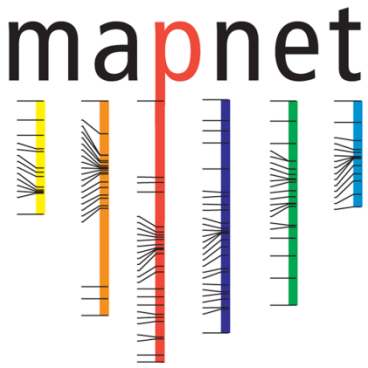

1-2 Sept 2016, Nelson, New Zealand

ABSTRACTS

Oral 1. Predicting routes of horizontal gene transfer using completely sequenced genomes

Heather Hendrickson, Massey University

Horizontal gene transfer (HGT) is a process by which large quantities of genetic information are transferred between bacteria in nature. Despite significant frequencies of DNA admixture there is ecological and phenotypic cohesion amongst higher taxonomic groups in the eubacteria. This suggests that there are mechanisms that impose structure or rules on HGT. Most bacterial genomes have a functional architecture composed of repeat sequences called Architectural IMparting Sequences or AIMS. These are 8 base pair sequences that have distributions in the genome that suggest that they function in orienting DNA associated enzymes to facilitate processes like DNA segregation. AIMS are phylogenetically conserved such that HGT events that occur between closely related bacteria will tend to preserve AIMS structure while more distant transfer events will disrupt AIMS structure therefore be detrimental to the organism.

AIMS are not under equally strong selection across the length of the chromosome. Selection on orientation and abundance is strongest at the terminus and eases towards the origin. In order to understand the degree to which AIMS shape bacterial genomes, we have analyzed naturally occurring disruptions in bacterial chromosomes. We have found that both inversions and insertions appear to be affected by AIMS distributions in bacterial genomes. Last, we used completely sequenced genomes to produce a predictive road map of paths of horizontal gene transfer between species based on AIMS compatibility between donors and recipients. According to these data, some HGT will non-reciprocity of genome compatibility between some donors and recipients. AIMS therefore suggest that some bacterial clades should be expected to be universal donors but not universal acceptors of genetic transfer.

Oral 2. The *Escherichia coli* nucleoid: an organized structure that is shaped by replication and transcription

Justin O'Sullivan, University of Auckland

To be supplied

Oral 3. PacBio methylome sequencing of *Neisseria meningitidis* – potential associations with disease

Una Ren, ESR

To be supplied

Oral 4. Causal modeling of relationships between genotype and phenotype using multi-omics data

Heather Cordell, University of Newcastle, UK

Over the past 10 years, genome-wide association studies (GWAS) have been extraordinarily successful at identifying genetic variants associated with common, complex disorders. However, a typical GWAS gives little insight into the underlying biological mechanism through which the associated genetic variants are implicated in disease. Genetic variants identified through such investigations represent the first step along the causal pathway to disease development, and one strategy to help elucidate the underlying causal mechanisms is to make use of data of different types, including genetic data, "omics" data such as measurements of DNA methylation and gene expression, and clinical variables related to disease phenotype. A variety of methods can be used to model the relationships between these different variables. In this talk I will focus on methods that assume at least a proportion of subjects have been measured on all variables of interest. I will outline the methodological approaches that we have been exploring based on causal inference techniques, and present the results of computer simulations and real data analyses illustrating the utility of these approaches.

Oral 5. R/qtl2: System genetic analysis in multi-parent populations

Karl Broman, University of Wisconsin

To be supplied

Oral 6. Variance GWAS

Wenhua Wei, University of Otago

To be supplied

Oral 7. Interactive exploration of genome-scale datasets

Nick Burns, Ruth Topless, James Boocock, Michael A Black, Tony Merriman
Department of Biochemistry, University of Otago, Dunedin, NZ

Genome-scale analyses are now well-established in the study of complex human diseases. The variety of statistical tests have extended from the traditional genome-wide association study (GWAS) to include a wide range of functional and spatial traits. There is a need for analytical and exploratory tools to combine and compare results across multiple genome-scale analyses. Unfortunately, the sheer size of these datasets imposes severe restrictions on the types of questions that can be explored. Current genome browsers and exploratory tools are mostly restricted to simple lookups and summaries which miss the potential to discover more compelling relationships.

The Genotype-Tissue Expression (GTEx) project is a ready example of the problems researchers face when dealing with such large, heterogeneous datasets. Combined, the genotype, gene expression and expression quantitative trait loci (eQTL) datasets total more than 500GB of data. The GTEx portal provides a user-friendly interface to explore single-tissue summaries of this data but there are no means by which to compare results across tissues or genes, nor to directly contrast the gene expression profiles with interesting eQTLs. The sheer size of these data necessitates such restrictions, but limits the exploratory potential of such a rich dataset.

We have developed a data warehouse which can facilitate full, unrestricted exploration of genome-scale datasets, which we have applied to the gene expression and eQTL datasets from GTEx together with various GWAS summary datasets. The benefits of deduplication and optimised data access strategies reduce the problems of scale normally associated with these data. This presentation will demonstrate an interactive interface to the data warehouse, using R and Shiny, which provides rich and intuitive visualisations via a web browser, allowing users to quickly explore loci across the GTEx datasets, including investigating relationships to other loci of interest.

Oral 8. Conservation genomics

Brad Shaffer, University of California LA

To be supplied

Oral 9. Kakapo genome

Bruce Robertson, University of Otago

To be supplied

Oral 10. A conservation genomic approach for maximising genetic diversity in a critically endangered New Zealand bird.

