencing projects and further molecular phylogenetic analyses, should contribute to resolving the still enigmatic phylogenetic relations within the genus. This study was supported by the National Science Centre Poland (grant no. DEC-2012-04-A-NZ3-00572 and DEC-2015-18-M-NZ2-00394).

GSA2018_APCC699186

Identifying Homozygous Genotypes Associated with Early-Onset Lymphoma in Bullmastiff by Melting Curve Analysis

GSA: Genetics and medicine

GSA2018 Abstract - Poster

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Lymphoma is one of the most common hematopoietic malignancies affecting both humans and dogs. In dogs, some breeds are particularly predisposed to lymphoma, and Bullmastiff is one of them. Based on our laboratory's previous research on Bullmastiff dogs affected by lymphoma, a region of the size of 1.2Mb on CFA13 containing five haplotypes composed of 20 single-nucleotide polymorphisms (SNPs) are highly associated with early-onset lymphoma in the dog. According to the GWAS, a high percentage, 77.8%, of affected Bullmastiff dogs were found homozygous on those SNPs compared to low frequency of the unaffected Bullmastiff dogs or other breeds. On top of that, five SNPs are specified to determine whether the dog is homozygous or heterozygous through the related regions and hereby categorized into Risk, Carrier, and Normal groups. In a bid to distinguish genotypes of the tested samples quickly and economically, we developed a method combining quantitative real time polymerase chain reaction (qrt-PCR or qPCR) and SYBR Green melting curves. The concordance and accuracy of our results elucidated that melting curves analysis enable the identification of the homozygosity of SNP in a rapid, inexpensive, and reliable way. CFA13 has been reported to be related to several canine hereditary diseases while it also contains proto-oncogene c-MYC and a region that is syntenic with human PVT1. In short term, our study is expected to be helpful for revising breeding strategies for Bullmastiff population; long-term objective will be to improve the health of both dogs and humans. Link for Fig. https://drive.google.com/file/d/1sXjy-rjYL353hkuO3vbiGCLVRJZmg_WD/view?usp=sharing

GSA2018_APCC67611

Identifying the drivers of landscape genetic structure in a range expanding damselfly

GSA: Evolutionary genetics and comparative genomics

GSA2018 Abstract - Oral

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Identifying the drivers of landscape genetic structure in a range expanding Damselfly Range shifts induced by climate and environmental change have been documented in many insect taxa, leading to disruptions of genetic connectivity between populations with compounding effects of habitat fragmentation. The effect of the intervening landscape on gene flow is important as maintaining genetic diversity facilitates adaptation to changing local environments. Populations of the damselfly, Ischnura elegans, have been documented to be undergoing a poleward range expansion in northern Europe and show environmental selection signatures and decreased genetic admixture towards its range limit in Sweden. Using 3,809 Single Nucleotide Polymorphisms (SNPs) derived from RAD sequencing data, we evaluated landscape genetic connectivity between 25 populations of I. elegans (n = 426 individuals) extending ~600km from southern Sweden to the northern expansion front. Using landscape resistance modelling of non-linear relationships with genetic distance, we test for effects of mean annual temperature and land cover on gene flow. A model selection approach based on mixed effects modelling and Akaike Information Criterion (AIC) was used, which showed that genetic distance is best explained by a non-linear relationship with mean annual temperature, indicating threshold effects on genetic connectivity. Additional resistance to gene flow was found for landscape variables, which was most explained by urban/industrial developed areas and forests. Increased landscape resistance to gene flow towards the northern range limit was associated with lower relative abundance at sites and

larger body size. Threshold effects of temperature on landscape genetic connectivity were similar to patterns of environmental selection previously characterised in the same populations. Our study has implications for using landscape genetic resistance modeling to disentangle demographic from selection processes within heterogeneous landscapes during range expansion.

GSA2018_APCC629172

Illuminating the evolutionary history of the endomesoderm gene regulatory network: ChIP-Seq of Wnt pathway components in Sycon capricorn

GSA: Development and cellular genetics

GSA2018 Abstract - Poster

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By modifying gene regulatory networks (GRNs) present the last common ancestor of the Eumetazoa (cnidarians and bilaterians), animals evolved a diversity of complex body plans. Yet, the question of what specific changes in GRNs facilitated the emergence of complex animal bodies remains largely unexplored. My Honours project explores the evolutionary history of a GRN which specifies endomesoderm and its derivatives (gut and muscle tissue) during embryogenesis. Though endomesoderm specification in Eumetazoa is well-studied, little is known about the mechanisms that specify its potential homologues in earlier evolving metazoans. Sponges (Porifera) are considered one of the oldest surviving animal lineages, and are emerging models for exploring evolution of developmental GRNs. Components of the canonical Wnt pathway (a key element of the endomesoderm GRN) are expressed in a potential endomesoderm homologue – the choanoderm – in the calcareous sponge Sycon. However, whether the Wnt pathway plays a role in sponge choanoderm specification similar to its role in eumetazoan endomesoderm is unknown. Using custom-raised antibodies against TCF, the transcription factor of the canonical Wnt pathway, and a ChIP-Seq protocol developed for Sycon capricorn, I will identify Wnt target genes and compare them to those known to be involved in endomesoderm specification in cnidarians. The results of my study will shed light on the evolutionary relationship between the sponge choanoderm and the eumetazoan endomesoderm.

GSA2018_APCC65880

Integrated karyotyping of woodland strawberry (Fragaria vesca) with oligopaint FISH probes

APCC6: Cytogenetics in the genomics era (Plants and Animals)

APCC6 Abstract - Poster

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Chromosome identification is critical for many aspects of cytogenetic research. However, for Fragaria vesca, definitive identification of the individual chromosomes is almost impossible because of their small size and high similarity. Here we demonstrated that bulked oligonucleotide (oligo) probes can be used as chromosome-specific DNA markers for chromosome identification in F. vesca. Oligos specific to an entire pseudochromosome in the draft genome of F. vesca were identified and synthesized as a library. In all, we synthesized 6 oligo libraries corresponding to 6 pseudochromosomes of F. vesca, respectively. These libraries were amplified and labeled as probes for fluorescence in situ hybridization (FISH). Two rounds of multicolor FISH analysis were sequentially conducted on the same metaphase cells with each round including three probe libraries, which permitted simultaneous identification of all chromosomes of F. vesca. Moreover, 45S and 5S rDNA were mapped to chromosomes 1, 2 and 7, respectively. A karyotype of metaphase chromosomes was constructed, which was the first FISH-based molecular cytogenetic karyotype of F. vesca. Our study can serve as a basis for future comparative cytogenetic research through cross-species chromosome painting using bulked oligo probes and will facilitate applied breeding technologies that rely upon identification of chromosomes in the genus Fragaria.

GSA2018_APCC677166

Inter-individual Genetic Variability in The Expression Frequency of The Human Common Fragile site FRAXB among Healthy Individuals

APCC6: Other

APCC6 Abstract - Oral

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Cancer cells are characterized by their ability to grow at an uncontrolled, quickened and indefinite pace. Point mutations in tumor suppressor genes and proto-oncogenes, changes in chromosome copy numbers and structure are various forms of genome instability that constitute a hallmark of cancer. Several studies suggest a strong correlation and molecular colocalization between cancer breakpoints and breaks at nonrandom genomic loci called common fragile sites (CFSs). This finding highlights the importance of CFSs in oncogenesis. CFSs are site-specific breaks that are observed on metaphase chromosomes when cells are cultured under stress conditions. They are found in all individuals and are mainly induced by low doses of aphidicolin (Aph), a partial inhibitor of DNA polymerases alpha, delta and epsilon. In this study, we propose to determine the expression frequency of FRAXB, a CFS located at Xp22.31 and one of the most CFS expressed in the human genome, in healthy persons of different ages. Blood samples from several donors were collected, cultured and lymphocytes were then treated with 0.2 µg/ml of Aph. RP11-483M24, a probe specific for FRAXB hotspot of breakage was used in FISH in order to detect FRAXB breaks. One hundred fluorescent signals were analyzed per donor using fluorescence microscopy. The results show that the percentage of breakage ranges from 6% to 25% with several folds increase among females in the twenties and from 3% to 19% among males of the same age suggesting the presence of CFS expression variability among individuals of the same age and gender. Such inter-individual variability in CFS bre akages could be due to personal difference in the genetic background and can explain why there is variability in cancer incidence among individuals with the same age subjected to the same environmental conditions and mutagens. This finding can be used as a biomarker for cancer prevention.

GSA2018_APCC654184

Interplay between the epigenome & chromatin signaling enzymes in metastatic cancers

GSA: Genetics and epigenetic regulation

GSA2018 Abstract - Oral

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Though all genes exist within every cell in the human body, only a defined gene expression program is executed at any time via reprogramming of the epigenome in response to environmental cues. These dynamic events are elegantly orchestrated by writer and eraser enzymes; generating a ‘histone code’ within the epigenetic landscape of genes. The implications of targeting a novel class of epigenetic enzymes are beginning to be appreciated in clinical oncology.In this seminar, I will present my laboratory’s recent work unravelling a key role for such writers in metastatic cancers. I will also discuss our development of novel classes of liquid biopsy based biomarkers and companion epi-inhibitors as well as potential implications for the utility of epi-therapy.

GSA2018_APCC68058

Investigating potential roles for FUBP1 and CIC in oligodendroglioma using Drosophila models

GSA: Development and cellular genetics

GSA2018 Abstract - Oral

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Glioma, the most common primary brain tumour, is a highly heterogeneous disease arising from glial cell types including astrocytes and oligodendrocytes. These slow growing but invasive tumours are difficult to treat with unpredictable outcomes, necessitating an analysis of their molecular basis to improve prognosis and treatment. Far upstream binding protein 1 (FUBP 1), a single stranded DNA binding protein, was first discovered as a transcriptional activator of the MYC oncogene. Consistently with this function, FUBP1 is upregulated in many tumour types, including breast, liver, bladder, kidney lung cancer and glioblastoma. However, in oligodendroglioma FUBP1 is one of the top-ranked mutated genes predicted to result in loss-of-function, suggesting that FUBP1 has capacity for anti-proliferative effects in this context. Interestingly, FUBP1 mutations frequently co-occur with loss-of-function mutations in Capicua (CIC), a downstream regulatory factor of receptor tyrosine kinase (RTK) signaling pathways. Although FUBP1 and CIC are predicted to behave as tumour suppressors in glial lineage tumours, the capacity of mutations in either gene alone or in combination for driving glioma has not been fully understood. Hence, in order to identify the effect of deleted FUBP1 and CIC in glial cells and brain development, our research employs Drosophila which has been widely used for studying mechanisms of tumourigenesis due to the conservation of key molecular pathways involved in human cancer. In Drosophila, Capicua/CIC has characterized roles in regulating cell fate, while the role of human CIC has not been fully elucidated. Moreover, Drosophila FUBP1 (called Psi) plays conserved molecular functions in regulating expression of MYC. Therefore, the individual and cooperative effects of FUBP1/Psi and/or CIC/Capicua depletion on glial lineage behaviour and brain development in Drosophila will provide insights into molecular mechanisms underlying glioma development. Additionally, we aim to determine whether FUBP1 and/or CIC mutation(s) drive tumorigenesis, using mouse xenograft glioma models.

GSA2018_APCC68747

Investigating the evolution of complex, novel traits using whole genome sequencing and molecular palaeontology

GSA: Evolutionary genetics and comparative genomics

GSA2018 Abstract - Oral

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96 Normal 0 false false false EN-US X-NONE X-NONE /* Style Definitions */ table.MsoNormalTable {mso-style-name:"Table Normal"; mso-tstyle-rowband-size:0; mso-tstyle-colband-size:0; mso-style-noshow:yes; mso-style-priority:99; mso-style-parent:""; mso-padding-alt:0cm 5.4pt 0cm 5.4pt; mso-para-margin:0cm; mso-para-margin-bottom:.0001pt; mso-pagination:widow-orphan; font-size:12.0pt; font-family:Calibri; mso-ascii-font-family:Calibri; mso-ascii-theme-font:minor-latin; mso-hansi-font-family:Calibri; mso-hansi-theme-font:minor-latin;} Understanding how new biochemical pathways evolve in a sexually reproducing population is a complex and largely unanswered question. We are using PacBio whole-genome sequencing and deep population resequencing to explore the evolution of a novel biochemical pathway in yeast over several thousand generations. Growth of wild Saccharomyces cerevisiae strains on the pentose sugar xylose is barely perceptible. A mass-mated starting population was evolved under selection on Xylose Minimal Media with forced sexual mating every two months for four years, producing a population that can grow on and utilise xylose as its sole carbon source. We are now using a novel “molecular

palaeontology” approach to trace the evolutionary process and identify functionally significant loci under selection. Populations at seven key time points during their evolution have been sequenced using Illumina short-read sequencing. In addition, all the parental strains from the founding population have been subject to PacBio de novo whole-genome sequencing and assembly. By constructing reliable whole genomes of the ancestors of our populations, we can the trace evolution of these populations over time. We can therefore track the trajectory of allele frequencies through time, identifying the contributions of different founding strains and novel mutations. We are using these data to estimate the proportions and regions of the genome that have evolved neutrally, under purifying selection, or adaptively in response to xylose selection. Our unique array of both extant and past, but not extinct, populations allow us to test popular models of molecular evolution.

