Abstracts for the 36th Human Genetics Society of Australasia Annual Scientific Meeting Canberra, Australia
July 22–25, 2012

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*This information was correct at the time of printing and is subject to change.

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Kate	Dunlop	Providing Written Information for Consumers on Rare Genetic Conditions - Attitudes Towards and the Use of Genetics Information Sheets	Poster Display
Kate	Dunlop	Evaluation of a General Practitioner Education Module on First Trimester Screening and the Associated Genetic Counselling Skills	Poster Display
Richard	Dunstan	Primary Ciliary Dyskinesia (PCD) Causing Idiopathic Tachypnoea in Newborn	Poster Display
Desiree	DuSart	Population-Based Carrier Screening for Cystic Fibrosis in Victoria: The Past 6 Years Experience	Oral Presentation
Matthew	Edwards	Molybdenum Cofactor Deficiency due to MOCS2 Mutation	Poster Display
Matthew	Edwards	Primary Ciliary Dyskinesia (PCD) Causing Idiopathic Tachypnoea in Newborn	Poster Display
Matthew	Edwards	The Importance of Confirming CFTR Mutation Genotypes for Affected Individuals and the Significance of This Information for Extended Family Members, Clinical Care and Future Reproductive Decision Making	Poster Display
Maha	Eid	In Vitro Study of the Anti Oxidant Effect of Astaxanthin and Vitamin E on Cultured Lymphocyts from Fanconi Anemia Patients	Poster Display
Stewart	Einfeld	Changing Clinical Interpretation of Chromosomal Microarrays Over Time	Oral Presentation
Carolyn	Ellaway	OTC - Energy and Protein Requirements of a 2 Year Old Noumean Girl Presenting with Unstable Ammonium Levels	Poster Display
Carolyn	Ellaway	Liver Transplantation from an Adult Donor with an Unrecognized Urea Cycle Disorder to a Paediatric and an Adult Recipient	Oral Presentation
Carolyn	Ellaway	Deletion of a Novel Phosphatase Results in a Rett Syndrome-Like Phenotype	Oral Presentation
Aoife	Elliott	The Effect of Continuous Haemodiafiltration and Intermittent Haemodialysis on Plasma Amino Acid and Ammonia Profiles for the Metabolic Patient	Poster Display
E	Elliott	Inborn Errors of Metabolism: Significant Impacts on Families and Health Services	Oral Presentation
Kerry	Emslie	The Differences Between Discovery and Diagnostic DNA Methylation Measurements May Impede Clinical Application	Oral Presentation

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First Name	Last Name	Title	Presentation Type
M.L	Engvall	Adenosine Kinase Deficiency, a Novel Cause of Hypermethioninemia Presenting as a Slowly Progressive Encephalopathy with Epilepsy, Myopathy and Liver Disease	Oral Presentation
Anoop	Enjeti	When Size Does Matter - Little Things Mean a Lot	Poster Display
John	Entwistle	Epilepsy with Cognitive Deficit and Autism Spectrum Disorders: Prospective Diagnosis by Array CGH	Oral Presentation
Jamie	Errico	Propionic Acidaemia, Nephrotic Syndrome and Ischaemic Gut: a Nutritional Challenge	Oral Presentation
Jane	Estrella	OI Type III-FKBP10 in Non-Consanguineous Australian Families	Oral Presentation
Tiffany-Jane	Evans	SPN Array Technology Identifies Novel Candidate Loci for Hnpcc/Lynch Syndrome	Oral Presentation
Kerry	Fagan	Fish Reveals Hidden Complexity of Copy Number Variants	Oral Presentation
Peter	Farlie	Mouse Models of Human Skeletal Dysmorphologies	Oral Presentation
Susan	Fawcett	Decision Making Following Genetic Counselling for Second Trimester Ultrasound Marker for Aneuploidy - A Review of Outcomes at the Royal Women's Hospital Genetic Counselling Service, Melbourne, Australia	Poster Display
Frank	Ferman	Childhood Spastic Diplegia in a Novel Form of Non-Ketotic Hyperglycinaemia, Associated with Pyruvate Dehydrogenase Complex Deficiency, Caused by Glrx5 Deficiency of the Iron-Sulfur Cluster	Oral Presentation
I.	Ferrero	Identification and Characterization of New Mitochondrial Disease Genes	Oral Presentation
Michael	Field	Postnatal Mothers Attitudes to Newborn Screening for Fragile X Syndrome	Oral Presentation
Peter	Field	Numerical and Structural Abnormalities of the Y Chromosome in Infertility Investigations	Oral Presentation
Michael	Field	The Australian Familial Pancreatic Cancer Cohort: Screening for Pancreatic Cancer in High Risk Australians	Poster Display
Peter	Field	Offering CFTR-PGD Without a Delay for Work Up; Reality or Fiction?	Oral Presentation
Peter	Field	FMR1 Gene Small Triplet Repeat Alleles Associated with POF/POI and Early Menopause	Oral Presentation
Michael	Fietz	Niemann-Pick Disease Type C Caused By Maternal Uniparental Isodisomy of Chromosome 18	Oral Presentation
Michael	Fietz	Unusual Radiographic Features of a 9 Year Old Girl with Attenuated Mucopolysaccharidosis Type VI	Poster Display
Maree	Flaherty	Genome-Wide Analysis Using Next-Generation Sequencing For Disease-Gene Identification In Genetically Heterogeneous Eye Diseases	Oral Presentation