Galla, S.J. [1], Hale, M.L. [1], Santure A.W. [2], Steeves, T.E. [1]

[1]School of Biological Sciences, University of Canterbury, Christchurch, NZ

[2]School of Biological Sciences, University of Auckland, Auckland, NZ

Endangered species management can utilise captive breeding for translocation as a technique to prevent extinction and enhance species recovery. Captive breeding programmes often make pairing decisions based on available pedigree data to minimise inbreeding and maximise genetic diversity in an effort to maintain the ability to adapt to environmental change (i.e., evolutionary potential). However, captive pedigrees are often shallow (<5 generations deep), incomplete and error-prone. While genetic-based techniques (microsatellites) offer these programmes a way to estimate genetic relatedness among individuals, emerging evidence indicates that genetic-based measures of relatedness based on microsatellites are relatively poor indicators of genome-wide diversity, particularly in genetically impoverished endangered species, and a better indication of genome-wide diversity should be obtained from genomic-based measures of relatedness based on genome-wide single nucleotide polymorphisms (SNPs). Here, I will provide a comparison of pedigree-based, genetic-based and genomic-based estimates of relatedness in kakī (*Himantopus novaezelandiae*) from the 2015/2016 breeding season as part of a larger effort to determine which of these approaches is the most efficient and effective for maintaining evolutionary potential in this critically endangered New Zealand endemic. Using kakī as a proof-of-concept, this research can be applied to other poorly-pedigreed captive breeding translocation programmes in New Zealand, and worldwide.

Oral 11. Assembling the South Island robin genome to understand inbreeding depression: the pitfalls of *de novo* assembly faced by ecological geneticists

Helen R. Taylor[1], James V. Briskie[2], Kim M. Rutherford[1], Neil J. Gemmell[1]

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Assembling genome sequences *de novo* has become relatively common practice for ecological geneticists seeking to understand the genomic mechanisms underlying various traits and processes. However, ecological geneticists are not generally bioinformaticians and are often inexperienced in genome sequencing and assembly. Unfortunately, there is a lack of information in the scientific literature regarding guidelines for *de novo* assembly and "best practice" can vary substantially between taxa. We are currently assembling a *de novo* genome for the South Island robin (*Petroica australis australis*). We aim to use this genome to elucidate the links between inbreeding depression and poor male fertility, a subject that has rarely been assessed in wild populations, has almost exclusively been examined in mammals, and has not yet been approached using full genome sequencing. Here, we will outline the steps we have taken to ensure the best quality assembly possible (pre-assembly quality control procedures), as well as comparisons between different assembly software. We will explain some of the pitfalls we have faced as ecological geneticists new to genome assembly, and how we have overcome these issues, emphasising the need for general guidelines for de novo assembly non-model species. Finally, we will explain how we hope to use our robin genome to assist in seeking out the signatures of inbreeding that might explain decreases in male fertility in this, and other, species.

Oral 12. Whole-transcriptome profiling of flexible sexual phenotypes in a model sex-changing fish, the bluehead wrasse.

Erica V. Todd[1], Hui Liu[1], Melissa Slane-Lamm[2], John Godwin[2], Neil J. Gemmell[1]

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[2] Department of Biological Sciences, North Carolina State University, Raleigh, NC 27695, USA.

Sex is increasingly seen as a continuous, rather than a dichotomous trait. Sex is phenotypically plastic in many marine fishes and results from environmentally-sensitive differential gene regulation. Bluehead wrasse (*Thalassoma bifasciatum*) are highly-social reef fish and well-studied models of sexual plasticity. These diandric, protogynous (female-first) hermaphrodites have three sexual morphs as adults whose development is plastic and socially cued. Bluehead wrasse mature as male (primary males) or female, but each have the capacity to become dominant (secondary) males later in life. Large, brightly coloured secondary males actively defend and court a harem of females, whereas primary males are female-mimics that employ a 'sneaker' mating strategy. Using whole-transcriptome RNA-sequencing (RNA-seq) we have explored the molecular basis of plastic sexual phenotypes in the bluehead wrasse brain and gonad. Differential expression analysis identified thousands of genes important in the maintenance of the primary male, secondary male, and female phenotypes. In the brain, secondary males had the most distinct expression patterns, whereas expression profiles of primary males reflect their female-like behaviour, not their male sex. We find that isotocin (homologue of mammalian oxytocin) is overexpressed in secondary males, supporting recent evidence for its regulatory role in teleost social interactions, especially those related to dominance and rank. Gonadal expression profiles were strongly sex-biased, although secondary males upregulated genes involved in androgenesis and in the maintenance of secondary sexual characteristics (i.e., colouration and territoriality). Further investigations into the molecular basis of sexual plasticity are now underway, including transitions between alternative phenotypes and comparisons of gene expression patterns in evolutionarily divergent systems.

Oral 13. Genetic diversity in Tasmanian Atlantic Salmon and prospects for GWAS and genomic prediction

James Kijas, CSIRO

To be supplied

Oral 14. Genomes, gene discovery and fish immunity

Steve Bird, Waikato University

To be supplied

Oral 15. Making a splash: using genomics to aid the New Zealand aquaculture industry

Rachael Ashby, University of Otago/AgResearch

To be supplied

Oral 16. Phenotyping and genomic resources for domestication of the marine finfish snapper (*Chrysophrys auratus*)

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[1]The New Zealand Institute for Plant & Food Research Limited, Nelson, New Zealand
[2]School of Biological Sciences, Victoria University of Wellington, Wellington, New Zealand
[3]Molecular Ecology and Evolution Group, Department of Biology, Lund University, Sweden

Mapping the genetic basis of phenotypic variation is a major challenge for biology. The recent development of next generation sequencing technologies has dramatically improved our ability to address this challenge, particularly in non-model species which have limited genomic resources. Australasian snapper (*Chrysophrys auratus*) is one such species that has limited genomic resources and is of interest for fisheries and as a potential aquaculture species. A range of phenotyping and genomic resources are being developed at Plant and Food Research to support domestication and genotype-phenotype mapping in this species. In this talk, I will discuss the development of these resources including 1) genotyping of a three generation pedigree using Genotyping by Sequencing (GBS), 2) development of automated software for rapid phenotyping from images, 3) identification of QTLs associated with growth rate, and 4) development of a recently assembled genome.