GSA2018_APCC68557

Investigating the role of MYD88 in Lymphoma

GSA: Development and cellular genetics

GSA2018 Abstract - Poster

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Lymphoma is the sixth most common form of cancer, with 90% of cases belonging to a heterogeneous group of lymphomas called Non-Hodgkin Lymphoma (NHL). One of the most common types of NHL, Diffuse Large B Cell Lymphoma (DLBCL), is characterized into two subtypes on the basis of differential gene expression: Activated B Cell (ABC) and Germinal Centre (GC), of these ABC is the most aggressive and least curable form. Recurrent somatic mutations in MYD88 were initially identified in ABC-DLBCL patients. MYD88 is an adaptor protein that transmits innate immune signalling from Toll-like receptor (TLR) and interleukin-1 (IL-1) receptor to the nuclear factor-kappa B (NF-□B) pathway, for immune and inflammatory responses. The most frequent MYD88 mutation in lymphoma is a single amino acid variant L265P, identified in approximately 30% of ABC-DLBCL and 90% of Waldenström Macroglobulinemia (WM) patients. Apart from the L265P mutation, additional mutations in MYD88 have been reported in lymphoma. Moreover, genome-wide analysis of ABC-DLBCL and WM indicates that MYD88 L265P is preferentially found in combination with mutations in the B-cell antigen component CD79B and chemokine receptor CXCR4, which result in further induction of NF-κB pathway.We are currently investigating the role of MYD88 in both normal development and immune cell function. For this purpose, we have developed Drosophila melanogaster and mouse model systems. In the fly model, we are investigating potential effects of Myd88 knockdown and CRISPR-generated Myd88 I335P substitutions (designed to mimic oncogenic MYD88 L265P) on hematopoietic development

and immunity. In the mouse model, compound mutations (MYD88 loss-of-function together with MYD88 L265P) are being examined to determine potential of MYD88 L265P as a therapeutic target. Further to this, we are testing the combined effect of the MYD88 L265P mutation and CD79B mutations in our B cell lymphoma mouse models.

GSA2018_APCC650187

Isolation and characterization of centromeric DNA sequences in diploid and allotetraploid Brassica species

APCC6: Comparative, evolutionary and population cytogenetics

APCC6 Abstract - Poster

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Polyploidy is an important event during species evolution, especially in flowering plants. The Brassica genus provides a well system for studying chromosome evolution, since it containing three basic diploid species and three allotetraploid species. Centromeres, as an important unit of chromosomes, exhibit a critical role in genome polyploidization events during evolution. However, the sequence components of centromeres in Brassica species are largely unknown. Here, we employed ChIP-seq based method to isolate the centromeric DNA sequences in both diploid and allotetraploid Brassica species. It is revealed that a conserved centromeric satellite DNA element, named CentBr, is the most abundant sequence in the five of the six Brassica species except for the diploid species B. nigra, in which the retrotransposon sequences exhibit the most abundance. Futhremore, using FISH analysis, we identified that: 1) sereval sequences are conserved in all six species, which mightbe the most ancient centromeric elements in Brassica species; 2) several sequences are specific in B.rapa , B. nigra or B. oleracea, which were speculated to be newly formed elements after these species divergence; 3) futhermore, several sequences are with similar signals in both B.rapa and B. oleracea, but absent in B. nigra. However, interestingly, we do not find any sequence observed in B.rapa and B. nigra but not B. oleracea, or in B. oleracea and B. nigra but not B.rapa. All these results indicate that B.rapa and B. oleracea are more close in between on evolution, but both separate from B. nigra, which centromeres are composed of most retrotransposons. The reason of this phenomena might be that B. nigra was under less evolution pressure because of less application in production than B.rapa and B. oleracea. These results will provide new insights into Brassica genome evolution process from centromeric DNA sequences viewpoint.

GSA2018_APCC63252

Isolation of centromeric repeated sequence from common long-tailed macaque (Macaca fascicularis fascicularis) by construction of genomic fosmid library and chromosome mapping

APCC6: Cytogenetics in the genomics era (Plants and Animals)

APCC6 Abstract - Poster

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Common long-tailed macaques (Macaca fascicularis fascicularis, 2n=42) are commonly used non-human primate models in a wide range of scientific experiments. Their whole genome sequence is available; however, the assembly process remains complicated for repeated non-coding sequences in heterochromatic regions such as at the centromeres. To upgrade whole genome structure, highly repeated DNA sequences were isolated by constructing a genomic fosmid library prepared from long-tailed macaque genomic DNA (gDNA) and fluorescence in situ hybridization (FISH) mapping. A total of 768 clones were hybridized with long-tailed macaque gDNA, while 14 clones exhibiting intense positive signals were collected to examine their chromosomal localization. Only five clones were mapped to the centromeric region of all chromosomes, except for the Y chromosome with smaller size compared to X and Y chromosomes of other macaques. This suggests that centromeric repeated sequences were separately homogenized between X chromosome-autosomes and Y chromosome, possibly leading to degeneration of the Y chromosome in long-tailed macaque. Different FISH signal intensities were observed among the five clones and chromosomes, suggesting that each repeated clone might contain a variety of repeated sequences in the genome. All those five clones were sequenced and determined the molecular structure of centromeric heterochromatin.

GSA2018_APCC62195

Karyotype Analysis of Six Lilium Species by FISH Technique

APCC6: Applied cytogenetics (e.g agriculture and biomedical)

APCC6 Abstract - Poster

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Lilies have many attractive characteristics such as fascinating fragrance, shape, and beauty. Various lily species have been widely used as a food and are equally versatile as medicine in the Middle East and Eastern Asia. Today, lily is one of the most important ornamental plants used for cut flowers, potted plants, and garden plants all over the world. Lilium species are interesting materials for cytogenetic studies. Most of Lilium species were known as a diploid (2n=2x=24) with basic chromosome number is x=12 but with the exception of triploid (2n=3x=36) species in L. tigrinum. Fluorescent in situ hybridization (FISH) is a powerful molecular cytogenetic technique using chromosomal markers that reveal the positions of specific genes, such as ribosomal DNAs, thereby making it easy to identify individual chromosomes. We have constructed detailed karyotypes of L. sargentiae, L. henryi, L. davidii, L. formosanum, L. leichtlinii, and L. regale. Only one pair (two loci) of 5S rDNA signal was detected in all six lily species. Two pairs (four loci) of 45S rDNA signals were found in L. henryi, three pairs (six loci) in L. formosanum, four pairs (eight loci) in L. sargentiae, L. davidii, and five pairs (ten loci) in L. leichtlinii, L. regale. The results clearly demonstrate that FISH analysis could be an important tool to elucidate phylogenies among the section Sinomartagon at the chromosomal level. (This work was carried out with the support of “Cooperative Research Program for Agriculture Science and Technology Development (Project No. PJ012804)” Rural Development Administration, Republic of Korea)

GSA2018_APCC682122

Karyotypes of two Australian native freshwater fish Golden perch and Murray cod (Percichthydae)

APCC6: Comparative, evolutionary and population cytogenetics

APCC6 Abstract - Oral

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Karyotypic data from Australian native freshwater fishes are scarce, having been described from only few species. Here we present karyotypic data from two Australian iconic native freshwater species, golden perch (Macquaria ambigua) and Murray cod (Maccullochella peelii). Both species have diploid chromosome 2n = 48. All chromosomes of Golden perch are telocentric while in Murray cod 20 are acrocentric and 28 are telocentric. Using standard and molecular cytogenetics techniques we have also identified male heterogametic (XX/XY) sex chromosomes system in both species. We also mapped repetitive sequences and NOR and discussed chromosome evolution within Percichthyidae fishes.

GSA2018_APCC653119

Know your enemy: a population genetics approach to management of a native Macadamia pest

GSA: Ecological genetics

GSA2018 Abstract - Oral

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Macadamia lace bugs are very economically important pests. Macadamias are native to subtropical eastern Australia, and northern New South Wales produces almost half of world macadamia nut output. However, this production is severely threatened by macadamia lace bugs (Ulonemia spp.). We are using genetic methods to address three aspects of lace-bug control. First, Ulonemia is a member of the huge global family Tingidae, which includes many serious pests. These species are likely to have different pest characteristics, insecticide susceptibilities, etc., yet often it is difficult to say how many species are present. Therefore, we aimed to improve knowledge of species identification and genetic structure. We have shown that there is more than one species in the Australian orchards, contrary to some previous suggestions. Secondly, the small, densely packed, orchards in northern New South Wales are managed independently, but this may not be optimal if the bugs frequently disperse between orchards. Therefore, we are genetically assessing dispersal of Ulonemia. We used SNPs and several metrics (Jost's D, G"st, Shannon Mutual Information) to determine the relationships between U. decoris populations in northern New South Wales. Preliminary results indicate high movement over surprisingly large scales. Thirdly, gene exchange between wild and plantation populations of Ulonemia might affect control outcomes, so we will investigate exchange levels, using the same genetic methods. Our results will provide macadamia growers crucial information, to help design optimal integrated pest management practices for of these serious pests.

GSA2018_APCC61816

Landscape genomics of a widespread agricultural pest, the wingless grasshopper (Phaulacridium vittatum)

GSA: Evolutionary genetics and comparative genomics

GSA2018 Abstract - Oral

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Examining adaptive genetic variation in pests is beneficial for forecasting pest impacts andaiding management decisions, especially in the context of shifting climate regimes. Whilepests are often widespread with high gene flow, local adaptation may still occur,highlighting the concurrent need to examine the interaction of gene flow and landscapeconnectivity with patterns of selection. The wingless grasshopper (Phaulacridium vittatum)is a widely distributed native herbivore in Australia and is a major pest of agriculturalpastures, crops and tree nurseries. Using a combination of landscape genetic resistancemodelling, Fst outlier and Environmental Association Analysis (EAA), we disentangle the roleof landscape and environmental variables on connectivity and adaptive responses. Wesampled P. vittatum across a ~900 km environmental sampling gradient at 19 sites in NewSouth Wales, Australia. Each individual was genotyped at 11,464 single nucleotidepolymorphisms (SNPs) obtained from a modified ddRAD protocol. Genetic structureanalysis revealed low levels of genetic differentiation across the latitudinal gradient. Fstoutlier methods detected a total of 20 outlier SNPs and 260 SNPs were identified by EAA.Amongst 260 SNPs, greater numbers of SNPs were associated with temperature, latitude,and few associations were found with foliage cover, soil moisture and soilacidity, indicating correlation of allelic diversity with climatic and environmental features.Annotation of candidate SNPs indicated gene functions relevant for environmentaladaptation. Our study finds signatures of selection in P.vittatum along environmentalgradients despite low landscape resistance to gene flow, which has implications forspatially-explicit management of this agricultural pest.

GSA2018_APCC69554

Marsupial and monotreme cathelicidins: characterisation and antimicrobial activity

GSA: Genetics and medicine

GSA2018 Abstract - Oral

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The rise in antimicrobial resistance and paucity of new antimicrobial compounds calls for alternatives to traditional antibiotics. Antimicrobial peptides (AMPs) have emerged as potential candidates. Cathelicidins are a major family of AMPs in mammals which form part of innate immunity through antimicrobial and immunomodulatory functions. Marsupial and monotreme cathelicidins are of particular interest due to their involvement in protecting immunologically naive young during development in the pouch via expression in the pouch lining and milk, where they modulate microbial flora and provide passive immunity. As such, the cathelicidin gene family has expanded in marsupials and monotremes, with a high number of cathelicidins in the tammar wallaby, gray short-tailed opossum and platypus. However, our knowledge is limited to these species and functional studies are lacking. We charcaterised cathelicidins in the Tasmanian devil, koala and echidna, and investigated the antimicrobial function of all known marsupial and monotreme cathelicidins. As expected, cathelicidins have expanded in the Tasmanian devil and koala, resulting in a high number of cathelicidins which were widely expressed throughout the body, including in pouch lining and milk. Only a single cathelicidin was identified in the echidna due to the quality of the genome. Out of 26 cathelicidins tested, six displayed broad-spectrum antibacterial activity against gram-negative and positive bacteria, including methicillin-resistant Staphylococcus aureus (MRSA). One koala cathelicidin rapidly inactivated Chlamydia pecorum and significantly reduced the number of chlamydial inclusions in vitro. Cathelicidin activity was reduced in the presence of serum and whole blood, and peptides displayed varying levels of haemolytic and cytotoxic activity. Many cathelicidins did not display antimicrobial activity and future work is required to explore their potential immunomodulatory properties. These results have advanced our understanding of cathelicidins in marsupials and monotremes on a genetic and functional level, and highlight their potential as novel therapeutics in the future.

GSA2018_APCC610193

McGISH identifies the parental genomes in artificial hybrids of the wind orchid Neofinetia falcata (syn. Vanda falcata)

APCC6: Cytogenetics in the genomics era (Plants and Animals)

APCC6 Abstract - Poster

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The epiphytic wind orchid, Neofinetia falcata (now transferred in the genus Vanda) is distributed throughout the Japan, Korea and China. Initially, it was popular among shoguns and samurai during the Edo period in Japan. Since then, a variety of mutants locally known as “Fuukiran” (orchid of the rich and noble in Japanese) are still maintained and cultivated for their subsequent improvement of horticultural traits. Neof. falcata has been hybridized with Vanda, Ascocentrum, Rhynchostylis, Phalaenopsis etc. to introduce useful traits, such as cold tolerance, fragrance, and the dwarf growth habit etc in the cultivars. In this study, chromosome painting by multi-color GISH accurately distinguished the parental genomes in the inter-generic artificial hybrids viz. Ascofinetia Cherry Blossom (Neof. falcata×Ascocentrum ampullaceum), Ascofinetia Petite Bouquet (Ascf. Cherry Blossom×Asctm. ampullaceum), Darwinara Charm (Neof. falcata×Vascostylis Tham Yuen Hae), Neosedirea Summer Stars（Neof. falcata×Phalaenopsis japonica）Neostylis Lou Sneary (Neof. falcata×Rhynchostylis coelestis), Neostylis Pinky (Neof. falcata×Rhynchostylis gigantea), Yonezawaara Blue Star (Neostylis Lou Sneary×Vanda coerulea). McGISH was performed using total genomic DNAs of the parental species as probes. Firstly, in two-way crossing, both parental genomes that contribute 19 chromosomes respectively to the hybrids were clearly distinguished by GISH. The allocation of parental genomes at interphase as well as prophase nuclei was also observed. Basically, these hybrids were diploid in nature however occasional occurrence of triploids, tetraploids and mixoploids were also encountered. In plural crossing beyond the

genus, the genome of one of the parental species that used in the last crossing was clearly identified. However, a mixing of genomes was observed in the remaining half chromosomes. The identification of genomes architecture vis-a-vis origin of chromosomes in economically valued orchids/hybrids with a complicated lineage is quite prerequisite for subsequent crop improvement programs.