Maree	Flaherty	PAX6 Mutation Spectrum in an Australian and New Zealand Aniridia Cohort	Oral Presentation
Janice	Fletcher	Obesity in Adult Patients With Phenylketonuria	Poster Display
Robin	Forbes	The Experiences of Parents of Children with Developmental Delay, Autism or Intellectual Disability Who Receive an FMR1 Grey Zone Result	Oral Presentation
Michael	Forbes-Smith	The Differences Between Discovery and Diagnostic DNA Methylation Measurements May Impede Clinical Application	Oral Presentation
Rachel	Fraser	SNP-Arrays for the Study of Plasma cell Myeloma	Oral Presentation
Rebecca	Freedman	Receiving Enzyme Replacement Therapy for a Lysosomal Storage Disorder; Exploring the Experiences of Young Patients and Their Families	Oral Presentation
Michael	Freelander	Primary Ciliary Dyskinesia (PCD) Causing Idiopathic Tachypnoea in Newborn	Poster Display
Hudson	Freeze	An Additional Chime Syndrome Case Due to Compound Heterozygous Mutations in PIGL	Poster Display
Marisa	Friedrich	Childhood Spastic Diplegia in a Novel Form of Non-Ketotic Hyperglycinaemia, Associated with Pyruvate Dehydrogenase Complex Deficiency, Caused by Glrx5 Deficiency of the Iron-Sulfur Cluster	Oral Presentation
Kathryn	Friend	A Family With an ARG134CYS Mutation In COl1A1 and Overlapping Phenotypes of Oteogenesis Imperfecta and Ehlers-Danlos Syndrome	Poster Display
Kathie	Friend	Sequence Analysis oftThe Nemo Gene in a Cohort of Patients Without the Common Nemo?4–10 Deletion	Poster Display
Shuang	Fu	The Differences Between Discovery and Diagnostic DNA Methylation Measurements May Impede Clinical Application	Oral Presentation
Lydia	Gaffney	Decision Making Following Genetic Counselling for Second Trimester Ultrasound Marker for Aneuploidy - A Review of Outcomes at the Royal Women's Hospital Genetic Counselling Service, Melbourne, Australia	Poster Display
Devika	Ganesamoorthy	A Genetic Approach to the Problem of Monitoring Kidney Transplant Rejection	Oral Presentation
A	Gardner	Mutations in PRRT Cause Benign Familial Infantile Epilepsy and Infantile Convulsions and Choreoathetosis Syndrome	Oral Presentation
Don	Garrett	FMR1 Gene Small Triplet Repeat Alleles Associated with POF/POI and Early Menopause	Oral Presentation

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First Name	Last Name	Title	Presentation Type
Dave	Garrick	Genome-wide Analysis of Sequence Variation Underlying Tissue-Specific Transcription Factor Binding and Gene Expression	Oral Presentation
J	Gecz	Mutation in STXBP5L Associated with an Early Onset Neurodegenerative Disorder	Oral Presentation
J	Gécz	Mutations in PRRT Cause Benign Familial Infantile Epilepsy and Infantile Convulsions and Choreoathetosis Syndrome	Oral Presentation
D	Ghezzi	Identification and Characterization of New Mitochondrial Disease Genes	Oral Presentation
Richard	Gibbons	Genome-wide Analysis of Sequence Variation Underlying Tissue-Specific Transcription Factor Binding and Gene Expression	Oral Presentation
Joanne	Gibson	Mitochondrial Defects In RETT Syndrome	Oral Presentation
S	Gijsen	Mutation in STXBP5L Associated with an Early Onset Neurodegenerative Disorder	Oral Presentation
C	Gilissen	Pitfalls and Successes of Exome Sequencing: Mutations in a Novel Gene Cause Walker-Warburg Syndrome And Defective Glycosylation Of a-Dystrgoglycan	Oral Presentation
D	Gill	Childhood Spastic Diplegia in a Novel Form of Non-Ketotic Hyperglycinaemia, Associated with Pyruvate Dehydrogenase Complex Deficiency, Caused by Glrx5 Deficiency of the Iron-Sulfur Cluster	Oral Presentation
Lynn	Gillam	Genetic Carrier Testing in Healthy Siblings: Investigation of Current Practice and Health Professionals' Views	Oral Presentation
Emma	Glamuzina	Two Extremes of Ornithine Transcarbamylase Deficiency in Males	Oral Presentation
Himanshu	Goel	Postnatal Mothers Attitudes to Newborn Screening for Fragile X Syndrome	Oral Presentation
Wendy	Gold	Whole Exome Sequencing Identifies a Novel Missense Mutation in Two Affected Children with a Suspected Rare Mendelian Disorder	Oral Presentation
Wendy	Gold	Mitochondrial Defects In RETT Syndrome	Oral Presentation
Wendy	Gold	HDAC6 Inhibitors: A Novel Therapy for RETT Syndrome	Poster Display
H	Goldberg-Stern	Mutations in PRRT Cause Benign Familial Infantile Epilepsy and Infantile Convulsions and Choreoathetosis Syndrome	Oral Presentation
Jack	Goldblatt	An Exploratory Dysmorphometric Analysis to Define 3d Facial Phenotypic Signatures as a Foundation for Non-Invasive Monitoring of Lysosomal Storage Disorders	Oral Presentation
Leena	Gole	Case Study: Characterisation and Implications of Ring Chromosome 21 in a Female Carrier	Poster Display
A	Goodwin	The Australian Familial Pancreatic Cancer Cohort: Screening for Pancreatic Cancer in High Risk Australians	Poster Display