Keywords: Snapper, QTL, GBS, genome, New Zealand

Oral 17. Genomics assisted yield improvement in alfalfa - Are we making more hay?

Charles Brummer, University of California, Davis

To be supplied

Oral 18. Mānuka genome assembly using chromosome conformation capture Hi-C analysis

David Chagné*[1], Amali Thrimawithana[1], Ross Crowhurst[1], Ivan Liachko[2], Shawn Sullivan[2], David Lewis[1], Julie Ryan[1], Munazza Saeed[1], Tracey Van Stijn[3], Jeanne Jacobs[3], Rudiger Brauning[3], Shannon Clarke[3], Cecilia Deng[1], Helge Dzierzon[1], Ella Grierson[1], Elena Hilario[1], Kathy Schwinn[1]

*presenting
1 The New Zealand Institute for Plant & Food Research Ltd
2 PhaseGenomics Ltd
3 AgResearch Ltd

We have evaluated a novel strategy combining capture of chromatin interaction within the nucleus, next-generation sequencing (NGS) and new bioinformatics methods (Hi-C) for developing a near-complete pseudo-chromosome assembly of the mānuka (*Lepstospermum scoparium* 'Crimson Glory') genome. The method relies on the folded confirmation of chromosomes inside the nucleus and the assumption that segments of the same chromosome are in closer 3D proximity with other segments of the same chromosome than to segments of other chromosomes. We fixed and captured chromosome fragments that were in close proximity and then sequenced them using NGS. The Hi-C bioinformatics approach utilizes the probability that chromosome fragments within paired-end NGS reads are close in proximity and this probability can be used to aid scaffolding. Using the Hi-C analysis, we inferred the location of previously unanchored contigs and created chromosome length super-scaffolds, enabling a ~100 times increase in the N50 scaffold length compared with assembly in the absence of this analysis. Furthermore, the Hi-C technique allowed the separation of the mānuka genome from contigs assembled from associated endophytic fungal and bacterial species. The newly assembled mānuka genome was compared to the genome of *Eucalyptus grandis* and high density genetic maps of mānuka constructed using genotyping-by-sequencing. The mānuka genome sequence will help shed new light on the genetic control of unique characters such as nectar and foliage biochemical composition, flowering time and disease resistance.

Oral 19. “Genotyping-by-sequencing” platform to recover genetic relatedness

Klápště, J.[1], Telfer, E.[1], Dungey, H.[1]

[1]Forest Genetics, Scion, 49 Sala Street, Rotorua, New Zealand.

Current developments in next generation sequencing technologies have enabled the implementation of genomics in organisms with no reference genomes, as is the case for most forest trees. The availability of genomic resources and success of genomic selection achieved in animal and crop breeding has now turned the attention of breeders towards the implementation of genomic selection in forest tree breeding programs. Genomic predictions capitalize on capturing genetic relatedness, co-segregation and linkage disequilibrium (LD) between markers and causal variants (quantitative trait loci - QTL). However, common forest tree genome properties such as large size, fast LD decay and large effective population size hinder the ability of genomic prediction models to capture LD between markers and QTLs, which is usually the most stable part of genomic prediction. This means that genomic prediction models in forest trees rather depend on the quality of relatedness recovery, and should be used to predict only related genetic material.

Our study is focused on the investigation of a “Genotyping-by-sequencing” (GBS) platform to recover genetic relatedness. This platform is cost efficient to genotype large training populations but usually suffers a large amount of missing data and low sequence depth. Under such conditions, it can be problematic to perform reliable missing data imputation and relatedness estimations in species with a large effective population size. We deployed tools specifically proposed for GBS data[1] to construct a genomic relationship matrix. Our analysis found there was benefit in avoiding missing data imputation and taking read depth into account for the reliable recovery of genetic relatedness. This strategy can efficiently identify genomic outliers and remove them from the training population, since the informativeness and ability to predict such individuals would be very limited.

1. Dodds, KG., McEwans, JC., Brauning, R., Anderson, RM., van Stijn, TC., Kristjánsson, T, Clarke, SM. (2015). *Construction of relatedness matrices using genotyping-by-sequencing data.* BMC Genomics 16:1047. Doi: 10.1186/s12864-015-2252-3

Oral 20. Predicting the future of C3 plants using a modified small structural protein

Nick Roberts, AgResearch

To be supplied

Oral 21. GWAS for weight in Samoans

Steve McGarvey, Brown University

To be supplied

Oral 22. Identification of biologically informative uncommon genetic variants by resequencing in extreme phenotype

Tony Merriman, University of Otago

To be supplied

Oral 23. Insulin resistance and diabetes - insights from monogenic causes

Rinki Murphy
University of Auckland

Identifying those with diabetes or severe insulin resistance disorders that result from a single gene defect has not only enabled improved diagnostic and clinical management of such patients, but has also resulted in key biological insights into the pathophysiology of the increasingly common forms of diabetes and insulin resistance. This presentation will outline the major types of human monogenic disorders that I have found in my clinical practice that result in pancreatic beta cell forms of early-onset diabetes or severe insulin resistance manifesting in absence of obesity at a young adult age. The lessons they provide for current understanding of the molecular pathogenesis of common type 2 diabetes and insulin resistance are highlighted.