GSA2018_APCC616158

Measurement of nuclear DNA content of the Onobrychis accessions maintained in The Margot Forde Germplasm Center, NZ as a measure of genebank accession identity

GSA: Evolutionary genetics and comparative genomics

GSA2018 Abstract - Poster

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The genus Onobrychis contains more than 150 species. Some of these species are cultivated widely in some parts of the world as forage legume due to their beneficial traits such as, high protein content, high yield, high digestibility, cold tolerance and drought tolerance. Taxonomic identification of the Onobrychis species is challenging due to variation and lack of morphological diversity. Additionally, species of this genus have different base chromosome number of either n=7 or n=8 and polyploidy is a common phenomenon in the genus.Identification of the species within the genus Onobrychisand determining their chromosome numbers are crucial before initiating any plant breeding program for the genus. This is beacause of the potential issues such as genetic incompatibility and sterility that may occur during the crossings, which may waste the limited available resources such as material and human resources. In this study, we used flow cytometry to determine nuclear DNA content of 38 Onobrychis acessions from nine species, maintained by The Margot Forde Germplasm Center (MFGC), Palmerston North, New Zealand for the first time. Based on the results of the study, 2C nuclear DNA content of the accessions varied between 1.0 and 2.87 pg. These results indicate that the collection includes both diploids and polyploids. Except one accession of O. viciifolia and two accessions of O. transcaucasia, all other accessions of these two cultivated species are tetraploid (2n=28). Therefore, these three accessions should be further investigated to confirme their label information by distinguishing the accessions taxonomically. Further, the results of the study indicate that all accessions or the contents of their seed packets are pure as they don't include plants with different ploidy levels or misidentified seeds although only a few plants per accession were analysed.Acknowledgements: This study was supported by a TUBITAK grant (TOVAG-215O526).

GSA2018_APCC630130

Methylation-sensitive DArTseq analysis to identify sex-linked markers in an Agamid lizard

GSA: Evolutionary genetics and comparative genomics

GSA2018 Abstract - Poster

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Chromosomes may contain sex-specific genes or sequences that can reveal their sex determination mode (XY or ZW) when sex chromosomes are cryptic or homomorphic. Sex –linked markers can also reveal sex determining genes and even the genetic architecture of sex chromosomes. It has been hypothesised that the differential methylation of the promoters of genes at sex-specific temperatures is involved in sex determination and also involved in differentiation of sex chromosomes, yet a full understanding of this mechanism is still yet unknown. Although, differences in gene methylations occurring between genetic males and females have been observed in several species of fishes and

turtles, information on these differences at genetic marker level is yet lacking. Methylation DArTseq, a genotyping by sequencing (GBS) approach, employs a combination of genome complexity reduction and next generation sequencing methods and have the potential to identify differential gene methylation between sexes. In this study, we investigated oriental garden lizard (Calotes versicolor) genome by using this method to discover sex-linked markers that are differentially methylated. Both methylation sensitive and non-sensitive restriction enzymes were used (m-DArTseq and DArTseq) and we analysed in silico DArTseq markers (presence and absence) for sex-linkage. From m-DArTseq, 12 markers were identified to be substantially methylated in females and 2 in males (from 7 females and 7 males). Of these, 2 markers were only found in females. We also identified 241 female and 142 male specific markers from DArTseq, of which 60 were only found in females and 5 were only found in males (from 4 females and 4 males). Our preliminary data suggest, like other dragon lizards, Calotes versicolor also has a female heterogametic (ZZ/ZW) system. We are currently screening larger sample size to validate the efficacy of these markers.

GSA2018_APCC66249

Microsatellite repeat motif distribution: computational whole genome sequence analysis vs chromosome mapping reveals different levels of copy number variation and localization in common long-tailed macaque (Macaca fascicularis fascicularis) genome

APCC6: Cytogenetics in the genomics era (Plants and Animals)

APCC6 Abstract - Poster

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Microsatellites, as tandem repeats of small stretches of DNA motifs, are widespread in genomes. Availability of the whole genome sequence of common long-tailed macaque (Macaca fascicularis fascicularis, MFA), a commonly used non-human primate model for biomedical research, has made it possible to carry out an overall microsatellite analysis. However, the assembly process remains complicated for repeated non-coding sequences, leading to failure of computational analysis for identification of chromosome-specific microsatellite distributions and their copy numbers. Here, fluorescence in situ hybridization (FISH) mapping with 16 microsatellite repeat motifs and telomeric (TTAGGG)n was performed on MFA chromosomes. Hybridization signals of (TTAGGG)n sequences were observed at chromosomal telomeric ends and dispersed (AT)n, (CA)n and (GC)n were mapped to all chromosomes with different signal intensities. Interestingly, (CAG)n were localized to MFA7q interstitially, suggesting that amplification of these microsatellite motifs occurs independently in the genome of MFA. No signal was observed from the other 12 microsatellite motifs ((GA)n, (CAT)n, (CGG)n, (AAT)n, (CGC)n, (CAA)n, (AAAC)n, (AATC)n, (ACGC)n, (AATG)n, (AGAT)n and (AAGG)n), although computational whole genome sequence analysis showed plausible physical locations. This indicates that copy number variation among subspecies of long-tailed macaques or copy number of these sequences may be too low for FISH mapping sensitivity. These results provide a reference for identification of chromosome-specific microsatellite markers which can facilitate understanding regarding the structure of the long-tailed macaque genome and genetic linkage map construction.

GSA2018_APCC626101

Mitochondrial DNA, Mother’s Curse and Me

GSA: Other

GSA2018 Abstract - Oral

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Mitochondria are central to eukaryotic life. Mitochondria retain their own genome (mtDNA) and this molecule has important roles in energy production and a growing list of other processes. Mitochondrial DNA is almost universally inherited solely through the female lineage, which provides opportunity for selection to operate asymmetrically between males and females. A growing body of work suggests strongly that mtDNA mutations may have stronger effects on phenotype in males than females — an idea I termed 'Mother's Curse’. In this talk I will review the development of the Mother's Curse hypothesis and the still accumulating evidence that supports it. I will also explore some areas in evolution and ecology where this theory might have impact or application, including a new form of biocontrol, and a few unexpected twists that have emerged from this work over the years. Finally, because science discovery is seldom, if ever linear, I will also discuss a few unexpected twists that emerged from this work over the years and some of the questions we have yet to resolve.

GSA2018_APCC61314

Molecular cytogenetic screening for chromosomal abnormalities in agricultural species

APCC6: Applied cytogenetics (e.g agriculture and biomedical)

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Chromosome translocations have been shown to reduce fertility in agricultural species leading to reduced pregnancy rates and/or small litter sizes. With an increasing emphasis in modern farming on the use of a small population of males for artificial insemination, the potential economic and environmental costs of using subfertile boars and bulls are significant. An increased need for novel tools to facilitate rapid, cost effective chromosome translocation screening is therefore required. Current screening is performed by karyotyping; however, this relies on significant

expertise and is limited in its ability to identify cryptic translocations. To address this problem, we developed a novel method for translocation screening in pigs and cattle using subtelomeric probes and multi-target fluorescence in situ hybridisation. Probes designed to localise in the subtelomeric region of the p-arm and q-arm of each chromosome were directly labelled with FITC or Texas Red. These were subsequently applied to a ‘Multiprobe’ device, enabling simultaneous detection of each chromosome on a single slide - significantly shortening analysis time. Initial experiments led to the discovery of a series of incorrectly mapped regions in the porcine genome assembly, highlighting the importance of accurate physical mapping of newly sequenced genomes. Multiple porcine chromosome translocations were identified using this approach, including a cryptic translocation that was determined to have resulted in a loss of ~£20,000 to the breeder. This system therefore provides a robust and comprehensive method of screening for chromosomal abnormalities in agricultural species, with the benefit of potentially large cost savings to the industry.

GSA2018_APCC61690

Molecular Cytogenetics for Plant Genetics and Breeding

APCC6: Cytogenetics in the genomics era (Plants and Animals)

APCC6 Abstract - Oral

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Physical mapping of unique nucleotide sequences on specific rice chromosome regions was performed using a combination of chromosome identification and highly sensitive fluorescence in situ hybridization (FISH). FISH is an effective method for the physical mapping of genes and repetitive DNA sequences on chromosomes. Increases in the detection sensitivity of smaller DNA sequences and improvements in spatial resolution have ushered in a new phase in FISH technology. Thus, it is now possible to perform FISH on somatic chromosomes, pachytene chromosomes, and even on extended DNA fibers (EDFs). Pachytene-FISH allows the integration mapping of genetic linkage maps and quantitative chromosome maps. Visualization methods using FISH can reveal the spatial organization of the centromere, heterochromatin/euchromatin, and the terminal structures of rice chromosomes. Furthermore, EDF-FISH and the DNA combing technique can resolve a spatial distance of 1 kb between adjacent DNA sequences, and the detection of even a 300-bp target is now feasible. The copy numbers of various repetitive sequences and the sizes of various DNA molecules were quantitatively measured using the molecular combing technique. Analysis of nuclear DNA and proteins would reveal nuclear structure and gene functions in plant development. This study describes the significance of these advances in molecular cytogenetics in rice and Brassicaceae. I discusses future applications in plant genetics and breeding studies using high sensitivity visualization techniques.

GSA2018_APCC69735

Molecular evolution of CTNNB1 and WAC gametologous genes reveal independent strata of sex chromosomes in caenophidian snakes under a short time period

APCC6: Specialised chromosomes (sex chromosomes and B chromosomes)

APCC6 Abstract - Oral

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Caenophidian snakes exhibit ZZ/ZW type genetic sex determination with female heterogamety (ZW). The W chromosomes are highly degenerated showing interspecific size variation, centromere position, and amount of heterochromatin. Comparison of the structures and functions of Z and W homologous genes (gametologous genes) is, therefore, important to understand the process of sex chromosome differentiation in caenophidian snakes. Here, partial sequences of two gametologous genes (CTNNB1 and WAC) from 24 caenophidian snakes were analyzed and the state of mutation was compared with nuclear autosomal and mitochondrial genes. Results indicated that the WACW gene had greater size variation resulting from insertions-deletions (indels) than the CTNNB1W gene in the caenophidian lineage. Average Ka/Ks in the coding region of both genes was very low (1), suggesting these gametologous genes presentednegative selection. Molecular phylogenetic plac ement showed caenophidian snake CTNNB1Z genes clustered together, while caenophidian snake CTNNB1W genes were grouped with henophidian snake CTNNB1 genes. Similar results were found in most WACZ and WACW genes, except for WACW and WACZ genes of Achrochordus javanicus (Elephant trunk snake) grouped in the same clade of each species. This result differed, with a phylogenetic relationship based on nuclear autosomal and mitochondrial genes, suggesting that evolutionary strata occurred independently at the WAC gene located on the long arm of ZW chromosomes in caenophidian snakes.

GSA2018_APCC658140

Monitoring post-release survival of the Northern Corroboree frog using Environmental DNA

GSA: Ecological genetics

GSA2018 Abstract - Oral

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Corroboree frogs are two of Australia's most iconic species. Like many any other amphibian species, populations are in decline due to the wide spread chytrid fungus Batrachochytrium dendrobatidis. With species listed as endangered (Northern Corroboree frog) or critically endangered (Southern Corroboree frog), continuing their survival through breeding programs and translocations has been essential. Evaluating the success of amphibian breeding programs and translocations can be difficult, with monitoring efforts restricted to certain times of year and often limited to the detection of adult male frogs using traditional shout-response surveys. Environmental DNA (eDNA) involves the detection of trace amounts of DNA organisms release into their environment and can detect both sexes of a target species across multiple life stages. The use of Environmental DNA as a monitoring tool could provide valuable information on the capacity of individuals to survive translocation efforts. In this study, we use eDNA as a monitoring technique, capable of detecting the presence of corroboree frog DNA non-invasively. An assay was developed targeting both species of Corroboree frog. The assay was tested on eDNA water samples from tanks containing captive reared Northern Corroboree frog eggs and tadpoles housed within a breeding facility. Northern Corroboree frog eggs were translocated to three ponds in Namadgi National Park with DNA detected at 3/3 ponds and remaining detectable at 2/3 ponds after 78 days. Incorporation of eDNA surveys into current monitoring efforts may be useful to monitor post-release survival of translocated populations in a non-invasive manner. Such knowledge can be used to inform the management of translocated populations or future release strategies.

GSA2018_APCC62885

Muscleblind regulates cell-type-specific alternative splicing of Drosophila Dscam2

GSA: Development and cellular genetics

GSA2018 Abstract - Oral

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Alternative splicing (AS) generates proteome diversity much needed in the nervous system, where a limited set of genes specifies more than 1015 neuronal connections. If AS plays a role in specifying neuronal connections, it needs to be regulated in a cell-type-specific manner. Recent work from our lab has shown that this is the case for the transmembrane protein, Down syndrome cell adhesion molecule 2 (Dscam2). We demonstrated that two alternative isoforms of Drosophila Dscam2 are expressed in distinct neuronal cell types and that this expression pattern is crucial for proper neuronal wiring. In our current work, we sought to identify the mechanism underlying Dscam2 cell-type-specific AS. In an RNAi screen, we knocked-down ~160 RNA binding proteins and identified muscleblind (mbl) as a regulator of Dscam2 AS. Mbl loss of function de-represses one Dscam2 isoform, whereas mbl overexpression activates the other. Analysis of mbl expression suggests that it is regulated in a cell-specific manner that correlates with the repressed Dscam2 isoform. Consistent with this, overexpressions of mbl lead to neuronal outgrowth defects that resemble Dscam2 isoform mis-expression. Understanding the mechanism underlying cell-type-specific Dscam2 AS could give us insights into how functional diversity is achieved by the proteome.