Christopher	Gordon	Mouse Models of Human Skeletal Dysmorphologies	Oral Presentation
Lisa	Gordon	Decision Making Following Genetic Counselling for Second Trimester Ultrasound Marker for Aneuploidy - A Review of Outcomes at the Royal Women's Hospital Genetic Counselling Service, Melbourne, Australia	Poster Display
Neil	Granter	When Size Does Matter - Little Things Mean a Lot	Poster Display
Padriac	Grattan-Smith	14q13 Microdeletion: a Recognizable Contiguous Gene Syndrome Encompassing Brain/Thyroid/Lung and Dental Phenotype	Oral Presentation
Stephanie	Grehan	Numerical and Structural Abnormalities of the Y Chromosome in Infertility Investigations	Oral Presentation
Desma	Grice	SPN Array Technology Identifies Novel Candidate Loci for Hnpcc/Lynch Syndrome	Oral Presentation
Adrian	Griffiths	Haemoglobin E and Co-Existing Alpha Thalassaemia in the South Australian Population	Poster Display
Lyn	Griffiths	Molecular Genetics of Migraine; Implications for Therapeutic Development	Oral Presentation
John	Grigg	Genome-Wide Analysis Using Next-Generation Sequencing For Disease-Gene Identification In Genetically Heterogeneous Eye Diseases	Oral Presentation
John	Grigg	PAX6 Mutation Spectrum in an Australian and New Zealand Aniridia Cohort	Oral Presentation
Sean	Grimmond	The Australian Familial Pancreatic Cancer Cohort: Screening for Pancreatic Cancer in High Risk Australians	Poster Display
Bronwyn	Grinton	Mutations in PRRT Cause Benign Familial Infantile Epilepsy and Infantile Convulsions and Choreoathetosis Syndrome	Oral Presentation
B	Grisart	Pitfalls and Successes of Exome Sequencing: Mutations in a Novel Gene Cause Walker-Warburg Syndrome And Defective Glycosylation Of a-Dystrgoglycan	Oral Presentation
Alexandra	Groves	An Exploration of the Clinical Experiences and Attitudes of Australian Health Professionals in Regard to Direct-To-Consumer (Dtc) Genetic Testing	Poster Display
Sachin	Gupta	Childhood Spastic Diplegia in a Novel Form of Non-Ketotic Hyperglycinaemia, Associated with Pyruvate Dehydrogenase Complex Deficiency, Caused by Glrx5 Deficiency of the Iron-Sulfur Cluster	Oral Presentation
T.	Haack	Identification and Characterization of New Mitochondrial Disease Genes	Oral Presentation

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First Name	Last Name	Title	Presentation Type
E	Haan	Mutation in STXBP5L Associated with an Early Onset Neurodegenerative Disorder	Oral Presentation
Eric	Haan	Epilepsy with Cognitive Deficit and Autism Spectrum Disorders: Prospective Diagnosis by Array CGH	Oral Presentation
E	Haan	Mutations in PRRT Cause Benign Familial Infantile Epilepsy and Infantile Convulsions and Choreoathetosis Syndrome	Oral Presentation
Eric	Haan	Sequence Analysis oftThe Nemo Gene in a Cohort of Patients Without the Common Nemo?4–10 Deletion	Poster Display
M	Habord	Mutation in STXBP5L Associated with an Early Onset Neurodegenerative Disorder	Oral Presentation
Andrew	Hallahan	5 Year Follow Up of 2 Siblings with Krabbe Disease Transplanted at 4 and 5 Weeks of Age Following Postmortem Diagnosis in a Sibling	Oral Presentation
Jane	Halliday	Implementing a Two-Stage Written Consent Process for Newborn Screening in Victoria	Oral Presentation
Jane	Halliday	International Sarcoma Kindred Study (ISKS) - Knowledge of Genetics Amongst Patients, Families and Health Professionals	Oral Presentation
Jane	Halliday	Public Health Genomics: a New Core Subject for the University Of Melbourne, Master Of Genetic Counselling	Poster Display
Jane	Halliday	Should we be Offering Women a Choice of the types of Chromosome Conditions they want Prenatally Diagnosed?	Oral Presentation
Jessica	Hansen	Postnatal Mothers Attitudes to Newborn Screening for Fragile X Syndrome	Oral Presentation
Michael	Harbord	Epilepsy with Cognitive Deficit and Autism Spectrum Disorders: Prospective Diagnosis by Array CGH	Oral Presentation
Iain	Hargraves	Mitochondrial Defects In RETT Syndrome	Oral Presentation
James	Harraway	Genetic Testing for Maturity Onset Diabetes of the Young in Australia	Oral Presentation
James	Harraway	Detection of Gene Mutations Causing Noonan Syndrome: Two Case Studies	Oral Presentation
Mark	Harris	Genetic Testing for Maturity Onset Diabetes of the Young in Australia	Oral Presentation
Emma	Harrison	Challenges Faced by Health Care Interpreters Practicing in Genetic Counselling Services	Poster Display
John	Hawley	From Mitochondria to Movement: The Genetics of Athletic Performance	Oral Presentation
John	Hawley	Skeletal Muscle: The Locus of Control for Metabolic Health	Oral Presentation
Janette	Hayward	Family Testing For a Microdeletion Found on Microarray CGH in a Prenatal Setting	Poster Display
Luke	Heaps	PAX6 Mutation Spectrum in an Australian and New Zealand Aniridia Cohort	Oral Presentation
Milton T.W.	Hearn	Signature Peptides as Biomarkers for the Identification of Haemoglobinopathies	Poster Display