Oral 24. The Dynamic and Hypervariable Nature of *LPA*

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High lipoprotein(a) levels are a major risk factor for cardiovascular disease[1]. Significantly elevated or lowered levels of lipoprotein(a) are caused by variation in the *LPA* gene[2]. Of these, the most significant is copy number variation at a repeat region of LPA (KIV_2), there being 2-40 copies of the repeat at any one allele. The size of this *LPA* repeat region has been shown to be the basis of 30-70% of lipoprotein(a) phenotypic variance[2]. A second major genetic determinant of this phenotypic variance is predicted to be sequence variation located in the *LPA* repeat array[3]. Identifying such variation brings the challenges inherent in sequencing highly repetitive regions.

In this project, next generation sequencing methods were used to reveal the sequence variation across the *LPA* gene in 48 Caucasian individuals. A focus has been placed on the KIV_2 repeat array, including targeted deep sequencing of the two exons encoded in the repeat. Amplicon generation was achieved with PCR-based methods (sequenced at an average coverage of 2,500 reads per exon) and RPA-based methods (sequenced at an average coverage of 12,000 reads per exon). These two methods were coordinated to reduce the significance of any amplification errors or bias towards certain repeat types.

So far, this custom method has identified 10 new sequence variants within the *LPA* repeat array – the total number of known variants in this region now being 28[3]. Two of the novel variants have only been identified in individuals with low or null lipoprotein(a) levels, and are predicted to change the structure of the encoded protein. Other novel variants are illustrative of recent repeat expansion and concerted evolution across the *LPA* repeat region, detailing the dynamic evolution of the *LPA* gene.

1. Kamstrup PR., Nordestgaard BG. *Elevated lipoprotein (a) levels, LPA risk genotypes, and increased risk of heart failure in the general population*." JACC: Heart Failure 4.1 2016

1. Kronenberg F., Utermann G. *Lipoprotein(a): resurrected by genetics.* J Intern Med. 2013

2. Noureen A., Fresser F., Utermann G., Schmidt K. *Sequence variation within the KIV-2 copy number polymorphism of the human LPA gene in African, Asian, and European populations.* PloS one. 10(3), e0121582 2015

Oral 25. Inbreeding depression in Eastern and Western Polynesians and metabolic disease

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Homozygosity caused by consanguineous union has long been associated with an increased prevalence of rare Mendelian disorders. In contrast, the role of homozygosity in relation to quantitative traits and complex disease susceptibility is less well established. Recent work has shown that an increased burden of homozygous DNA segments is associated with reduced height and cognitive ability in diverse human populations. The ROHgen Consortium study investigates the mechanism of inbreeding depression by exploring different measures of homozygosity. Long homozygous segments arise from recent common ancestry and bring all variants, including rare causal variants, into a homozygous state. In contrast, shorter homozygous segments may arise from more ancient common ancestry and thus capture only the dominance effects of more common variants. Multivariate linear models have been used to show that FROH (the fraction of all autosome in a run of homozygosity >1.5Mb in length) is more predictive of inbreeding depression than traditional estimates of inbreeding coefficient based on individual SNP homozygosity.

Here, we assess the scope of inbreeding depression in individuals from the Eastern (n=692) and Western Polynesian (n=568) cohorts submitted to the study. We analysed the effect of FROH on nine quantitative metabolic traits of interest. Initial findings show that both cohorts have increased inbreeding depression when compared to the sample UK population. Using FROH there is greater variation in inbreeding depression when using the more traditional measures.

Oral 26. SheepGenomesDB used to identify genomic features impacted by domestication and selection

James Kijas, CSIRO

To be supplied

Oral 27. Sequence variation currently not utilized in genomic selection of New Zealand dairy cattle

Chris Couldrey, Livestock Improvement Corporation

To be supplied

Oral 28. High-density genotyping of the New Zealand sheep industry

Suzanne Rowe, AgResearch

To be supplied

Oral 29. Genomic Analysis of a High Vitamin C Trait in *Actinidia* Hybrids

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Kiwifruit of the genus *Actinidia* are still early in domestication and harbour substantial genetic variation that can be accessed by hybridisation. The species *A. eriantha* is notable for containing very high vitamin C concentrations (> 600 mg/100 g fresh weight) but has small fruit. Analysis of vitamin C concentrations in A. *eriantha* hybrid populations revealed evidence for bimodal segregation of vitamin C contents, consistent with a major gene. We are conducting genetic analysis of this trait using whole-genome sequencing methods to develop strategies for tagging and introgressing consumer traits from these polyploid interspecific crosses, to support breeding of novel fruit types.

To identity genome regions linked with VitC content we conducted Pool-GWAS by sequencing four total DNA pools of N>=20 individuals sampled from eleven tetraploid (*A. chinensis* var. *deliciosa x A. eriantha*) x (*A. chinensis* var. *deliciosa x A. chinensis* var. *chinensis*) families. Pool were as follows: high vitamin C/high fruit weight; high vitamin C/low fruit weight; low vitamin C/high fruit weight; low vitamin C/low fruit weight. Global scans for association of allele frequencies with vitamin C content using the Popoolation2 pipeline revealed significant associations across a 7-Mb region on a single chromosome of the *A. chinensis* reference genome. Analysis of gene and repeat density and of recombination in the *A. chinensis* genome support the hypothesis that this is a region with restricted recombination. This suggests that the high vitamin C trait may be suited to marker-aided selection. Inspection of bam alignments revealed that local haplotypes consistent with different donor genome origins could be readily distinguished, confirming that mapping by sequencing will be a highly effective strategy for analysis of *Actinidia* hybrids. Progress in developing validation strategies, and alternative workflows for poolseq analysis will be discussed.