GSA2018_APCC65527

Museums are an untapped reservoir of genomic data: novel substrates and new horizons

GSA: Evolutionary genetics and comparative genomics

GSA2018 Abstract - Oral

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Natural history collections provide some of the deepest insight into the temporal dynamics of molecular and morphological evolution. Technological advances in DNA sequencing have allowed us to push the boundaries of what is considered suitable material for genomic analysis. Here I describe two new initiatives at the Australian National Wildlife Collection that investigate novel substrates for genomic analysis. Firstly, I will introduce a project focused on whole genome sequencing from formalin preserved specimens. I will explore the limitations of current technology and describe the novel triage methodology implemented to assess the likelihood of DNA extraction and subsequent sequencing success. Secondly I will introduce the Ovogenomics Project, focused on whole mitochondrial genome sequencing from modern avian eggshell collections. Avian eggshells are a bio-ceramic material, once thought to be devoid of DNA. However, maternal nuclei are incorporated into and sequestered within the shell. Here we utilise these unexpectedly optimal preservation conditions to sequence intra-crystalline DNA from Neognath eggshells. We also discuss the trade-off between collecting molecular data and destructive sampling of egg specimens. At ANWC alone, these two initiatives have mobilised over 40,000 new specimens for genomic analysis and have the potential to provide evolutionary insight over the 138 year collecting period.

GSA2018_APCC68498

Network analysis of signaling pathways reveals new mechanisms regulating the development and evolution of reproductive output in Drosophila

GSA: Development and cellular genetics

GSA2018 Abstract - Oral

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All developmental processes rely on a complex interplay of gene regulatory network (GRN) interactions. A major challenge is to determine not only which GRNs function in a given developmental process, but also how GRNs and their interactions are modulated over evolutionary time to generate morphological variations that impact fitness and thus are subject to natural selection. We are using the developing ovary of Drosophila as a system to address these challenges. The ovary comprises a number of egg-producing structures called ovarioles, and ovariole number is positively correlated with egg-laying, making ovariole number a trait that likely impacts fitness. Ovariole number is species-specific, highly heritable and also influenced by environmental factors including nutrition. We had previously identified the Hippo signaling pathway as a major regulator of ovariole number. Here we present the results of a genetic screen aimed at uncovering new Hippo-dependent and -independent modulators of reproductive capacity. We provide evidence that all major signaling pathways impact either ovariole number or egg-laying or both, and that some of them act synergistically with Hippo signaling to do so. A network analysis of the screen phenotypes yielded three major and novel outcomes. First, we find that egg-laying and ovariole number are regulated by non-overlapping genetic modules. Second, we predict the involvement of previously untested genes in ovariole number regulation. Finally, we distinguish between genetic modules that are likely to be major “on/off” switches for reproductive capacity, and modules that are more likely to provide the “fine tuning” dials through which evolutionary change generates ovariole number variation across species.

GSA2018_APCC648134

Newly Found Repetitive DNA in Four Araliaceae Tree Species

APCC6: Comparative, evolutionary and
population cytogenetics

APCC6 Abstract - Poster

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Araliaceae is also called as ginseng family, and the herbs in this family have high medicinal and economic value. Many studies show that repetitive DNA sequence makes up a big part of the whole genome size in plant species. Nevertheless, there is little information on repetitive sequence available in Araliaceae genome for its breeding and genomic research. Here, the cytogenetic analysis of four Araliaceae species, Aralia elata (2n = 24), Dendropanax morbifera (2n = 48), Eleutherococcus sessiliflorus (2n = 48), and Kalopanax septemlobus (2n = 48), will help us to understand the distribution of the newly found repetitive DNA sequence. Low-coverage next generation sequencing whole genome sequence of these four species was analyzed by using RepeatExplorer. A 160-bp satellite DNA (satDNA) was identified in all four species and a 156-bp satDNA was identified in E. sessiliflorus and K. septemlobus. Fluorescence in situ hybridization (FISH) technique was applied to confirm the distribution of 5S and 45S rDNAs, and newly found repeats on the mitotic chromosomes. FISH results showed that only one pair of signals of both rDNA families appeared in all four species. The 160-bp satDNA located at centromeric or interstitial or terminal region of all species except K. septemlobus, but the 156-bp satDNA located only in E. sessiliflorus at telomeric regions of some chromosomes. These results will be useful for the comparative cytogenomic and breeding studies among the species.

This work was carried out with the support of “Cooperative Research Program for Agriculture Science & Technology Development (PJ013119) Rural Development Administration, Republic of Korea”.

GSA2018_APCC613136

Newly Found Repetitive DNA in Four Araliaceae Tree Species

APCC6: Comparative, evolutionary and
population cytogenetics

APCC6 Abstract - Poster

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Araliaceae is also called as ginseng family, and the herbs in this family have high medicinal and economic value. Many studies show that repetitive DNA sequence makes up a big part of the whole genome size in plant species. Nevertheless, there is little information on repetitive sequence available in Araliaceae genome for its breeding and genomic research. Here, the cytogenetic analysis of four Araliaceae species, Aralia elata (2n = 24), Dendropanax morbifera (2n = 48), Eleutherococcus sessiliflorus (2n = 48), and Kalopanax septemlobus (2n = 48), will help us to understand the distribution of the newly found repetitive DNA sequence. Low-coverage next generation sequencing whole genome sequence of these four species was analyzed by using RepeatExplorer. A 160-bp satellite DNA (satDNA) was identified in all four species and a 156-bp satDNA was identified in E. sessiliflorus and K. septemlobus. Fluorescence in situ hybridization (FISH) technique was applied to confirm the distribution of 5S and 45S rDNAs, and newly found repeats on the mitotic chromosomes. FISH results showed that only one pair of signals of both rDNA families appeared in all four species. The 160-bp satDNA located at centromeric or interstitial or terminal region of all species except K. septemlobus, but the 156-bp satDNA located only in E. sessiliflorus at telomeric regions of some chromosomes. These results will be useful for the comparative cytogenomic and breeding studies among the species.
This work was carried out with the support of “Cooperative Research Program for Agriculture Science & Technology Development (PJ013119) Rural Development Administration, Republic of Korea”.

GSA2018_APCC64236

Novel families of centromeric satellite DNA in Asian swamp eel (Monopterus albus) related to a complex network with transposable elements

APCC6: Cytogenetics in the genomics era (Plants and Animals)

APCC6 Abstract - Oral

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Centromeres exhibit highly complex organization and are often flanked by satellite DNA (stDNA) related to their functional maintenance which includes cell division. Here, four novel centromeric stDNA families (MALREP-A, MALREP-B, MALREP-C and MALREP-D) were isolated from genomic DNA of the Asian swamp eel (Monopterus albus, 2n=24), considered to be the lowest chromosome number known in teleosts (2n=42–48), by construction of a genomic fosmid DNA library, characterized by in silico, fluorescence in situ hybridization (FISH) mapping, and filter hybridization. Respective lengths and GC contents of the sequences were 233 bp and 54.3% for MALREP-A, 233 bp and 53.2% for MALREP-B, 293 bp and 52.9% for MALREP-C, and 293 bp and 52.2% for MALREP-D. Principle component analysis showed the existence of several subfamilies in each stDNA family, suggesting large stDNA diversity in centromeric region. Three centromeric stDNA families (MALREP-A, B and D) were located on all chromosomes with different FISH signal intensities, except for MALREP-C localized to 11 pairs, suggesting that centromeric stDNA were separately homogenized among chromosomes. These stDNA families only hybridized with genomic DNA of Asian swamp eel, but no signal was observed even for other teleosts and vertebrates. Similar results, based on in silico mapping, were recorded for whole genome sequences of several vertebrates, suggesting that these stDNA sequences were differentiated rapidly or specifically amplified in the Asian swamp eel genome. Sequences homologous to all families of repeated sequences were searched in Repbase. Most stDNA families showed partial homology with GYPSY and MuDR retrotransposons. This suggests that these centromeric stDNAs were derived from retrotransposons which acted as multifunctional modular elements and essentially shared propagation mechanisms and synergy to shape the genome

GSA2018_APCC698194

Novel genotype of spotted-tailed quoll (Dasyurus maculatus) present on Kangaroo Island (South Australia) prior to extirpation

GSA: Ecological genetics

GSA2018 Abstract - Poster

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Spotted-tailed quolls (Dasyurus maculatus) occupied Kangaroo Island (KI), South Australia, for over 50,000 years but became locally extinct following European settlement of the island in 1836. As the top-order mammalian predator on KI when Europeans arrived, spotted-tailed quolls played a significant role in maintaining healthy ecosystem function. The reintroduction of spotted-tailed quolls to KI could return some of these ecological benefits, as welll as establish another refuge population of these animals that are under threat on the Australian mainland and Tasmania. Two subpecies of spotted-tailed quoll (mainland and Tasmanian) are recognised; however, the subspecies that inhabited KI in the past is currently unknown. While the extant subspecies are difficult to distinguish based on skeletal morphology, they are genetically distinct. Here, we extracted ancient DNA from five fossil specimens from Kelly Hill Cave (KI), morphologically identified as D. maculatus. Using next-generation sequencing, we confirmed the morphological identifications of the fossil specimens, and amplified a 450 bp region of the mitochondrial D-loop to determine the subspecific affiliation(s) of KI's D. maculatus, and therefore the subspecies that would be the most appropriate candidate for reintroduction of spotted-tailed quolls to KI. We find that all fossil specimens are most closely related to the Tasmanian subspecies, but form a distinct monophyletic clade. As this clade may prove to be a new subspecies with further research, we advocate that any reintroduction of D. maculatus to KI seeks to establish a genetically diverse comparative population, possibly sourcing individuals from both the mainland and Tasmania.

GSA2018_APCC687197

Novel splicing mechanisms to immunise intronic transposon insertions in Drosophila

GSA: Genetics and medicine

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A large proportion of eukaryotic genomes are comprised of repetitive sequences such as Transposable elements, transposons in short. Uncontrolled propagation of transposons causes catastrophic DNA damage on one hand, occasional new transposon insertions provide additional regulatory elements for the host gene expression. From the genetic screen for factors involved in transposon silencing in Drosophila germline, we identified factors that help the splicing of piwi gene, the central gene in the piRNA pathway in Drosophila. RNA sequencing revealed that there are two independent mechanisms that facilitate the splicing of two different introns of piwi. Interestingly, both introns contain evolutionarily recent transposon insertions, which likely made the splicing of those introns difficult. We found an evolutionary arms race where transposons harm the host defence pathway while the host immunises transposon insertions by aiding the splicing of suboptimal introns.

GSA2018_APCC64094

Physical Chromosome Mapping of repetitive DNA sequences in Bienertia sinuspersici using Fluorescence in situ hybridization

APCC6: Cytogenetics in the genomics era (Plants and Animals)

APCC6 Abstract - Poster

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A number of important crops are C4 plants that could produce more sugar than C3 plant in conditions of high light and temperature. Bienertia sinuspersici (2n =2x=18) is one of the few plant species that could perform SSC4 photosynthesis without having a typical Kranz anatomy in its leaf. A cytogenetic mapping is carried out using two satellite and two unknown DNA sequences through FISH technique. A satellite DNA sequences (rnd-6-1241-3+rnd-6-1241-2) with 814 copies have hybridized in the interstitial and centromeric regions of nine chromosomes. Another type of satellite DNA (rnd-3-39-1; 21,480 copies) are dispersedly distributed in the interstitial

regions of 18 chromosomes. Unknown DNA sequences (rnd-3-22-2-1; 54,943 copies) have the same pattern of signal distribution with the previous one. Lastly, another unknown DNA sequences have entirely hybridized the regions of all chromosomes (rnd-3-22-2-1; 61,450 copies). A symmetrical karyotype was observed with chromosome length that ranges from 9.21 µL to 13.15 µL. The gathered data will be utilized to understanding the genetic constitution of this species. (This work was supported by the Cooperative Research Program for Agriculture Science & Technology Development (project No. PJ01095307) of the Rural Development Administration Republic of Korea)

GSA2018_APCC62733

Predicting Shannon’s information for genes in finite populations – including conservation applications

GSA: Ecological genetics

GSA2018 Abstract - Oral

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This study provides predictive equations for genetic Shannon’s information in a finite population, which are intuitive and simple enough to see wide scale use in molecular ecology and conservation biology. A comprehensive profile of genetic diversity contains three complementary components: numbers of allelic types, Shannon's information and heterozygosity. Currently heterozygosity has greater resources than Shannon’s information, such as more predictive models and integration into more mainstream genetics software. However, Shannon's has several advantages over heterozygosity as a measure of genetic diversity, so it is important to develop Shannon's information as a new tool for molecular ecology. Past efforts have dealt with expectations for Shannon’s at genetic equilibrium, but dynamic forecasts are also vital. In particular, we must be able to predict loss of genetic diversity when dealing with finite populations, because they risk losing genetic variability, which can have an adverse effect on their survival. We present equations for predicting loss of genetic diversity measured by Shannon's information. We also provide statistical justification for these models by assessing their fit to data derived from simulations and then applying those equations on live drosophila populations. The predictive models will enhance the usefulness of Shannon's information as a measure of genetic diversity; they will also find practical applications in pest control and conservation.

GSA2018_APCC61886

Quantifying the effects of sex-biased transmission on local adaptation in Drosophila

GSA: Genetics of quantitative traits

GSA2018 Abstract - Oral

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Recent theory on the genetic basis of local adaptation predicts that X-linked genes should nearly always make a larger contribution to local adaptation than autosomal genes. To test this prediction, we expanded upon a widely-used reciprocal cross method for estimating the influence of the X-chromosome on phenotypic divergence between populations (captured by the X-linked divergence metric, IX, from Reinhold 1998). In the present study, we developed indices for estimating the contributions of the X-chromosome (IX), the autosomes (IA), and cytoplasmic and Y-linked genes (ICY) to phenotypic divergence between populations. We used reciprocal crosses from two geographically distinct populations of Drosophila melanogaster collected from along a latitudinal gradient to generate F1 and F2 males that were then phenotyped for a series of four ecologically important and clinally variable traits: heat resistance, desiccation and starvation resistance, and wing size (a proxy for body size). In contrast to the theoretical predictions, we find that divergence is dominated by autosomal genes, and genes with uniparental inheritance (e.g., mitochondria, Y). We discuss how attributes of genome architecture and sex-biased demography may contribute to these results.