William	Heath	Proteomic and Metabolic Analysis of a Mitochondrial Complex I Deficiency Mouse Model Obtained by Retroviral Insertion In The NDUFS4 Gene	Oral Presentation
Eliana	Henao	Pooling/Bootstrap-based GWAS (pbGWAS) Identifies New Loci Modifying the Age of Onset in PSEN1 E280A Alzheimer's Disease	Poster Display
E.	Herlenius	Mutations in PRRT Cause Benign Familial Infantile Epilepsy and Infantile Convulsions and Choreoathetosis Syndrome	Oral Presentation
Amy	Herlihy	International Sarcoma Kindred Study (ISKS) - Knowledge of Genetics Amongst Patients, Families and Health Professionals	Oral Presentation
Amy	Herlihy	Public Health Genomics: a New Core Subject for the University Of Melbourne, Master Of Genetic Counselling	Poster Display
Sarah	Heron	Mutations in PRRT Cause Benign Familial Infantile Epilepsy and Infantile Convulsions and Choreoathetosis Syndrome	Oral Presentation
Honey	Heussler	Complex IV Deficiency in a Girl with 22Q11.2 Deletion and Adjacent Segmental Uniparental Disomy	Poster Display
Chelsee	Hewitt	Proteomic and Metabolic Analysis of a Mitochondrial Complex I Deficiency Mouse Model Obtained by Retroviral Insertion In The NDUFS4 Gene	Oral Presentation
Doug	Higgs	Genome-wide Analysis of Sequence Variation Underlying Tissue-Specific Transcription Factor Binding and Gene Expression	Oral Presentation
Gladys	Ho	Complementation between Specific Phenylalanine Hydroxylase (Pah) Missense Mutations and Correlation to Clinical Phenotype in Phenylketonuria (PKU)	Oral Presentation
Bree	Hodgson	Mutations in PRRT Cause Benign Familial Infantile Epilepsy and Infantile Convulsions and Choreoathetosis Syndrome	Oral Presentation
A	Hoischen	Mutation in STXBP5L Associated with an Early Onset Neurodegenerative Disorder	Oral Presentation
Elizabeth	Holiday	SPN Array Technology Identifies Novel Candidate Loci for Hnpcc/Lynch Syndrome	Oral Presentation
Lucy	Holland	Developing a Youth Friendly Model of Genetic Counselling	Oral Presentation
Mark	Holloway	Haemoglobin E and Co-Existing Alpha Thalassaemia in the South Australian Population	Poster Display

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First Name	Last Name	Title	Presentation Type
Noemi	Horvath	SNP-Arrays for the Study of Plasma cell Myeloma	Oral Presentation
Louanne	Hudgins	Prenatal Diagnosis of Rasopathies	Oral Presentation
Louanne	Hudgins	Counselling Issues Related to Prenatal Diagnosis / Cell Free Fetal DNA (Please note the Session Details are to be Confirmed and are Subject to Change)	Oral Presentation
Louanne	Hudgins	Noninvasive Prenatal Testing: How Far do we go and What do our Patients Really Want?	Oral Presentation
Jim	Hughes	Genome-wide Analysis of Sequence Variation Underlying Tissue-Specific Transcription Factor Binding and Gene Expression	Oral Presentation
Yvonne	Hull	Epilepsy with Cognitive Deficit and Autism Spectrum Disorders: Prospective Diagnosis by Array CGH	Oral Presentation
Lynn	Hulston	Adventures in Prenatal Testing	Oral Presentation
Maureen	Humphrey	Propionic Acidaemia, Nephrotic Syndrome and Ischaemic Gut: a Nutritional Challenge	Oral Presentation
J	Humphries	The Australian Familial Pancreatic Cancer Cohort: Screening for Pancreatic Cancer in High Risk Australians	Poster Display
Lauren	Hunt	Psychological Impairment Particularly in Mothers Following Sudden Cardiac Death in the Young	Oral Presentation
Lauren	Hunt	Utility of Genetic Testing in Hypertrophic Cardiomyopathy in Queensland's Statewide Cardiac Genetics Clinic	Poster Display
Matthew	Hunter	Postnatal Mothers Attitudes to Newborn Screening for Fragile X Syndrome	Oral Presentation
Val J	Hyland	Genotype/Phenotype Classification of a Family with a Novel Missense Mutation in NOG, Surgical Implications for Stapedectomy	Poster Display
Frank	Ierino	A Genetic Approach to the Problem of Monitoring Kidney Transplant Rejection	Oral Presentation
Jodie	Ingles	Psychological Impairment Particularly in Mothers Following Sudden Cardiac Death in the Young	Oral Presentation
Jodie	Ingles	Case Presentation - 'Getting Over the Shock of it' the Impact of Implantable Defibrillators'	Oral Presentation
Jodie	Ingles	The Australian National Genetic Heart Disease Registry	Oral Presentation
Anita	Inwood	5 Year Follow Up of 2 Siblings with Krabbe Disease Transplanted at 4 and 5 Weeks of Age Following Postmortem Diagnosis in a Sibling	Oral Presentation
Anita	Inwood	Unusual Radiographic Features of a 9 Year Old Girl with Attenuated Mucopolysaccharidosis Type VI	Poster Display
Liane	Ioannou	No Thanks' - Why Pregnant Women Choose not to Have Cystic Fibrosis Carrier Screening	Poster Display
Xenia	Iona	Mutations in PRRT Cause Benign Familial Infantile Epilepsy and Infantile Convulsions and Choreoathetosis Syndrome	Oral Presentation