Oral 30. CpG DNA hypomethylation occurs in human Autosomal Dominant Polycystic Kidney Disease

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The DNA methylation status of the genome has been shown to contribute to development and progression of many cancers. However, the role of DNA methylation in Mendelian diseases has not been extensively studied. The process of cystogenesis in the Mendelian disease Autosomal Dominant Polycystic Kidney Disease (ADPKD) shares similarities with many neoplasms, due to the characteristic large fluid-filled cysts that develop within the kidney. Most ADPKD patients carry a mutation in one copy of the *PKD1* gene, but loss of function of the second *PKD1* allele contributes to disease progression. It has been hypothesised that an epigenetic mechanism such as DNA methylation could contribute to a change in *PKD1* expression in the second allele, playing a role in cyst progression.

To understand the role of DNA methylation in ADPKD, whole genome-scale methylomes interrogating a 30-fold enrichment of the CpG islands of human ADPKD kidney tissue (n=2) were generated using reduced representation bisulfite sequencing, and compared to those of normal kidney tissue (n=3). Comparative data between the two groups indicated overall hypomethylation in the ADPKD tissue, which is similar to the methylation alteration often seen in cancer. There were 25 significantly differentially methylated fragments (DMFs) identified, and 52% of these were hypomethylated. DMFs were identified with an ANOVA test with FDR <0.25 and a 15% difference in methylation status. There was an overall enrichment of DMFs overlapping gene bodies compared to other genomic elements. A hypermethylated DMF was also found within the *PKD1* gene body, overlapping an intron/exon junction. Methylome analysis of additional ADPKD tissue samples, and cell lines derived from individual cysts, will be used to expand these data sets, to develop a rationale for targeting DNA methylation in ADPKD as a suitable therapeutic strategy.

Oral 31. Old challenges made harder: Optimising linkage map construction in the modern genomics era

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Genome-wide genotyping is now affordable for most commercial breeding programmes in New Zealand. However, in non-model organisms such as coniferous trees, the lack of comprehensive reference genome sequences means genetic maps need to be developed based on linkage analyses. Linkage maps have many applications, most recently as resource for genomic selection. Using genotypic data generated from a 44K exome capture panel we have conducted linkage analysis in two outcrossed full sib families of *Pinus radiata* D.Don. A total of 31 165 and 21 319 single nucleotide polymorphism (SNP) markers were identified as segregating in each of the families, respectively, based on observed genotypic variation in parents as well as offspring (*n* = 90 - 93). Clustering analyses (LOD 8.0) showed almost all markers mapped to a cluster with >200 markers. Twelve clusters were identified in both families, equal to the haploid number of chromosomes. Marker order in each cluster was determined using a maximum likelihood algorithm implemented in JoinMap 4.1{Van Ooijen, 2006 #3609}. However, the resulting linkage maps were substantively longer than expected based on previously constructed maps, in part due to the accumulation of errors and difficulties placing inaccurately genotyped loci. To obtain biologically realistic estimates of map length, we reduced the number of markers to 100 per cluster, and then reconstructed maps of the three clusters in both families. This resulted in substantively shorter maps than those constructed using all markers. Decreasing the number of markers to 50 per cluster resulted in further reductions in linkage group lengths, indicating further optimisation is needed. Nonetheless our results show that vast increases in the volume of polymorphisms generated by modern sequencing-based genotyping technologies does not necessarily result in higher quality genetic maps, thus substantively fewer – and carefully selected - markers are needed to produce biologically valid genetic maps.

1. Van Ooijen J: *JoinMap® 4, Software for the calculation of genetic linkage maps in experimental populations.* Kyazma BV, Wageningen 2006, **33**:10.1371.

Poster G1. Screening loss of function mutations in fgr/badh2 gene for conferring fragrance in rice varieties of Pakistan

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University of Karachi, Karachi, Pakistan

Abstract:
Fragrance in rice is conferred by mutations resulting in loss of function of the fgr/badh2 gene product along with accumulation of 2-acetyl-1-pyrroline (2-AP). This fgr/bdh2 gene corresponds 8bp deletions and three single nucleotide polymorphisms (SNPs) in exon 7. Although, many varieties are reported to be fragrant without having these known mutations, suggesting the involvement of other genes/mutations in rice. It is need of the day to identify the multiple mutations sites in fgr/bdh2 gene for 2AP accumulation and characterize the fragrance in a wide range of genetic resources of rice available for this trait. In this study, loss of function mutations of badh2 genes were characterized in aromatic and non-aromatic rice varieties of Pakistan to resolve the ambiguities linked to the genetic basis of fragrance along with quantification of 2 AP through GC-MS. Genetic analysis revealed that many aromatic varieties exhibited 8-bp deletion and 3 SNPs with strong accumulation of 2-AP. However, amount of 2 AP is highly variable among those varieties indicating involvement of additional factors controlling the intensity of aroma. Moreover, badh2 gene was found to be intact in non-aromatic varieties with no AP accumulation. Thus, it is suggested that this mutation is not only responsible for the 2AP synthesis and accumulation but there are other genetic factors for controlling the development of 2 AP metabolism pathways which are needed to explore. Such investigations will be very important in developing molecular-assisted breeding of aromatic rice with multiple genetic sources of 2AP leading to development of more aromatic varieties.