GSA2018_APCC629168

Recombination between genes with fitness interactions – how much is enough? Empirical data and simulations

GSA: Ecological genetics

GSA2018 Abstract - Oral

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Many of the effects on fitness population genetics is concerned with are due not to single locations in the genome, but due to the interaction of genetic variants at multiple locations in the genome. Of particular interest are 'epistatic' interactions, where a particular combination of genetic variants is required to produce an effect, and the effect cannot occur with any other combination. In diploids, models of these effects are difficult to analyse mathematically, as they are strongly influenced by meiotic recombination, a process which can both assemble and destroy combinations of genetic variants. Additionally, these interactions can be hard to detect in empirical studies, and so are regularly not considered. As a result, there is little consensus on when high levels of recombination might be expected, or how strongly recombination affects beneficial or deleterious fitness effects controlled by epistatic interactions. We address this question by conducting a meta-analysis, using data drawn and curated from Drosophila melanogaster studies in FlyBase. We developed a protocol for the extraction of studies relating genetic combinations and phenotypically detectable effects on fitness. Using the extracted data, we analysed the relationship between the rate of recombination and effect on fitness with a statistical model. We also ran simulations under a two-locus Wright-Fisher model with recombination and epistatic selection. The results of both approaches indicated a tendency for genetic combinations with some effect on fitness to have reduced meiotic recombination. Two possible explanations for this are that the variants controlling such interactions may only be retained in the population if they appear at locations with a low rate of recombination between them, or that such interactions lead to selection for lower rates of recombination.

GSA2018_APCC67831

Reconstruction of female heterogametic sex determination from admixture of female and male heterogametic systems in a frog

APCC6: Specialised chromosomes (sex chromosomes and B chromosomes)

APCC6 Abstract - Oral

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The Japanese soil-frog, Glandirana rugosa, consists of multiple populations having different sex-determining mechanisms, which are XX-XY and ZZ-ZW types. Both the XY and ZW sex chromosomes are 7th largest in the complement (2n=26) and are morphologically differentiated between the homologues. The geographic populations with XY sex chromosomes are distributed in central Japan (Tokai and Kinki districts), while those with ZW sex chromosomes are classified into two different groups: one is distributed in western regions of northern Japan (Tohoku district), called ZW population, while the other is in central Japan (Kinki district), called Neo-ZW. In the central Japan (Kinki district), the two different populations, Neo-ZW in the west and XY population in the east, are panmictic and, in fact, are intermingled with each other where heterogametic males and females are found. Recently, we have found that the most eastern populations of the Neo-ZW, which are very close to the intermingling populations, shared the same mitochondrial haplotypes with XY populations, and thus it was designated Neo-ZW2 population to be differentiated from the original population, Neo-ZW1. Then, to uncover the evolutionary mechanisms of Neo-ZW2 populations, we investigated constitution of the nuclear genomes and sex-linked genes by SNPs analysis. Based on the results, we concluded that the Neo-ZW2 populations originated from hybridization between the Neo-ZW1 and XY populations in the past, and interestingly the Z chromosomes originated from the Neo-ZW1 while W chromosomes originated from X chromosomes of the XY population. The convergent evolution of female heterogamety is discussed based on the artificial crossing experiments between the Neo-ZW1 and XY populations at laboratory.

GSA2018_APCC68989

Regulation of epigenetic silencing and chromatin architecture by the noncanonical SMC protein SMCHD1

GSA: Genetics and epigenetic regulation

GSA2018 Abstract - Oral

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My primary interest is in understanding the molecular mechanisms of epigenetic silencing. For this talk I'll focus on the epigenetic regulator SMCHD1, which we have previously shown has a critical role in developmental epigenetic silencing of the inactive X chromosome and clustered genes on autosomes, for example imprinted genes. Recent work from us and others also suggests SMCHD1 is a potential drug target in disease. This has been revealed due to the heterozygous SMCHD1 mutations found in two different diseases: facioscapulohumeral muscular dystrophy (FSHD) vs a rare congenital craniofacial development disorder, bosma arhinia and micropthalmia (BAMS). There is much controversy about how these mutations alter SMCHD1 function. I will present our biochemical studies related to these mutations, which suggest that BAMS results from gain of function mutations in SMCHD1, while FSHD is strictly associated with loss of function mutations. I will then present our functional genomic studies on how SMCHD1 elicits gene silencing and how it is targeted to its genomic binding sites. Our studies reveal for the first time that SMCHD1 is a novel regulator of long-range chromatin interactions, and add it to the canon of proteins required for Hox gene silencing. Furthermore, they suggest a model where Smchd1 brings about transcriptional silencing by insulating the chromatin, limiting access to other chromatin modifying proteins. Together these studies expand our understanding of how SMCHD1 functions in epigenetic control in normal development and disease.

GSA2018_APCC616161

River Structure and Scale as Determinants of Population Structure in the Eastern Basins of Australia

GSA: Bioinformatics and genomics

GSA2018 Abstract - Poster

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After some 20 years of river structure theoretical and field studies in genetics, their structure and scale effects are poorly understood for Australia. Some field observations conflict with theory developed from and explored by modelling. In this poster we illustrate current work using extensive genomic SNP datasets (DaRTSeq) from shrimp, turtle, smelt and yabby sampling, along the main Eastern river systems. Our research is aimed at examining the relationship between aquatic species' genetic and riverine distance measures, and the relative importance of river network topologies (linear vs dendritic) in shaping these. The results of testing theoretical predictions of constrained gene flow in dendritic systems, and increased among-site diversity among headwater populations in different taxa and basins are shown and discussed.

GSA2018_APCC688123

Sex reversal and sex-determining mechanism in an oviparous skink (Bassiana duperreyi): a lizard with XX:XY sex chromosomes and sex reversal at cold incubation temperatures

GSA: Ecological genetics

GSA2018 Abstract - Poster

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Sex of most species in animal kingdom has classically been considered to be determined via one of two mechanisms: genotypic sex determination (sex determined at the time fertilization by genetic factors which independent of environmental influence) or environmental sex determination (sex determined by environmental factors that act after fertilization). However, in Bassiana duperreyi and Pogona vitticeps, genetic sex determination can be over-ridden by extreme temperatures during incubation resulting in sex reversal. Altered climate regimes are increasingly being considered one of the major threats for reptiles, especially for species with temperature-dependent sex determination, possibly leading to population declines and extinctions. The aim of this project is to determine the ecological and evolutionary implications of sex reversal in the wild. Therefore, putative Y-chromosome markers will be identified using subtractive genomics. We will then use these markers to assess the rate of sex reversal across a high altitude to low altitude gradient from the Australian Alps to the Victorian coast to determine the rate of sex reversal in nests and among adults.This project will address our knowledge gap on understanding the ecological consequences of global warming on reptiles and our new data will stimulate the development of predictive models that could help predict the effect on global warming on reptile populations in Australia on reptile sex ratios. In addition, this project will serve as a model for species at risk of extinction from climate change to focus on future research to test on-the-ground management strategies to mitigate the effects of climate in local populations.

GSA2018_APCC66132

Sex reversal in labrid fishes involves novel sex genes and developmental regression through a pluripotent-like state

GSA: Genetics and epigenetic regulation

GSA2018 Abstract - Oral

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Fishes exhibit remarkably diverse and plastic sexual development. This includes functional adult sex change, which has evolved repeatedly and appears in 27 teleost families. While the sex change process and its evolutionary advantages are well known, how environmental cues drive such dramatic changes in sexual identity, and the molecular processes involved, have been longstanding questions. Female-male sex change (protogyny) is common in the wrasses (Labridae), where individuals reproduce first as females, but routinely reverse sex in the absence of a socially dominant male. This process involves complete gonadal restructuring where no identifiable male tissues exist in the gonad prior to sex change. We used social manipulations to generate a sex change time-series in each of two

distantly-related wrasses - the Caribbean bluehead (Thalassoma bifasciatum) and New Zealand spotty (Notolabrus celiodotus) - and applied transcriptomic and whole-methylome approaches to identify the primary trigger and subsequent molecular cascade involved in gonad remodelling. We show that redirection of sexual fate involves extensive transcriptional and epigenetic reprogramming of gonadal cells. Our data suggest that the environmental stimulus may involve stress, that repression of the aromatase gene (encodes the enzyme that converts androgens to estrogens) triggers a cascaded collapse of a feminising expression network, and that neofunctionalisation of duplicated sex-pathway genes may underpin the notable sexual plasticity of teleost fish. Significantly, we find that sex change involves specific epigenetic reprogramming consistent with developmental regression through a pluripotent-like state. As the first genome-scale investigation of natural sex change in any vertebrate, our results have important implications for understanding the evolution and functioning of vertebrate sex determination and developmental systems. Our data also underscore the role of epigenetics and pluripotency in sex determination, and offer a unique perspective on the relationship between DNA methylation, pluripotency and developmental capacity in non-mammalian vertebrates.

GSA2018_APCC626115

Sex reversal is a geographically and temporally widespread phenomenon in the central bearded dragon

GSA: Ecological genetics

GSA2018 Abstract - Oral

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The central bearded dragon (Pogona vitticeps) displays ZZ/ZW genetic sex determination, but incubation temperatures greater than 32˚C can cause sex reversal, in which ZZ genotypic males develop as phenotypic females. The species is widespread, spanning temperate to arid climatic regions across Australia. If sex in this species can be affected by environmental conditions, questions therefore arise as to where and how often sex reversal occurs in wild populations, and whether these instances correlate with climatic region or changing trends in climate. Here, we report a substantial increase in the geographic and temporal sampling of sex in this species, including the identification of several sex-reversed females in previously unstudied regions. The genotypic sexing of over 100 P. vitticeps tissues taken from specimens collected and stored in the South Australian Museum and Australian Biological Tissue Collection revealed that sex reversal does occur in South Australian populations, in addition to previously reported occurrences of sex reversal in a well-studied population on the border of QLD-NSW. This expansion in the spatial and temporal record of sex reversal will vastly improve our ability to develop predictive and explanatory models of landscape-scale sex dynamics in this species, shedding light on the capacity for changes in climate to influence sex determination modes in this thermally sensitive reptile.

GSA2018_APCC6921

Silene latifolia - model for the sex chromosome evolution in plants

APCC6: Specialised chromosomes (sex chromosomes and B chromosomes)

APCC6 Abstract - Oral

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Silene latifolia (white campion) is a dioecious species possessing heteromorphic sex chromosomes, heterogametic in males (24, XY) and homogametic in females (24, XX). Due to their relatively recent origin and heteromorphic nature, the sex chromosomes in white campion are extensively studied. Although many aspects of sex chromosome evolution in S. latifolia were already described, the sex determining region, genes involved in sex determination and potential epigenetic control of sex expression are still poorly understood. We have found, using genomic and cytogenetic techniques, that various DNA repeats (DNA transposons, retroelements and satellites) are not only specifically distributed on the Y, but also significantly affect the X chromosome structure. These results indicate that some DNA repeats (namely Ogre and Athila elements) are under sex specific control. In addition, immunostaining, using antibodies against active (e.g., H3K9ac, H3K4me) and inactive histone modifications (e.g., H3K9me, H3K27me), allowed us to differentiate gene rich regions and to decipher the epigenetic state of the X and Y chromosomes. Finally, transcriptome profiling of hermaphroditic individuals, induced by epigenetic drug treatment, enabled us to identify candidate genes for sex expression. These genes are now studied by reverse genetic approaches (e.g., by CRISPR/Cas9 nucleases) to confirm their function in sex determination. This work was supported by the Czech Science Foundation project No. P501/12/G090.

GSA2018_APCC63223

Sister chromosome pairing leads to clonal gametes in dojo loach, a teleost fish

APCC6: Comparative, evolutionary and population cytogenetics

APCC6 Abstract - Oral

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Dojo loach, Misgurnus anguillicaudatus (2n = 50), is a freshwater fish distributed in East Asia. There are two genetically-diverse groups A and B within a single species and they reproduce bisexually. However, unisexual lineages with clonal genotypes reproduce by gynogenesis in certain localities of Japan. They are considered to originate from past hybridization event between the two groups, but mechanisms responsible for clonal gametogenesis have not been disclosed yet. Here, we succeeded to distinguish chromosomes derived from each group by fluorescence in situ hybridization (FISH) with group-B specific probe and 5.8S + 28S rDNA probe. Then, hybrid origin of the clone was clearly demonstrated both in somatic as well as meiotic cells by FISH. In addition, we detected chromosomes originated from the two different groups in diploid sperm of sex-reversed clonal males. Thus, we proved that clonal gametes should be produced by a mechanism of premeiotic endomitosis, pairing between sister chromosomes formed by doubling of ancestral chromosome from each group before entering into meiosis. Sister chromosome pairing assures production of unreduced isogenic clonal gametes because sister chromosomes behave as homologous chromosomes and recombination occurs between identical elements of sister chromosomes. Our results give insights into the mechanisms underlying in alterations to meiosis for atypical reproduction, as well as hybrid origin of clonal vertebrate.