Joanne	Isbister	Decision Making Following Genetic Counselling for Second Trimester Ultrasound Marker for Aneuploidy - A Review of Outcomes at the Royal Women's Hospital Genetic Counselling Service, Melbourne, Australia	Poster Display
Robyn	Jamieson	Genome-Wide Analysis Using Next-Generation Sequencing For Disease-Gene Identification In Genetically Heterogeneous Eye Diseases	Oral Presentation
Robyn	Jamieson	PAX6 Mutation Spectrum in an Australian and New Zealand Aniridia Cohort	Oral Presentation
Gemma	Jenkins	Niemann-Pick Disease Type C Caused By Maternal Uniparental Isodisomy of Chromosome 18	Oral Presentation
David	Jessup	TBA (Abstract Not Received at Time of Printing)	Oral Presentation
David	Jessup	Panel Discussion on Cascade Testing for Cystic Fibrosis - the Family Perspective (Abstract Not Received at Time of Printing)	Oral Presentation
Amber	Johns	The Australian Familial Pancreatic Cancer Cohort: Screening for Pancreatic Cancer in High Risk Australians	Poster Display
Alexandra	Jolley	Mitochondrial Complex III Deficiency Associated with a Homozygous Mutation in TTC19	Poster Display
Zena	Junek	Predictive Value of Immunoreactive Trypsin for Detection of Cystic Fibrosis in NSW	Oral Presentation
Zena	Junek	Current Trends in Newborn Screening Refusal	Poster Display
Erik-Jan	Kamsteeg	Pitfalls and Successes of Exome Sequencing: Mutations in a Novel Gene Cause Walker-Warburg Syndrome And Defective Glycosylation Of a-Dystrgoglycan	Oral Presentation
Nadine	Kasparian	Experiences of Parents Considering Genetic Testing to Better Understand their Child(Ren)'S Risk of Developing Familial Hypertrophic Cardiomyopathy (HCM) or Long Qt Syndrome (LQTS)	Poster Display
Simranpreet	Kaur	Mitochondrial Defects In RETT Syndrome	Oral Presentation
H	Kayserili	Pitfalls and Successes of Exome Sequencing: Mutations in a Novel Gene Cause Walker-Warburg Syndrome And Defective Glycosylation Of a-Dystrgoglycan	Oral Presentation

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W.T	Keng	Allopurinol and S-Adenosylmethionine Therapies for Lesch Nyhan Disease: Malaysian Experience, Including a Girl	Oral Presentation
Craig	Kennedy	2011 Molecular Genetics Quality Assurance Program Review	Oral Presentation
Craig	Kennedy	Fish Reveals Hidden Complexity of Copy Number Variants	Oral Presentation
Rosalie	Kenyon	SNP-Arrays for the Study of Plasma cell Myeloma	Oral Presentation
Greg	Kesby	Family Testing For a Microdeletion Found on Microarray CGH in a Prenatal Setting	Poster Display
David	Ketteridge	Mitochondrial Complex III Deficiency Associated with a Homozygous Mutation in TTC19	Poster Display
Won Tae	Kim	Screening for the Other Known Cystic Fibrosis Mutations on the Lightscanner	Poster Display
S.A.	Kinkel	Proteomic and Metabolic Analysis of a Mitochondrial Complex I Deficiency Mouse Model Obtained by Retroviral Insertion In The NDUFS4 Gene	Oral Presentation
Judy	Kirk	The Australian Familial Pancreatic Cancer Cohort: Screening for Pancreatic Cancer in High Risk Australians	Poster Display
Edwin	Kirk	Experiences of Parents Considering Genetic Testing to Better Understand their Child(Ren)'S Risk of Developing Familial Hypertrophic Cardiomyopathy (HCM) or Long Qt Syndrome (LQTS)	Poster Display
Judy	Kirk	Oncology Health Professionals' Attitudes Towards Treatment Focused Genetic Testing	Poster Display
Sara	Kivity	Mutations in PRRT Cause Benign Familial Infantile Epilepsy and Infantile Convulsions and Choreoathetosis Syndrome	Oral Presentation
Jasper	Komen	Proteomic and Metabolic Analysis of a Mitochondrial Complex I Deficiency Mouse Model Obtained by Retroviral Insertion In The NDUFS4 Gene	Oral Presentation
A	Korczyn	Mutations in PRRT Cause Benign Familial Infantile Epilepsy and Infantile Convulsions and Choreoathetosis Syndrome	Oral Presentation
N	Koulouris	Fish Reveals Hidden Complexity of Copy Number Variants	Oral Presentation
R	Kumar	Mutation in STXBP5L Associated with an Early Onset Neurodegenerative Disorder	Oral Presentation
Stefanie	Kung	An Exploratory Dysmorphometric Analysis to Define 3d Facial Phenotypic Signatures as a Foundation for Non-Invasive Monitoring of Lysosomal Storage Disorders	Oral Presentation
M	Kwint	Mutation in STXBP5L Associated with an Early Onset Neurodegenerative Disorder	Oral Presentation
John	Land	Mitochondrial Defects In RETT Syndrome	Oral Presentation
A	Laskowski	Proteomic and Metabolic Analysis of a Mitochondrial Complex I Deficiency Mouse Model Obtained by Retroviral Insertion In The NDUFS4 Gene	Oral Presentation
H.Y.	Law	Integration of Multicentre Data to Further Characterise the Phenotypes of Rare Clinically Significant Haemoglobinopathy Mutations	Poster Display
Rod	Lea	Molecular Genetics of Migraine; Implications for Therapeutic Development	Oral Presentation
Anna	Leaver	Attitudes of Adults with Cystic Fibrosis Towards Population-Based Cystic Fibrosis Carrier Screening	Oral Presentation