Key Words:

Badh2 gene, fragrance, loss of function mutation, aromatic rice, 2 AP accumulation

Poster G2. Evaluation of Genomic Selection as a Tool for Pea Breeding

Carpenter, M.A.[1], Goulden, D.S.[1], Thomson, S.J.[1], Woods, C.J.[1], Alspach, P.A.[2], Kenel, F.O.[1], Frew, T.J.[1], Cooper, R.D.[1], Timmerman-Vaughan, G.M.[1]
The New Zealand Institute for Plant & Food Research Limited, [1]Lincoln, NZ, [2]Motueka, NZ.

The pea breeding programme at Plant & Food Research (PFR) uses marker assisted selection (MAS) in breeding for resistance to diseases such as powdery mildew, pea seed-borne mosaic virus and pea enation mosaic virus. This is a practical approach where disease resistance is a single gene trait, but not for diseases such as ascochyta blight, where susceptibility is polygenic. A solution may be found in genomic selection (GS), a promising breeding tool that uses genome-wide molecular marker data to predict phenotypic values and has potential to accelerate breeding for polygenic traits.
A training population of 205 individuals from the PFR pea breeding programme and germplasm collection has been genotyped using Genotyping by Sequencing (GBS) and phenotyped for three traits: MYC (ascochyta blight susceptibility), DTF (days to flowering) and TSWT (thousand seed weight). DTF and TSWT were determined from a single field trial whereas MYC was determined from two replicated randomised field trials in 2013 and 2015. Genomic selection analyses have been conducted using the R package BGLR (Bayesian Generalized Linear Regression) to explore several different models (GBLUP, Bayes A, Bayes B, Bayes C, BRR, BL and RKHS) and various parameters. These comparisons were done via cross-validation using replicated randomised testing sets of 50 lines. Overall the mean prediction accuracy did not vary much between the models. However, different testing/training partitions gave very different results such that the distribution of the prediction accuracies for each model is wide. Typical results for mean prediction accuracy were 0.4-0.5 for MYC and 0.6 for DTF and TSWT. The genotype data have also been used in a GWAS analysis of the three traits, which revealed several SNPs putatively associated with the traits. MYC was associated with SNPs in linkage groups 4 and 7, TSWT with linkage groups 1 and 5, DTF with linkage group 5.

Poster G3. QTL Mapping of Durable Stripe Rust Resistance in Wheat

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Durable disease resistance is a major target for plant breeders. The hexaploid wheat cultivar 'Monad' was released in New Zealand in 1993 and has remained resistant to stripe rust (*Puccinia striiformis* f. sp. *tritici*) for more than 20 years.

In order to understand the genetic basis for this resistance, a doubled haploid population was created from a cross between 'Monad' and the susceptible cultivar 'Tiritea'. The population of 219 lines was screened for response to natural infections of stripe rust and phenotyped for seed yield traits both with and without fungicide control. Unprotected plots of 'Monad' still achieved more than 94% of the fungicide controlled values for grain number, head weight and seed weight. In contrast, unprotected plots of 'Tiritea' dropped to 72%, 33% and 45% of their respective fungicide controlled values.

Genotyping was performed using the Wheat Illumina Infinium 90k SNP chip[1] and a genetic linkage map was constructed using the R package 'ASMap' from 8,107 polymorphic SNP markers.

Quantitative trait locus (QTL) mapping was performed using the R package 'qtl' and identified a large effect QTL for stripe rust resistance near the central region of chromosome 2B. This region is known to harbour the resistance genes *Yr5*, *Yr44*, and *Yr53*. An additional QTL for stripe rust resistance was also located on chromosome 7AL, but was only significant for two of the four disease severity scores over 2 years.

Identification of these QTL will allow this resistance to be tracked more easily into elite wheat breeding lines and help to maintain high levels of durable stripe rust resistance.

1. Wang, S., Wong, D., Forrest, K. et al. (2014) *Characterization of polyploid wheat genomic diversity using a high-density 90 000 single nucleotide polymorphism array*. Plant Biotechnology Jounral 12:787-796.

Poster G4. Genetic correlation between interleukin 23 receptor (*IL23R)* three variants (*rs7517847*, *rs11209026* and *rs11465804*) and gout susceptibility in a New Zealand population

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Gout is an auto-inflammatory disorder caused by deposition of monosodium urate (MSU) crystals in and around tissues in the presence of hyperuricemia. However, there are other factors that control progression from hyperuricemia to gout. The interleukin 23 receptor (*IL23R*) plays a role in promoting inflammation. An earlier study associated the *IL23R* variant *rs7517847* G-allele with the risk of gout in a Han Chinese population, [1] while others reported association of variants *rs11209026* and *rs11465804* with crohn's disease (CD) in Dutch cohort and CD and ulcerative colitis (UC) in Jewish and non-Jewish populations, respectively. [(2,3)] The aim of our study was to test for association between three *IL23R* polymorphisms (*rs7517847*, *rs11209026* and *rs11465804*) and gout in New Zealand (NZ) populations. A total of clinically-ascertained 2604 gout cases[4] and 2092 controls of European and New Zealand Māori and Pacific (Polynesian) ancestry were utilised. Taqman® genotyping of *IL23R* polymorphisms was carried out, followed by multivariate-adjusted association analysis in R 3.2.2 with gout as the outcome.

The minor G-allele of *rs7517847* showed a significant protective association with gout in the Western Polynesian group (OR=0.74, P_{OR}=0.029). However, the other *IL23R* variants were not associated with gout in any NZ population. A significant association was also found between the G-allele of *rs7517847* and the risk of gout in Polynesian males (OR=0.83, P_{OR}=0.03).