GSA2018_APCC65217

Spatial organization of three chromosome territories in the trisomic cell nucleus

APCC6: Chromosome architecture,
epigenetics and cell division

APCC6 Abstract - Poster

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Spatial positioning of chromosome territories (CTs) has been organized under specific manners, such as gene densities and physical sizes of each CT for the radial distribution in the cell nucleus. In this study, we focused on trisomic cells, which possess three homologous chromosomes as numerical abnormality compared with normal disomic cells possessing two homologous chromosomes. What kind of different spatial arrangements of the homologous CTs will be observed in the trisomic cell nucleus? Using human lymphoblastoid cell lines from Down syndrome persons with the standard typed trisomy 21 as well as human fibroblast cell line MK-7 possessing trisomy 13, we performed 3D-FISH analysis with the human chromosome 21 and 13 long arm-specific painting probes, respectively. After scanning images by the confocal microscopes, image stacks were utilized for measurement of the relative localization of three CTs. The results showed that in 21 trisomic cells two 21 CTs out of three CTs tended to be placed close to each other. These adjacent pair of 21 CTs would be derived from maternal origins and single apart CT would be derived from paternal one according to Omori et al (2017). In contrast, in 13 trisomic cells, three 13 CTs were uniformly distributed within the cell nucleus, and there were a higher tendency not to form the specific spatial adjacent pair of 13 CTs. However, significant decondensation was frequently observed in the single 13 CT out of three CTs, probably reflecting the status of gene expression of this CT. There was a difference in trisomy 21 and trisomy 13 cells suggesting that this may be affected not only by the difference between chromosomes 21 and 13 but also by the combination of parents derived from each CT in individual cell lines. Further studies would clarify the features of the spatial organization of CTs.

GSA2018_APCC689127

Structure and origin of the calcareous sponge microbiome

GSA: Ecological genetics

GSA2018 Abstract - Oral

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Sponges (Porifera) are likely the most ancient animal lineage, and therefore provide a unique insight into the evolutionary history of animal features, such as embryonic development, regeneration and immunity. Because of their well-known associations with microbes, sponges are also particularly well suited as models to study host-microbial interactions. The microbial component of an organism, known as the microbiome, has been found to play a crucial role in the biology of sponges. Moreover, the microbial community within sponges has been fruitfully mined for bioactive compounds, such as the toxic arenosclerin alkaloids. However, the majority of these studies involved demosponges, with the microbiomes of the remaining lineages, especially calcareous sponges, remaining largely unknown. Further, the mode of transmission of the sponge microbiome and its role in sponge development are not well understood. Taking advantage of multiple calcareous sponge genomes sequenced by our group for developmental genomics studies, we have set out to characterise the eukaryotic and prokaryotic microbial composition of several species of calcareous sponges, collected from the Norwegian fjords, and the eastern Australian coast. In particular, we are comparing microbiomes of closely related host species living in geographically distant locations as well as distantly related species living side-by-side. In order to determine the extent of vertical transmission of the microbiome, we are also comparing the calcareous sponge microbiomes between larval, juvenile and adult developmental stages. This study lays the foundations for defining the composition, structure and origin of the calcareous sponge microbiome.

GSA2018_APCC617191

Targeting the ribosomal RNA gene (rDNA) loci to treat cancer

GSA: Genetics and medicine

GSA2018 Abstract - Oral

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The role dysregulated transcription from the ribosomal RNA (rRNA) gene (rDNA) loci in eliciting a nucleolar stress response (NSR), which is heightened in cancer cells, is emerging as an exciting and significant area of cancer research. In the canonical NSR, disruptions to ribosome biogenesis (RiBi) result in p53 accumulation, as free ribosomal proteins (RPs) can bind and sequester MDM2, the ubiquitin ligase that targets p53 for degradation. In addition, though high throughput functional genome wide screening approaches we have uncovered a novel, atypical p53-independent NSR, associated with activation of DNA damage checkpoints, which has excellent potential for treating p53-null tumours. The validity of Pol I as a therapeutic target has been demonstrated by the selective Pol I transcriptional inhibitor, CX-5461which has entered clinical trials in Australia and Canada. Our mechanistic studies have demonstrated that CX-5461 kills tumour cells, in part, by triggering an acute NSR. Importantly, while drugs selectively targeting rDNA transcription to treat cancer are new, retrospective analyses of the targets for various chemotherapies (eg. classic platinum-based drugs) suggest that their efficacy is mediated in part through targeting rDNA transcription and the NSR. Despite the overwhelming evidence for dysregulated RiBi in cancer, only 1 selective Pol I inhibitor, CX5461, has entered clinical trials. Although extremely promising, our investigation into mechanism of action has revealed additional activities of CX5461, that may not only contribute to its efficacy but also to the toxicity profile and mechanism of acquired resistance. To address the need for new, improved Pol I inhibitors we have developed a series of orally available selective inhibitors with improved toxicology, tissue distribution and efficacy compared to CX-5461. Preliminary studies on our new lead compound, PMR-116, which has demonstrated improved survival (at MTD) will be discussed along with our studies on mechanism(s) which confer sensitivity and resistance to Pol I inhibition therapy.

GSA2018_APCC67099

Testing the Integrative Breakage Model: A multidisciplinary approach for the study of genome plasticity

APCC6: Chromosome architecture,
epigenetics and cell division

APCC6 Abstract - Oral

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Normal 0 21 false false false ES-TRAD JA X-NONE /* Style Definitions */ table.MsoNormalTable {mso-style-name:"Taula normal"; mso-tstyle-rowband-size:0; mso-tstyle-colband-size:0; mso-style-noshow:yes; mso-style-priority:99; mso-style-parent:""; mso-padding-alt:0cm 5.4pt 0cm 5.4pt; mso-para-margin:0cm; mso-para-margin-bottom:.0001pt; mso-pagination:widow-orphan; font-size:12.0pt; font-family:"Cambria","serif"; mso-ascii-font-family:Cambria; mso-ascii-theme-font:minor-latin; mso-hansi-font-family:Cambria; mso-hansi-theme-font:minor-latin; mso-ansi-language:ES-TRAD;}Unlocking the genomic basis of speciation is a research priority in biology fuelled by the ongoing debate on species concepts and facilitated by the availability of an unprecedented large number of genomic resources. Through comparative genomics of both closely and distantly related mammalian species our research group, along with others, has contributed to models that explain genome structure and evolution. Such reconstructions have revealed that the genomic regions implicated in structural evolutionary changes, disrupt genomic synteny, and are clustered in regions more prone to break and reorganize. In searching for the origin (and consequences) of this evolutionary instability, we provided insights on the genomic features that characterize evolutionary regions. In addition, we have also observed that changes in gene expression that are caused by genome reshuffling may have a selective advantage through the development of new adaptive characters specific to mammalian lineages. Consequently, given the diversity of factors associated with genome reshuffling it is most unlikely that the sequence composition of genomes is solely responsible for genomic instability during evolution. This view represents a new interpretative evolutionary hypothesis that has recently been unified by our research group as the ‘Integrative Breakage Model’, which postulates that the permissiveness of some genomic regions to undergo chromosomal breakage and genomic rearrangements could be influenced by chromatin conformation. Here we will provide evidence that suggest that certain properties of local DNA sequences, together with the epigenetic state of the chromatin, are effecting gene expression and are key elements in determining the genomic distribution of evolutionary breakpoints during the cell cycle in mammals.

GSA2018_APCC65064

Testing turnover: could coastal earthquake uplift provide opportunities for genetic change?

GSA: Ecological genetics

GSA2018 Abstract - Oral

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Recent research into processes that shape patterns of genetic diversity has revealed that dispersal among populations, even when frequent, often fails to result in gene flow, leading to distinct geographic structure. The failure of dispersal to facilitate gene-flow in such cases is presumed to be the result of ‘density-blocking’ or ‘Founder Takes All’ processes, whereby early immigrants densely colonise an area and inhibit (block) the establishment of later arrivals. The release of density-blocking should be achievable, however, through large-scale disturbance events, where original lineages are completely removed and new, immigrant lineages could gain a foot-hold. The November 2016 earthquakes in Kaikoura, New Zealand, resulted in massive coastal uplift that completely extirpated some existing, intertidal kelp populations, and partially destroyed others. This event has provided a unique opportunity to test the role of disturbance events in releasing density-blocking processes and enabling genetic turnover. Using genotyping by sequencing (GBS), we will pinpoint the precise source populations of kelp rafts reaching the uplifted area in the 1.5 years since the earthquakes. We hypothesise that a significant proportion of these potential colonisers will differ, genetically, from the previous lineages occupying the area, with drift kelp arriving from genetically-distinct populations along the entire east coast of the South Island, and from thousands of kilometres away in the sub-Antarctic. In the absence of established populations, any of these new lineages could, we predict, become the dominant inhabitants of the new coastline, resulting in genetic turnover. This research will help us to understand the importance of disturbance on the success of dispersal and on the development of spatial patterns of genetic structure.

GSA2018_APCC61025

The "Trojan Female Technique", a novel pest control method, combined with CRISPR/Cas9 to tackle current control limitations

GSA: Ecological genetics

GSA2018 Abstract - Oral

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The “Trojan Female Technique” (TFT), where females, carrying naturally occurring male sterility mitogenome haplotypes, are introduced into pest populations, is a promising new approach for biocontrol. “Trojan” females reproduce normally, but, the fertility of their male offspring is compromised, leading to a reduction in population growth and persistence over time (Gemmell et al. 2013). Recently, we demonstrated the suppressive effects of mtDNA haplotypes known to affect male, but not female, reproductive success in the common fruit fly, Drosophila melanogaster (Wolff et al. 2017). Population growth was reduced by introduction of the Trojan females, and this persisted over 10 generations with no reduction in TFT haplotype frequency. These results are an encouraging proof-of-concept for TFT as a eukaryotic pest control strategy. While promising, a major current limitation of the TFT approach is its dependence on naturally occurring mutations, so we are now exploring the possibility of introducing known TFT variants using gene editing approaches. One study has to date reported the use of the Crispr/cas9 transgenesis system to target mtDNA in HEK293 cell cultures. If this can be adapted to modify mtDNA in other systems it might provide a framework to introduce the suppressive mtDNA haplotypes into a limitless range of pest species while still retaining the species specificity of control. Currently, I am testing mtDNA gene editing in Drosophila melanogaster using the Crisp/cas9 system. Here I will talk on the special challenges associated with the gene editing of mitochondrial DNA including the issues of importation, copy number, and measuring the efficiency of edits.

GSA2018_APCC648181

The dihaploid genome architecture of a plant fungal pathogen shapes host adaptation

GSA: Bioinformatics and genomics

GSA2018 Abstract - Poster

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A long-standing biological question is how evolution has shaped the genomic architecture of dikaryotic plant pathogenic fungi. To answer this, high quality genomic resources that enable haplotype comparisons are essential. Short-read genome assemblies for dikaryotic fungi are highly fragmented and lack haplotype-specific information due to the high heterozygosity and repeat content of these genomes. Here we present a diploid-aware assembly of the wheat stripe rust fungus Puccinia striiformis f. sp. tritici based on long-reads using the FALCON-Unzip assembler. RNA-seq datasets were used to infer high quality gene models and identify virulence genes involved in plant infection referred to as effectors. This represents the most complete Puccinia striiformis f. sp. tritici genome assembly to date (83 Mb, 156 contigs, N50 1.5 Mb) and provides phased haplotype information for over 92% of the genome. Comparisons of the phase blocks revealed high inter-haploid diversity of over 6%. More than 25% of all genes lack a clear allelic counterpart. When investigating genome features that potentially promote the rapid evolution of virulence, we found that candidate effector genes are spatially associated with conserved genes commonly found in basidiomycetes. Yet candidate effectors that lack an allelic counterpart are more distant from conserved genes than allelic candidate effectors, and are less likely to be evolutionarily conserved within the P. striiformis species complex and Pucciniales. In summary, this haplotype-phased assembly enabled us to discover novel genome features of a dikaryotic plant pathogenic fungus previously hidden in collapsed and fragmented genome assemblies. In addition, it will enable the community to identify to date unknown effector proteins that are recognized by the plant immune system.

GSA2018_APCC682126

The evolution of flightless insects: natural selection underpins parallel clinal wing reduction in alpine stoneflies

GSA: Ecological genetics

GSA2018 Abstract - Oral

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Alpine insect biotas are frequently characterised by large numbers of wing-reduced species, but the ecological and evolutionary bases of this reductive evolution are poorly understood. We conduct phenotypic and genetic analyses of a wing-dimorphic New Zealand stonefly species to reveal increasing selection for a vestigial-winged phenotype with increasing altitude. Specifically, altitudinal transect sampling from two South Island streams reveals parallel increases in genotypic frequencies at a locus previously found to be associated with wing reduction in the Zelandoperla fenestrata complex. Strong clinal selection at this locus contrasts with genome-wide data from genotyping-by-sequencing. The concordant, steep clines observed at altitudes around 600-700 m in two neighbouring but phylogenetically-independent populations suggest that environmental gradients associated with exposure underpin this genetic and phenotypic diversity. Importantly, this study yields a highly novel example of ‘divergence with gene flow’ acting over fine spatial scales, illustrating that natural selection at one or a few genetic loci can potentially underpin dramatic cases of local adaptation in wild populations.

GSA2018_APCC69243

The FUBP1 Drosophila ortholog Psi interacts with the Mediator Complex to modulate transcription of MYC and regulate growth

GSA: Development and cellular genetics

GSA2018 Abstract - Oral

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Mammalian FUBP-family proteins play roles in both RNA processing and transcription, by binding single stranded nucleic acids via their KH domains. In Drosophila, the three mammalian FUBP proteins are represented by one ortholog, Psi. We have taken advantage of this reduced functional redundancy to demonstrate that an essential in vivo function of Psi is control of cell and tissue growth. Analysis of published Co-IP-mass spectrometry screens positioned Psi in an interactome predominantly comprised of RNA Polymerase II transcriptional machinery. Of great interest was the interaction with most subunits of the transcriptional Mediator (MED) complex, a known sensor of developmental signaling inputs. Moreover, manipulation of MED activity modified Psi-dependent growth, which suggests that Psi interacts with the MED complex to integrate developmental growth signals with transcription of growth regulators. Our work indicates that a key target of the Psi/MED network that impacts tissue growth is the MYC transcription factor. Further to this, transcriptome-wide expression analysis comparing Psi and MYC depleted wing imaginal discs suggests that in addition to MYC-dependent targets, Psi regulates expression of RNA processing machinery. Our current studies are directed towards identifying direct transcriptional targets of Psi.

GSA2018_APCC694151

The murky truth about metabarcoding data: how a lack of sufficiently stringent QA/QC protocols is impacting metabarcoding research.