Dirk	Lefeber	Pitfalls and Successes of Exome Sequencing: Mutations in a Novel Gene Cause Walker-Warburg Syndrome And Defective Glycosylation Of a-Dystrgoglycan	Oral Presentation
Melanie	Leffler	Predictors of Genetic Information Disclosure by CF Obligate Carriers to At-Risk Relatives	Poster Display
Monkol	Lek	Whole Exome Sequencing Identifies a Novel Missense Mutation in Two Affected Children with a Suspected Rare Mendelian Disorder	Oral Presentation
Amanda	Leo	Niemann-Pick Disease Type C Caused By Maternal Uniparental Isodisomy of Chromosome 18	Oral Presentation
Dillon	Leong	Proteomic and Metabolic Analysis of a Mitochondrial Complex I Deficiency Mouse Model Obtained by Retroviral Insertion In The NDUFS4 Gene	Oral Presentation
Nancy	Lerda	Haemophilia A: Molecular Diagnosis is not Always Straight Forward	Oral Presentation
Peter	LeSouef	An Exploratory Dysmorphometric Analysis to Define 3d Facial Phenotypic Signatures as a Foundation for Non-Invasive Monitoring of Lysosomal Storage Disorders	Oral Presentation
Sharon	Lewis	Attitudes of Adults with Cystic Fibrosis Towards Population-Based Cystic Fibrosis Carrier Screening	Oral Presentation
Sharon	Lewis	No Thanks' - Why Pregnant Women Choose not to Have Cystic Fibrosis Carrier Screening	Poster Display
R	Li	Integration of Multicentre Data to Further Characterise the Phenotypes of Rare Clinically Significant Haemoglobinopathy Mutations	Poster Display
Andrew	Lidral	GWAS Reveals New Recessive Loci Associated with Non-Syndromic Facial Clefting	Poster Display

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J	Liebelt	Epilepsy with Cognitive Deficit and Autism Spectrum Disorders: Prospective Diagnosis by Array CGH	Oral Presentation
Esther	Liet	When Size Does Matter - Little Things Mean a Lot	Poster Display
Carol	Lim	Predictive Value of Immunoreactive Trypsin for Detection of Cystic Fibrosis in NSW	Oral Presentation
Yung-Yao	Lin	Pitfalls and Successes of Exome Sequencing: Mutations in a Novel Gene Cause Walker-Warburg Syndrome And Defective Glycosylation Of a-Dystrgoglycan	Oral Presentation
Ming	Lin	Haemophilia A: Molecular Diagnosis is not Always Straight Forward	Oral Presentation
Joanne	Lind	Detecting Differential Allelic Expression Using Amplification Refractory Mutation System (ARMS) PCR	Oral Presentation
Joanne	Lind	Use of Technology in Medical Genetics Education: Lessons Learned from the UWS MBBS Program	Poster Display
Francisco	Lopera	Pooling/Bootstrap-based GWAS (pbGWAS) Identifies New Loci Modifying the Age of Onset in PSEN1 E280A Alzheimer's Disease	Poster Display
Rohan	Lourie	Detection of Gene Mutations Causing Noonan Syndrome: Two Case Studies	Oral Presentation
Karen	Lower	Genome-wide Analysis of Sequence Variation Underlying Tissue-Specific Transcription Factor Binding and Gene Expression	Oral Presentation
Vesna	Lukic	The Whole Exome Sequencing Approach to Identify Novel Genes in Mitochondrial Respiratory Chain Disorders	Oral Presentation
Samantha	Lupton	Detecting Differential Allelic Expression Using Amplification Refractory Mutation System (ARMS) PCR	Oral Presentation
Alan	Ma	PAX6 Mutation Spectrum in an Australian and New Zealand Aniridia Cohort	Oral Presentation
Ivan	Macciocca	Cardiac Chaos: A Hypothetical about Cardiac Genetic Services, Issues and Challenges	Oral Presentation
Ivan	Macciocca	Attitudes of Adults with Cystic Fibrosis Towards Population-Based Cystic Fibrosis Carrier Screening	Oral Presentation
Ivan	Macciocca	Experiences of Parents Considering Genetic Testing to Better Understand their Child(Ren)'S Risk of Developing Familial Hypertrophic Cardiomyopathy (HCM) or Long Qt Syndrome (LQTS)	Poster Display
Rebecca	Macintosh	Experiences of Parents Considering Genetic Testing to Better Understand their Child(Ren)'S Risk of Developing Familial Hypertrophic Cardiomyopathy (HCM) or Long Qt Syndrome (LQTS)	Poster Display
John	MacMillan	Molecular Genetics of Migraine; Implications for Therapeutic Development	Oral Presentation
John	MacMillan	Genotype/Phenotype Classification of a Family with a Novel Missense Mutation in NOG, Surgical Implications for Stapedectomy	Poster Display
Bridget	Maher	Molecular Genetics of Migraine; Implications for Therapeutic Development	Oral Presentation
Jessica	Malkoun	Evaluation of a General Practitioner Education Module on First Trimester Screening and the Associated Genetic Counselling Skills	Poster Display
G.M.S.	Mancini	Pitfalls and Successes of Exome Sequencing: Mutations in a Novel Gene Cause Walker-Warburg Syndrome And Defective Glycosylation Of a-Dystrgoglycan	Oral Presentation
Christopher S	Manning	Genotype/Phenotype Classification of a Family with a Novel Missense Mutation in NOG, Surgical Implications for Stapedectomy	Poster Display