In conclusion, our study indicates association of the *IL23R* variant *rs7517847* G-allele with gout in the NZ Western Polynesian population, replicating the Han Chinese finding[1]. However, other variants (*rs11209026* and *rs11465804*) did not exhibit association with gout susceptibility in the European or Polynesian populations. This suggests that the *IL23R rs7517847* variant may be involved in regulating the immune-mediated inflammatory response in gout pathogenesis in Western Polynesians.

1. Liu, S., He, H., Yu, R., Han, L., Wang, C. and Cui, Y., et al (2015). *The rs7517847 polymorphism in the IL-23R gene is associated with gout in a Chinese Han male population*. Modern Rheumatology. 25(3):449-52.

2. Weersma, R.K., Zhernakova, A., Nolte, I.M., Lefebvre, C., Rioux, J.D. and Mulder, F., et al (2008). *ATG16L1 and IL23R are associated with inflammatory bowel diseases but not with celiac disease in the Netherlands*. The American journal of gastroenterology. 103(3):621-7.

3. Duerr, R.H., Taylor, K.D., Brant, S.R., Rioux, J.D., Silverberg, M.S. and Daly, M.J., et al (2006). *A genome-wide association study identifies IL23R as an inflammatory bowel disease gene*. science. 314(5804):1461-3.

4. Wallace, S.L,. Robinson, H., Masi, A.T., Decker, J.L., McCarty, D.J. and Yu, T.F. (1977). *Preliminary criteria for the classification of the acute arthritis of primary gout*. Arthritis Rheum. 20:895–900.

Poster G5. Genetic association of inflammatory gene *PPARGC1B* with gouty arthritis in a New Zealand population

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Gout is an auto-inflammatory arthritis caused by deposition of crystallized monosodium urate in and around tissues. Elevated serum urate levels (hyperuricemia) trigger the formation of monosodium urate crystals (MSU). The genetic basis of hyperuricemia is increasingly well-characterised, however, the genetic basis of the innate immune-mediated inflammatory response in gout pathogenesis is still unclear. Previously, Chang et al (2016) reported association of the PPARGC1B missense variant *rs45520937* A-allele with gout susceptibility in a Taiwan Han Chinese population.

Our aim was to replicate this finding in a total of 2680 clinically ascertained gout cases[1] and 2195 controls from the New Zealand Polynesian (Māori and Pacific) population, and Europeans from New Zealand and Europe. Taqman® genotyping of PPARGC1B missense variant *rs45520937* was undertaken, followed by multivariate-adjusted association analysis in R 3.2.2 with gout as outcome.

We found a significant association of the PPARGC1B *rs45520937* A-allele with gout risk in the Polynesian sample set (OR= 1.17, P=0.02*) but not in Europeans (OR= 0.96, P=0.80). A stronger association was observed in males of higher Polynesian ancestry (OR= 1.47, P_{OR} =0.01*).

Our findings support a potential role of the inflammatory mediator PPARGC1B in the pathogenesis of gout.

References

1. Wallace, S.L., Robinson, H., Masi, A.T., Decker, J.L., McCarty D.J., Yu, T.F (1977) *Preliminary criteria for the classification of the acute arthritis of primary gout*. Arthritis Rheum 20:895–900.

Poster G6. Open Source, Open Data and Open Science Policy: Opportunities for New Zealand Genomics Research

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Genomics research is underpinned by Free / Libre and Open Source software – from the operating system level to the individual tools used to analyze data. Genomic scientists contribute to large open respositories of sequence data. The global move to open science (which includes transparancy, reproducibility and availability) is gaining traction in New Zealand public policy. The New Zealand Goverment Open Access and Licensing (NZGOAL) framework and soon the Software Extension to that framework (NZGOAL-SE) apply to all Crown Entities.

Getting beyond the buzzwords we will discuss the benefits of working in an open fashion with international examples appropriate to our MapNet community. We will describe the process by which the Software Extension was developed openly and collaboratively this year. We will also present a recap of the efforts of the Biospectra-by-Sequencing project to work openly and collaboratively for Nzinc., giving an update on the status of the project and plans for the future.

Poster G7. New molecular tools for interspecific hybridization of *Gentiana* sp.

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There are over 400 species of gentians, of which only a few are of economic importance as ornamental crops. Hot pink, red, purple and yellow flowered gentians are among the new genotypes and cultivars bred and developed by The New Zealand Institute for Plant & Food Research Limited (PFR). Developed from wide crosses among various *Gentiana* spp. these hybrids offer the New Zealand and international flower industry new options: different flower colours and forms tap into international demand for novel cut flowers, potted and landscape plants. Within the PFR breeding programme, new molecular tools are being developed and applied; for example, to confirm hybrid status of seedlings generated. Our progress in developing these tools is presented here.

Preliminary studies characterising *Gentiana* spp. using Sequence Characterized Amplified Regions (SCAR) markers, produced some usable assays. One SCAR primer set was identified to be specific for *G. mirandae* and another for *G. scabra*. Subsequent tests across a small number of hybrids verified that such molecular tools can be used to differentiate individuals within these *Gentiana* species. We anticipate further tests using publicly available chloroplastic markers and the High Resolution Melting (HRM) technique as well as microsatellite markers resolved using capillary electrophoresis will help to discriminate among *G. dinarica*, *G. angustifolia 'Alba', G. wilsonii, G. parryi, G. acaulis, G. freyniana, G. przewalskii, G. sino-ornata, G. paradoxa, G. lutea, G. mirandae, G. triflora, G scabra, G. septemfida* and their hybrids.