GSA: Ecological genetics

GSA2018 Abstract - Oral

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Metabarcoding can be broadly defined as the identification of multiple species via the high-throughput amplicon sequencing of environmental samples. This approach has been shown to be particularly powerful in studies examining complex assemblages of small and often cryptic organisms (e.g. microbes), as well as scenarios where samples may be degraded, with their constituents difficult to identify by optical means (e.g. faeces and gut-contents). While still a relatively new field of research, metabarcoding has not only transformed the way we obtain ecological data, but more importantly, our fundamental understanding of the distributions of taxa and their interactions with the environment. However, metabarcoding data is inherently noisy, with artefacts being produced in each step from sample collection to data processing. Collectively, these have the potential to substantially alter the data which underpins a study's findings. Here, I present a meta-analysis of the metabarcoding literature focusing on a number of QA/QC protocols pivotal for ensuring the reliability and transparency of data. The resulting evidence demonstrates a lack of adequate QA/QC implementation and/or reporting across the field, indicating that the findings of many studies may have been derived from data of unknown quality. While it is undoubtedly easy to pass judgement on a field that is still in its infancy, here I argue that in order for metabarcoding research to progress and to ensure it produces robust and reproducible findings, there is a critical need for a step-change in the way we monitor and report the quality of data.

GSA2018_APCC614150

The National Centre for Indigenous Genomics (NCIG): a community resource for research

GSA: Genetics and medicine

GSA2018 Abstract - Oral

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Research projects that develop perpetual community resources to support research, such as HapMap, UK Biobank and IGSR, play an increasingly important role in biomedicine. NCIG is establishing a resource of genome sequence data and related samples and records from Indigenous Australians. It is one of many community resource projects around the world that aim to ensure broad representation of the world's populations to ensure that the benefits of genomics are extended to all humans, irrespective of their ancestry. In the current era of "genomical" scale data and the availability of advanced machine learning techniques, resources such as NCIG needs to be appropriately managed to extract maximum value for all stakeholders. Towards that end, we are creating data repository in partnership with Nation Computational Infrastructure (NCI). This dynamic data repository integrates with biospecimen repository and in the future to be integrated with NCI's computational resources. NCIG, through this repository, will provides the essential link between Indigenous communities and research communities that is needed for sustainable benefit and capability development in both. Our data repository will ensure that the NCIG Collection is managed in line with governing principles and policies established by the Governance Board. Strong Indigenous governance ensures excellence in standards required for research involving Indigenous people and their involvement throughout the research life cycle. Enduring community engagement and partnerships that ensure a persitent connection between communities and the collection is at the heart of NCIG's operations.

GSA2018_APCC678165

The origin of a novel complex trait: butterfly eyespots

GSA: Development and cellular genetics

GSA2018 Abstract - Oral

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An important and still largely unanswered question in the field of evo-devo concerns the molecular changes that lead to the origin of novel complex traits. One such trait is the eyespot color patterns on the wings of nymphalid butterflies. My lab is combining a series of evo-devo approaches to identify the molecular and developmental changes underlying eyespot origins. A multi-species comparative phylogenetic approach led to the identification of the ancestral lineage where eyespots likely evolved, and also where four genes started becoming expressed in eyespot centers. A CRISPR/Cas9 gene editing approach identified three genes required for eyespot development that when mutated lead to wings without eyespots. Two of the genes evolved their new expression pattern concurrently with eyespot origins whereas the third gene evolved its expression at a later date in a subset of butterfly lineages. This means that genes that become associated with the development of a novel trait at a late stage in their evolution can become essential developmental genes. In addition, a combination of in situ gene expression with modeling work, led to reconstructions of mosaic mutant wing patterns and to the identity of two potential ligands involved in eyespot center differentiation via simple reaction-diffusion mechanisms. FAIRE-Seq further identified regulatory sequences flanking one of the genes, Distal-less, which is potentially involved in this reaction-diffusion network. When these regulatory sequences were deleted they led to eyespot deletions as well as disruptions of multiple other traits in the butterfly, indicating regulatory sequences with pleiotropic functions. This suggests that eyespots may have originated from co-opted limbs.

GSA2018_APCC670121

The Oz Mammals Genomics initiative: mammal genomics, evolution and conservation at a continental scale

GSA: Evolutionary genetics and
comparative genomics

GSA2018 Abstract - Oral

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The Australo-Papuan region has a unique mammal fauna, which faces unique threats and poses important evolutionary and ecological questions. Genomics has great potential to advance our understanding of the region's mammals and their conservation. The Oz Mammals Genomics Initiative brings together museum collections, researchers, data specialists and wildlife management agencies to comprehensively tackle rodent, bat and marsupial genomics at a continental scale. There are few published whole genomes for Australian marsupials. We are developing well-assembled genomes from a broadly representative range of marsupial taxa, to facilitate new insights into evolution and to provide reference data for conservation studies. Genome projects are now underway for seven priority species: the fat-tailed dunnart, brush-tailed rock-wallaby, eastern bettong, mountain pygmy possum, Leadbeater's possum, bare-nosed wombat and eastern barred bandicoot. Our current understanding of evolutionary relationships among many mammal taxa remains incomplete. To improve resolution of genus and species boundaries, we are generating comprehensive phylogenies of all extant and recently-extinct terrestrial mammals native to the Australo-Papuan region. We are using exon capture methods to sequence over 1000 genes from around 500 species of marsupial, rodent and bat, including samples from vouchered museum specimens wherever possible. This work will clarify taxonomic boundaries for several species of conservation concern, such as the Christmas Island flying-fox. Finally, the availability of reference genomes and phylogenies will provide a solid base for population-level studies. We are developing conservation genomic datasets for a selection of threatened mammal species. Using exon capture and genotyping-by-sequencing methods, we will measure genetic diversity and inbreeding, determine population structures, and identify adaptive variation. The inclusion of museum specimens as well as recently-collected samples will enable comparisons of modern and historic levels of genetic diversity. Conservation projects have been prioritised so that genomic data are contributing directly to urgent management decisions, for example to inform translocations, reintroductions and captive breeding.

GSA2018_APCC633102

The phylogeography of bacteria in the Antarctic

GSA: Ecological genetics

GSA2018 Abstract - Oral

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The Antarctic continent is generally assumed to be geographically isolated from the rest of the world. However, microorganisms have a high potential to disperse long distances through oceanic and atmospheric currents over short time periods, challenging our assumptions on its degree of isolation. In this study, we use biogeographic models that incorporate evolutionary and ecological processes to identify and explore putative dispersal events into Antarctica from other regions in several major soil bacterial groups. Our analyses identified approximately 50 overlapping terrestrial bacterial genera shared between the Antarctic and hundreds of sites around the world and shows that complex evolutionary dispersal pathways have contributed to the diversity of soil microbes in major ice-free locations in the Antarctic. In this presentation, we summarise these complex distributions by highlighting the phylogeographic patterns of the most abundant bacterial taxa and lineages, including lineages known for their ability to form symbiotic relationships with plants. These analyses provide informative baseline information that identifies particular lineages that have a higher evolutionary tendency to disperse into Antarctica. As climate continually changes in the continent, it is more likely that the Antarctic will experience changes in its resident microbial communities, either because the continent is colonized by new microbial species or because new environmental conditions promote the evolution of new microbial assemblages. Our evolutionary analysis of existing lineages and past dispersal events should provide critical information that will help to predict the nature and extent of the changes in Antarctica, necessary for informed management of the Antarctic biosphere into the future.

GSA2018_APCC63281

The roles of oncohistone H3.3 and ATRX abnormalities in cancers

APCC6: Chromosome architecture,
epigenetics and cell division

APCC6 Abstract - Oral

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One striking finding in the area of cancer epigenetics has been the identification of mutated histone genes (oncohistones) in paediatric glioblastomas (pGBMs). Two H3.3 mutations are found. The first mutation replaces lysine 27 with a methionine (K27M). The second one replaces glycine 34 by an arginine (G34R). H3.3G34R mutations always overlap with ATRX and p53 mutations, and these pGBMs are activated in the Alternative Lengthening of Telomeres (ALT) pathway, suggesting that H3.3G34R/ATRX/p53 mutations cooperate to drive ALT and GBM development. We have created cell models carrying H3.3G34R/ATRX/p53 mutations to recapitulate the initial driver epigenetic events that promote ALT. These mutants are compromised in heterochromatin formation at the telomeres, accompanied with increases in damage, transcription and formation of large PML bodies. KDM4 proteins are demethylases or epigenetic erasers that remove the methyl group from trimethylated H3K9 and H3K36. We find that H3.3G34R inhibits KDM4 catalytic function of and drives its aberrant distribution. As a result, it induces aberrant histone methylation pattern in genome and affects telomere chromatin maintenance. We propose KDM4 as a major driver that promotes ALT and the oncogenic process in GBMs. In the H3.3G34R/ATRX mutants, we also detect DNA copy loss at ATRX-bound ribosomal repeats, accompanied with severely reduced rRNA synthesis. ALT positive human sarcoma tumour samples are substantially reduced in rDNA copy. Moreover, ALT cancer cells show increased sensitivity to RNA polymerase I (Pol I) transcription inhibitor, suggesting the therapeutic potential of targeting Pol I transcription in ALT cancers. Our study provides insights into chromatin defects associated with ATRX/H3.3 mutations and development of ALT cancers.

GSA2018_APCC679171

Uncovering the principles underlying animal gene regulation

GSA: Evolutionary genetics and
comparative genomics

GSA2018 Abstract - Oral

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Comparative sequence conservation has been highly successful at producing detailed maps of coding and non-coding genes, under selective constraints, in sequenced genomes. Critically, we have a poor understanding of gene regulation — we cannot predict context-specific expression by studying sequence conservation across species. Recent studies have shown that the enhancers of genes responsible for tissue-specific phenotypes are evolving extremely rapidly and the majority of transcription factor binding sites have arisen in species-specific lineages between vertebrates. Using experimental and computational approaches, we demonstrate 1) gene regulatory network conservation between evolutionarily distant metazoans 2) ‘covert’ enhancers control deeply conserved developmental gene regulatory circuits. Our findings provide instructive examples of how regulatory circuits are rewired and provide key insights to understanding how tissue-specific gene expression patterns can be evolutionarily conserved despite constant flux in regulatory elements.

GSA2018_APCC671190

Understanding stem cell-niche interactions in the Drosophila germline

GSA: Genetics and medicine

GSA2018 Abstract - Poster

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The cellular microenvironment of the stem cell niche orchestrates the complex cellular interactions essential to stem cell self-renewal and differentiation. Proper organization and positioning of the niche is critical for preventing stem cell loss and tissue degeneration, or conversely, the emergence of cancer stem cells (CSCs) capable of unbridled proliferation. However, the mechanisms of oncogenic changes in the tumour microenvironment are still unclear. Here, we use the Drosophila model organism to demonstrate that the RNA recognition motif (RRM) protein Half-pint (Hfp) is important for germline stem cell niche signalling in vivo. Half-pint (Hfp), the Drosophila ortholog of the human tumour suppressor FIR, has been attributed to dual functions in mRNA splicing and transcription, particularly as a repressor of the MYC oncogene (Mitchell, 2010). Our exciting preliminary data demonstrate that depletion of the RNA recognition motif (RRM) protein Half-pint (Hfp) specifically in the Drosophila ovarian stem cell niche drives a phenomenon not previously reported; formation of an ectopic stem cell niche able to drive germline tumour formation. Interestingly the ability of depletion of Hfp in the stem cell niche to establish an ectopic niche, capable of non-autonomously driving germline stem cell tumours, was not dependent on MYC. Based on our observation that Hfp can bind genes in addition to MYC, we aim to determine 1) the direct Hfp binding targets in specifically in niche cells using Tandem-DamID and 2) identify targets responsible for the ectopic niche and germline tumour phenotype. Thus, we will identify the how Hfp regulates germline stem cell signalling, which will provide insight into oncogenic changes in the cancer microenvironment that might support cancer stem cell formation.

GSA2018_APCC633120

Unique mechanisms to silence the marsupial X

APCC6: Specialised chromosomes (sex chromosomes and B chromosomes)

GSA2018 Abstract - Oral

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Marsupial and eutherian X chromosome inactivation (XCI) share a common origin. This explains the overlapping histone code present on the inactive X in each group, but it fails to explain other fundamental molecular differences. In eutherians, the long noncoding RNA (lncRNA) XIST associates with epigenetic machinery and ribonuclear proteins to direct XCI. In marsupials, an independently evolved lncRNA – called RSX – is responsible for mediating silencing, but little is known about how it functions. Here we demonstrate that RSX interacts with many proteins responsible for epigenetic silencing, which it probably directs to the X chromosome to be silenced. Marsupial XCI is often considered the primitive cousin of eutherian XCI; however, our data highlight layers of independently evolved RNA mediated XCI complexities that are far from primitive.

GSA2018_APCC65492

Unique sex chromosome and sex-determining mechanism in Japanese native mammals, genus Tokudaia

APCC6: Specialised chromosomes (sex chromosomes and B chromosomes)

APCC6 Abstract - Oral

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In placenta mammals, a sex is genetically directed by inheritance of sex chromosomes at the time of fertilization. If the sex chromosome constitution is XX, the fertilized egg develops into a female, whereas if the constitution is XY, it becomes a male. Sex determination is actually carried out by the SRY (sex-determining region on Y) gene located on the Y chromosome. The expression starts in supporting cell precursors derived from coelomic epithelium and induces SOX9 (SRY-box 9) expression. The genus Tokudaia, Muridae, Rodentia does not adopt this sex-determining mechanism that is strictly conserved in mammals. Tokudaia consists of three species. Remarkably, two species, Tokudaia osimensis (Amami spiny rat) and Tokudaia tokunoshimensis (Tokunoshima spiny rat) have XO/XO sex chromosome constitution caused by lacking the Y chromosome. The SRY gene is also absent in these species. By contrast, the remaining one, Tokudaia muenninki (Okinawa spiny rat) has XX/XY sex chromosome constitution same as general mammals. However, their sex chromosomes have acquired the neo-X and neo-Y regions due to a fusion of a pair of autosomes to the X and Y chromosomes. Excessive duplicated copies of SRY are distributed on the Y chromosome, whereas, our previous studies indicated these were not functional. Tokudaia therefore have acquired a unique sex-determining mechanism independent of SRY. All species inhabit the southernmost islands in Japan. All are on the verge of extinction and successful artificial breeding has not been achieved, hence, we have studied Tokudaia using limited materials. In this session, I will show recent data of the unique sex chromosome evolution and the sex-determining mechanism in Tokudaia.