Nicole	Martin	Numerical and Structural Abnormalities of the Y Chromosome in Infertility Investigations	Oral Presentation
Nicole	Martin	Offering CFTR-PGD Without a Delay for Work Up; Reality or Fiction?	Oral Presentation
Nicole	Martin	FMR1 Gene Small Triplet Repeat Alleles Associated with POF/POI and Early Menopause	Oral Presentation
Amy	Martin	SPN Array Technology Identifies Novel Candidate Loci for Hnpcc/Lynch Syndrome	Oral Presentation
Ariel	Martinez	Pooling/Bootstrap-based GWAS (pbGWAS) Identifies New Loci Modifying the Age of Onset in PSEN1 E280A Alzheimer's Disease	Poster Display
Ariel	Martinez	Effects of LPHN3 on ADHD, Substance Use Disorder and Disruptive Behaviors	Poster Display
John	Massie	Attitudes of Adults with Cystic Fibrosis Towards Population-Based Cystic Fibrosis Carrier Screening	Oral Presentation
John	Massie	No Thanks' - Why Pregnant Women Choose not to Have Cystic Fibrosis Carrier Screening	Poster Display
John	Massie	Carrier Screening for Cytsic Fibrosis in Austrlia	Oral Presentation
John	Massie	Population-Based Carrier Screening for Cystic Fibrosis in Victoria: The Past 6 Years Experience	Oral Presentation
Isabelle	Maystadt	Pitfalls and Successes of Exome Sequencing: Mutations in a Novel Gene Cause Walker-Warburg Syndrome And Defective Glycosylation Of a-Dystrgoglycan	Oral Presentation
B	McClaren	The Experiences of Parents of Children with Developmental Delay, Autism or Intellectual Disability who Receive an FMR1 Grey Zone Result	Oral Presentation

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First Name	Last Name	Title	Presentation Type
Belinda	McClaren	No Thanks' - Why Pregnant Women Choose not to Have Cystic Fibrosis Carrier Screening	Poster Display
Louise	McCormack	Utility of Genetic Testing in Hypertrophic Cardiomyopathy in Queensland's Statewide Cardiac Genetics Clinic	Poster Display
Brett	McDermott	Complex IV Deficiency in a Girl with 22Q11.2 Deletion and Adjacent Segmental Uniparental Disomy	Poster Display
Mark	McEvoy	SPN Array Technology Identifies Novel Candidate Loci for Hnpcc/Lynch Syndrome	Oral Presentation
Alison	McEwen	Managing the Risks With Surgery: the Experiences of Nz Women With A Family History Of Breast And Ovarian Cancer	Oral Presentation
Julie	McGaughran	A Retrospective Study Of Cancer In Australian And New Zealand Cystic Fibrosis (Cf) Patients	Poster Display
Julie	McGaughran	Utility of Genetic Testing in Hypertrophic Cardiomyopathy in Queensland's Statewide Cardiac Genetics Clinic	Poster Display
James	McGill	5 Year Follow Up of 2 Siblings with Krabbe Disease Transplanted at 4 and 5 Weeks of Age Following Postmortem Diagnosis in a Sibling	Oral Presentation
Jim	McGill	The Effect of Continuous Haemodiafiltration and Intermittent Haemodialysis on Plasma Amino Acid and Ammonia Profiles for the Metabolic Patient	Poster Display
Jim	McGill	Complex IV Deficiency in a Girl with 22Q11.2 Deletion and Adjacent Segmental Uniparental Disomy	Poster Display
James	McGill	Unusual Radiographic Features of a 9 Year Old Girl with Attenuated Mucopolysaccharidosis Type VI	Poster Display
George	McGillivray	Is it Too Late for One Family to be Able to Utilise Prenatal Testing for Tuberous Sclerosis And Have A Healthy Baby?	Poster Display
Ivan	McGown	Genetic Testing for Maturity Onset Diabetes of the Young in Australia	Oral Presentation
Ivan	McGown	Detection of Gene Mutations Causing Noonan Syndrome: Two Case Studies	Oral Presentation
Ivan M	McGown	Allopurinol and S-Adenosylmethionine Therapies for Lesch Nyhan Disease: Malaysian Experience, Including a Girl	Oral Presentation
Pauline	McGrath	So, What Exactly is That You Do? Providing Genetic Education to Midwives and Nurses	Poster Display
L	McGregor	Epilepsy with Cognitive Deficit and Autism Spectrum Disorders: Prospective Diagnosis by Array CGH	Oral Presentation
Lesley	McGregor	Sequence Analysis oftThe Nemo Gene in a Cohort of Patients Without the Common Nemo?4–10 Deletion	Poster Display
Matt	McKenzie	Proteomic and Metabolic Analysis of a Mitochondrial Complex I Deficiency Mouse Model Obtained by Retroviral Insertion In The NDUFS4 Gene	Oral Presentation
John	Mcreanor	Unusual Radiographic Features of a 9 Year Old Girl with Attenuated Mucopolysaccharidosis Type VI	Poster Display
Avis	McWhinney	The Effect of Continuous Haemodiafiltration and Intermittent Haemodialysis on Plasma Amino Acid and Ammonia Profiles for the Metabolic Patient	Poster Display
Avis	McWhinney	Complex IV Deficiency in a Girl with 22Q11.2 Deletion and Adjacent Segmental Uniparental Disomy	Poster Display
S	Mead	The Australian Familial Pancreatic Cancer Cohort: Screening for Pancreatic Cancer in High Risk Australians	Poster Display
B	Meiser	Exploring Beliefs about Causes and Heritability of Major Depressive Disorder among Chinese Australians	Oral Presentation