Poster G8. Genomics in New Zealand Forestry

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Tree breeding in New Zealand has initiated the move to genomics. Tree breeding programmes in NZ are currently all with exotic species, with recorded pedigree depths of between two and four generations from unimproved. The combination of well-structured populations and the ability to clonally test across a number of environments is an advantage in these programmes. However, the limited number of generations means that these populations are still highly heterogeneous. Furthermore, most experimental trials are planted with wind-pollinated trees and have long generation times, both of which present a significant challenge to breeding programmes.

Genomics can help address many of the challenges faced by tree breeders: providing marker-based relationships in wind-pollinated populations where only one parent is known, selecting for wood quality traits that are very expensive to measure, and accelerating the rate of delivery of genetic gain. The Radiata Pine Breeding Company, in partnership with Scion, is leading genomics research for radiata pine, and have developed a 44K exome capture panel to enable the implementation of genomic selection. The development of such extensive genomics resources has historically been unachievable for smaller breeding programmes, such as those for eucalypts and Douglas-fir. However, recent years have seen internationally-developed high density SNP arrays become available for these species {Howe, 2013 #1990;Silva-Junior, 2015 #2383}, which is enabling a paradigm shift in our approach to breeding.

We report on the progress being made towards the introduction of genomics and genomics research in radiata pine, Douglas-fir and in a number of eucalypt breeding programmes. We present early results from the existing multi-species *Eucalyptus* SNP chip EuCHIP60K that has been used in the first application of genomics to a eucalypt breeding programme in NZ. We also report on our plans to effectively incorporate genomics for a number of other forest-based species.

1. Howe, G.T., et al., *A SNP resource for Douglas-fir: De novo transcriptome assembly and SNP detection and validation.* BMC Genomics, 2013: p. 137.
2. Silva-Junior, O.B., D.A. Faria, and D. Grattapaglia, *A flexible multi-species genome-wide 60K SNP chip developed from pooled resequencing of 240 Eucalyptus tree genomes across 12 species.* New Phytol, 2015. **206**(4): p. 1527-40.

Summary of Abstracts for the Poster Session Template

No.	Title	Presenter	Institutions
G1	Screening loss of function mutations in fgr/badh2 gene for conferring fragrance in rice varieties of Pakistan	Saddia Galani, Shagufta Sahar, Abid Azhar	Karachi Institute of Biotechnology and Genetic Engineering (KIBGE) University of Karachi, Karachi, Pakistan
G2	Evaluation of Genomic Selection as a Tool for Pea Breeding	Carpenter, M.A.[1], Goulden, D.S.[1], Thomson, S.J.[1], Woods, C.J.[1], Alspach, P.A.[2], Kenel, F.O.[1], Frew, T.J.[1], Cooper, R.D.[1], Timmerman-Vaughan, G.M.[1]	The New Zealand Institute for Plant & Food Research Limited, [1]Lincoln, NZ, [2]Motueka, NZ.
G3	QTL Mapping of Durable Stripe Rust Resistance in Wheat	Paul A. Johnston[1], Catherine Munro[1], Matthew Cromey[2], Soonie Chng[1], Vijitha Meiyalaghan[1], Merle Forbes[1], Steve Shorter[3]	[1]The New Zealand Institute for Plant & Food Research Limited, NZ [2]The Royal Horticultural Society, UK [3]PGG Wrightson Ltd, NZ
G4	Genetic correlation between interleukin 23 receptor (*IL23R)* three variants (*rs7517847*, *rs11209026* and *rs11465804*) and gout susceptibility in a New Zealand population	Shaukat, A.[1], Leaupepe, K.[1], Phipps-Green, A.[1], Dalbeth, N.[2], Stamp, L.[3], Hindmarsh, J.H.[4] and Merriman, T.[1]	[1]Department of Biochemistry, University of Otago, NZ [2]Department of Medicine, University of Auckland, NZ [3]Department of Medicine, University of Otago, NZ [4]Ngati Porou Hauora Charitable Trust, NZ
G5	Genetic association of inflammatory gene *PPARGC1B* with gouty arthritis in a New Zealand population	Shaukat, A.[1], Phipps-Green, A.[1], Dalbeth, N.[2], Stamp. L.[3], Hindmarsh, J.H.[4]	[1]Department of Biochemistry, University of Otago, NZ

		and Merriman, T.[1]	[2]Department of Medicine, University of Auckland, NZ [3]Department of Medicine, University of Otago, NZ [2]Ngati Porou Hauora Charitable Trust, NZ
G6	Open Source, Open Data and Open Science Policy: Opportunities for New Zealand Genomics Research	Elshire, R.J. [1]	[1]The Elshire Group Limited, Palmerston North, NZ
G7	New molecular tools for interspecific hybridization of *Gentiana* sp.	Tonya Frew[1], Claudia Wiedow[2], Gail Timmerman-Vaughan[1], Ryohei Kaji [2], David Chagne[2], Ed Morgan[2], Keith Funnell[2]	[1] The New Zealand Institute for **Plant** & **Food Research** Limited, Christchurch, NZ [2] The New Zealand Institute for **Plant** & **Food Research** Limited, Palmerston North, NZ
G8	Genomics in New Zealand Forestry	Dungey, H.[1], Telfer, E.[1], Graham, N.[1], Shearer, A.[1,2], Li, Y[1], Klapste, J[1], Murray, M.[1,3], Macdonald, L.[1]	[1] Forest Genetics, Scion, Rotorua, NZ [2]University of Canterbury, Christchurch, NZ [3] University of Waikato, Hamilton, NZ