GSA2018_APCC648195

Using a xenograft model and RNA-seq to differentiate between the tumour and tumour microenvironment transcriptome.

GSA: Bioinformatics and genomics

GSA2018 Abstract - Poster

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In addition to the tumour cells, breast tumours contain other cell types such as fibroblasts, adipocytes, inflammatory and immune cells. Together with the extracellular matrix, these non-tumour cells compose the tumour microenvironment (TME). Complex interactions occur between tumour cells and the TME that can inhibit or stimulate tumour cell growth, metastasis and/or chemoresistance. While treatment of tumours with chemotherapeutic agents such as Abraxane, leads to apoptosis of tumour cells, it can also have consequences for the cellular makeup and transcriptional profile of the TME and these, like the increased infiltration of macrophages, may have detrimental effects. Transcriptome profiling of whole tumours from mice injected with tumour cell lines and treated with combinations of chemotherapeutic drugs has provided novel insights into pathways affected by different agents and diagnostic signatures. However, differences in the cellular composition of tumours from treated mice can have confounding effects and increase variation in whole tumour transcriptomes. To be able to examine the effects of co-treatment of Abraxane with another drug, we performed RNA-seq on tumours from mice injected with the human breast cancer cell line MDA-MB-231. We then separated reads mapped to the human and mouse genomes in silico, creating tumour and TME transcriptomes for control, Abraxane, drug and combination treated mice. Separation of the tumour and TME profiles revealed that while in the tumour cells, co-addition of the drug potentiated Abraxane's inhibitory effect on cell cycle genes and promotional effect on antigen presentation genes, in the TME it inhibited genes involved in the inflammation and migration responses and promoted genes involved in lipid and xenobiotic metabolism.

GSA2018_APCC646152

Using character-compatibility matrices in haploblock discovery.

GSA: Bioinformatics and genomics

GSA2018 Abstract - Oral

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Haplotype blocks (haploblocks) are contiguous stretches of DNA that have not been affected by recombination since the haplotype first evolved. Absence of recombination increases non-random association of alleles at polymorphic sites within the haploblock, and subsequently those polymorphic sites are mutually phylogenetically concordant. These properties enable efficient mapping of disease-associated loci, and enhances our ability to uncover historic demographic events and understand natural selection at discrete genomic regions. Common methods of discovering haploblocks can be divided into two groups: 1) linkage disequilibrium (LD) based methods and 2) methods based on gametic evidence of recombination (e.g. four-gamete test [FGT]). LD-based haploblock discovery methods lack specificity as they can tolerate a degree of recombination-induced noise, which leads to spurious block boundaries. Conversely, the FGT lacks sensitivity towards mutational noise. However, as diploid genomes are spanned by recurrent and back mutations, thereby causing haploblock boundaries to terminate prematurely and thus reducing the number of markers within a haploblock. In contrast to LD-based methods or the FGT, character-compatibility (Co) is another recombination-based method that can allow for removal of mutational noise thus retaining the specificity of the FGT without false-positive haploblock boundaries, as may be discovered by LD-based methods. We have for the first time employed Co-matrices to discover haploblocks in a single population from the 1000 Genomes Project dataset, and compared them to haploblocks discovered with LD-matrices. Our preliminary results suggest that haploblocks discovered using Co-matrices are more phylogenetically concordant and their boundaries terminate earlier than those discovered with LD-matrices, suggesting haploblocks discovered by LD-matrices erroneously extend over recombination points. Given increased specificity and sensitivity of Co compared to the LD-based and the FGT methods respectively, Co may significantly impact our ability to understand the evolutionary history of discrete regions of the human genome, with further possibilities for disease loci mapping.

GSA2018_APCC61424

Using meta-transcriptomics to reveal the hidden diversity of RNA viruses in aquatic hosts

GSA: Evolutionary genetics and comparative genomics

GSA2018 Abstract - Oral

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The total assemblage of viruses on earth is termed the ‘virosphere’, the vast majority of which is undescribed. It is futile to attempt to predict viral emergence events until there has been a dramatic increase in the number of viruses identified and sequenced. Hitherto, this task was greatly hampered by a strong sampling bias toward pathogens that cause disease in humans or in economically important animals and plants. This is set to change with the development of whole transcriptome (RNA) shotgun sequencing. RNA-Seq is a next generation sequencing technique that reveals the presence and quantity of transcriptomes, and can reveal the entire virus composition within an individual (i.e. the virome), as well as their abundance, as opposed to being limited to a specific pathogen. Using a ‘meta-transcriptomics’ approach I provide a comprehensive view of viruses in aquatic hosts. Viruses are abundant in marine and freshwater environments. Crucially, fish likely harbour a greater diversity of viruses than any other class of vertebrate, and all virus families previously thought to only infect mammals have recently been found in bony fish. These viruses are the likely progenitors of viruses that infect a broad range of hosts, including mammals, amphibians and birds. The recent identification of hepadnaviruses in fish has revealed many more instances of host jumping than previously realised, including from aquatic to terrestrial vertebrates. Marine mammals, such as whales, share this aquatic environment and so provide an opportunity to study viruses across diverse host types and draw comparisons with their terrestrial counterparts.

GSA2018_APCC634108

Using the q-profile to better detect landscape patterns of genetic differentiation

GSA: Ecological genetics

GSA2018 Abstract - Poster

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The q-profile is a continuum that includes common measures of genetic differentiation: allelic richness (q= 0), Shannon (q=1) and Fst (q=2). Using all three measures is expected to give the best chance of detecting genetic patterns, because each measure has specific sensitivities. For example, Fst is a commonly used measure of genetic distance; however, due to the overemphasis on common types (alleles), patterns of differentiation in rare alleles may be missed. Conversely, allelic richness has an overemphasis on rare types and is thus prone to sampling errors. Information measures (Shannon) weight alleles proportional to their abundance and allow for the detection of patterns for both rare and common alleles. While the benefits of Shannon measures of genetic diversity have been demonstrated in the theoretical literature, empirical genetic studies using Shannon are uncommon, with preference to Fst or allelic richness. By using all three, we test these measures on empirical data sets of plant populations across NSW as well as analogous simulated data to determine the scenarios to which each method is best applied.

GSA2018_APCC659148

Uterine recognition of pregnancy was an ancestral feature of the first live bearing mammals

GSA: Evolutionary genetics and
comparative genomics

GSA2018 Abstract - Oral

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In human pregnancy, recognition of a developing fetus within the uterus is essential to maintain uterine activity and support the embryo through an extended gestation. In most marsupials (with the exception of macropods), pregnancy is shorter than the estrus cycle, and for this reason it has been assumed that recognition of pregnancy is not necessary, and was a trait that evolved in the first eutherian (placental) mammals. To investigate whether uterine recognition of pregnancy might be more widespread in live bearing mammals, we examined reproduction in the grey short tailed opossum (Monodelphis domestica), a marsupial with what is assumed to have the most pleiotropic mode of pregnancy for live bearing mammals. We examined the morphological and gene expression changes in the uterus of females in the estrus cycle and compared these to the observed changes during pregnancy. We found that while the uterus undergoes substantial changes in pregnancy, these changes occur in a programmed fashion during estrus, and for the most part do not appear to be impacted by the presence of a fetus. However, transcriptionally we saw big differences between the uterus of pregnant and estrus animals. Gene onology analysis shows us that the genes up-regulated due to the presence of a fetus are involved in macromolecule transport, inflammation, and metabolic activity. Our results suggest that while the uterus exhibits programmed changes in response to ovulation, the transriptional landscape of pregnancy responds to the presence of a fetus, and upregulates suites of genes that may be essential for fetal support. These results are consistent with uterine recognition of pregnancy dating back to the origin of live birth in mammals.

GSA2018_APCC611118

Variants in the host genome may inhibit tumour growth in devil facial tumours: evidence from genome-wide association

GSA: Ecological genetics

GSA2018 Abstract - Poster

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Devil facial tumour disease (DFTD) has decimated wild populations of Tasmanian devils since its emergence. As the disease has the ability to avoid immune detection, it spreads rapidly both through populations and within individual hosts. Only a very few devils have been observed to spontaneously recover from the disease which is otherwise invariably fatal. In one north-western population, West Pencil Pine (WPP), it has been observed that some devils have spontaneously cleared the cancer where it had previously been considered 100% fatal. These individuals were sampled in the wild with evidence of DFTD tumours that were recorded to have shrunk or disappeared on subsequent sampling trips. We sequenced the whole genomes of 11 Tasmanian devils and combined them with 2 previously published devil genomes to obtain a sample set of 7 devils with regressed tumours and 6 controls. A genome-wide association (GWAS) was conducted to determine if any regions segregated between cases (regressed devils) and

controls. Candidate regions arising from the genome were searched to identify any genes that may be involved in the ability for the host to overcome DFTD infection. Following targeted sequencing in an additional 13 controls and one regressed animal from WPP, the significant association of SNPs in and around the PAX3 gene was retained. The PAX3 gene is a regulatory gene involved with developmental pathways through regulation of intermediate targets such as bone morphogenic proteins (BMPs). As such PAX3 has been associated with the development and metastasis of many cancers due to its regulatory activity. PAX3 is also involved in angiogenesis through its interaction with VEGF. It is possible that PAX3 expression in the regressed devils results in reduced angiogenesis in the tumour microenvironment, leading to slowed tumour growth and eventual regression. Further research is required to confirm the involvement of PAX3 in DFTD regression.

GSA2018_APCC616111

Waking the sleeping dragon - epigenetic mechanisms that govern hibernation in the central bearded dragon.

GSA: Genetics and epigenetic regulation

GSA2018 Abstract - Oral

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Hibernation is a seasonal physiological state utilized by animals to conserve energy during winter. Hibernation is complex, involving large changes in metabolism and cellular function. While epigenetic mechanisms have been extensively studied in hibernating mammals, the role of epigenetics has yet to be uncovered in reptilian hibernation. Total transcriptomic profiles of the central bearded dragon (Pogona vitticeps) were generated for brain, heart and skeletal muscle at three time points: 1) late hibernation, 2) two days post-arousal, and 3) two months post-arousal to elucidate changes in gene expression. Additionally, reduced representation bisulfite sequencing was used to generate DNA methylation profiles for brain and skeletal muscle at the three time points to understand the role of DNA methylation in regulating the hibernation phenotype. Transcriptomic analysis revealed large changes in gene expression between hibernating and post-arousal individuals across all tissues, with chromatin modification and microRNA-mediated gene silencing genes overrepresented during hibernation. RRBS results suggest that global changes in DNA methylation does not play a substantial role in mediating phenotypic plasticity during the hibernation season. Rather, DNA methylation at a gene-specific level mediates this change, particularly across the transcriptional start site and first exon of genes. This study provides exciting insights into the molecular mechanisms that govern hibernation in the central bearded dragon, suggesting a complex interaction of a myriad of epigenetic regulatory elements.

GSA2018_APCC656173

What we can learn from sponges about the molecular and cellular bases of animal stem cells

GSA: Development and cellular genetics

GSA2018 Abstract - Oral

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The evolution of multicellular organisms is generally thought (and seems likely) to have been accompanied by the evolution of a stem cell system. Sponges, some of the early-evolved metazoans, have totipotent/pluripotent stem cells. Thus, uncovering the cellular and molecular bases of the sponge stem cells will not only be crucial for understanding the ancestral gene repertoire of animal stem cells, but will also give us clues for understanding the evolution of molecular mechanisms of maintaining multipotency (pluripotency) and differentiation ability during animal evolution. We have been working to clarify the molecular and cellular bases of the stem cell system of demosponges. Using an asexual reproduction system (gemmule hatching), the development of fully functional juvenile sponges from a group of stem cells alone, of a freshwater sponge, Ephydatia fluviatlis, we established methodologies for molecular studies and sequencing resources. Based on our results and previous histological studies, we proposed that the sponge stem cell systemconsists of two types of cells, archeocytes (mesenchymal pluripotent stem cells) and choanocytes (which generally function as food-entrapping cells but exhibit pluripotency in specific circumstances, for example, by undergoing meiosis to produce gametes). I would like to introduce our gene-expression and transcriptomic analyses of those two types of cells, especially archeocytes, and discuss the ancestral toolkit of animal stem cells and its biological significance.

GSA2018_APCC671182

When is a global repressor not a repressor? Genome-wide profiling of the Snail family member transcription factor Worniu in neural stem cells

GSA: Development and cellular genetics

GSA2018 Abstract - Oral

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The Snail family transcription factor Worniu is expressed specifically in neural stem cells (NSCs), and has been previously shown to be required for larval NSC maintenance. In synthetic reporter assays the protein functions as a global repressor; however, its direct binding targets in vivo remain unknown. Here, we describe the cell-type specific genome-wide binding profiles of Worniu in third instar larval NSCs using Targeted DamID technique. We relate these binding data to larval NSC transcriptional and chromatin state data, together with previously published transcriptional profiling data for both wild-type larval NSCs, and NSCs depleted for Worniu, to identify direct transcriptional targets. We find that Worniu binds to and represses a small number of genes involved in synaptic transmission, via a novel Trithorax-repressive chromatin state. Unexpectedly, however, we show that Worniu also binds promiscuously across open chromatin within the genome, including at active promoters and enhancers, inconsistent with a role as a global repressor. Combined, we suggest a model for Worniu's role in maintaining NSC identity within the brain.