Bettina	Meiser	An Exploration of the Clinical Experiences and Attitudes of Australian Health Professionals in Regard to Direct-To-Consumer (Dtc) Genetic Testing	Poster Display
Bettina	Meiser	Oncology Health Professionals' Attitudes Towards Treatment Focused Genetic Testing	Poster Display
Cliff	Meldrum	When Size Does Matter - Little Things Mean a Lot	Poster Display
Minal	Menezes	The Whole Exome Sequencing Approach to Identify Novel Genes in Mitochondrial Respiratory Chain Disorders	Oral Presentation
Sylvia	Metcalfe	The Experiences of Parents of Children with Developmental Delay, Autism or Intellectual Disability Who Receive an FMR1 Grey Zone Result	Oral Presentation
Kerry	Miller	Mouse Models of Human Skeletal Dysmorphologies	Oral Presentation
J	Millington	Fish Reveals Hidden Complexity Of Copy Number Variants	Oral Presentation
P	Mitchel	Exploring Beliefs about Causes and Heritability of Major Depressive Disorder among Chinese Australians	Oral Presentation
Gillian	Mitchell	International Sarcoma Kindred Study (ISKS) - Knowledge of Genetics Amongst Patients, Families and Health Professionals	Oral Presentation
Margaret	Mitchell	So, What Exactly is That You Do? Providing Genetic Education to Midwives and Nurses	Poster Display

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First Name	Last Name	Title	Presentation Type
Gillian	Mitchell	Oncology Health Professionals' Attitudes Towards Treatment Focused Genetic Testing	Poster Display
Laura	Molloy	The Experiences of Parents of Children with Developmental Delay, Autism or Intellectual Disability Who Receive an FMR1 Grey Zone Result	Oral Presentation
Sarah	Moore	SNP-Arrays for the Study of Plasma cell Myeloma	Oral Presentation
Francesca	Moore	HMG COA Synthase Deficiency- How Common is it and How Good are Our Markers?	Poster Display
Lina	Moreno	GWAS Reveals New Recessive Loci Associated with Non-Syndromic Facial Clefting	Poster Display
Chloe	Morgan	Detecting Differential Allelic Expression Using Amplification Refractory Mutation System (ARMS) PCR	Oral Presentation
Lucy	Morgan	Primary Ciliary Dyskinesia (PCD) Causing Idiopathic Tachypnoea in Newborn	Poster Display
David	Mowat	Changing Clinical Interpretation of Chromosomal Microarrays Over Time	Oral Presentation
David	Mowat	Mutations in Exons 41 and 42 of Fibrillin 1 Cause Short Stature of the Acromelic Type	Oral Presentation
David	Mowat	An Additional Chime Syndrome Case Due to Compound Heterozygous Mutations in PIGL	Poster Display
David	Mowat	14q13 Microdeletion: a Recognizable Contiguous Gene Syndrome Encompassing Brain/Thyroid/Lung and Dental Phenotype	Oral Presentation
David	Mowat	Targeted Therapy for Neuro-Developmental disorders Involving the mTOR Pathway	Oral Presentation
Glenda	Mullan	Changing Clinical Interpretation of Chromosomal Microarrays Over Time	Oral Presentation
J	Mulley	Epilepsy with Cognitive Deficit and Autism Spectrum Disorders: Prospective Diagnosis by Array CGH	Oral Presentation
J	Mulley	Mutations in PRRT Cause Benign Familial Infantile Epilepsy and Infantile Convulsions and Choreoathetosis Syndrome	Oral Presentation
Gillian	Mulvany	Challenges In Supplying And Managing A Low Protein Diet For The Postpartum Period In Ornithine Transcarbamylase Deficiency Heterozygotes	Poster Display
Maximilian	Muneke	Effects of LPHN3 on ADHD, Substance Use Disorder and Disruptive Behaviors	Poster Display
Craig	Munns	OI Type III-FKBP10 in Non-Consanguineous Australian Families	Oral Presentation
Kristi	Murphy	Family Testing For a Microdeletion Found on Microarray CGH in a Prenatal Setting	Poster Display
K	Murphy	Adventures in Prenatal Testing	Oral Presentation
Bobby	Ng	An Additional Chime Syndrome Case Due to Compound Heterozygous Mutations in PIGL	Poster Display
Wan Yi	Ng	Genome-Wide Analysis Using Next-Generation Sequencing For Disease-Gene Identification In Genetically Heterogeneous Eye Diseases	Oral Presentation
I	Ng	Integration of Multicentre Data to Further Characterise the Phenotypes of Rare Clinically Significant Haemoglobinopathy Mutations	Poster Display
L.H	Ngu	Allopurinol and S-Adenosylmethionine Therapies for Lesch Nyhan Disease: Malaysian Experience, Including a Girl	Oral Presentation
Andy	Nguyen	Haemoglobin E and Co-Existing Alpha Thalassaemia in the South Australian Population	Poster Display
Jillian	Nicholl	Epilepsy with Cognitive Deficit and Autism Spectrum Disorders: Prospective Diagnosis by Array CGH	Oral Presentation
Jillian	Nicholl	Mitochondrial Complex III Deficiency Associated with a Homozygous Mutation in TTC19	Poster Display
Rosie	Nicholls	First Impressions Count! Comparison of Three 'Family History Intake Processes'; Influence of Intake Process on Clinician and Genetic Counsellor Experience and Satisfaction	Poster Display
Catherine	Nicholls	Haemoglobin E and Co-Existing Alpha Thalassaemia in the South Australian Population	Poster Display
Catherine	Nicholls	Haemophilia A: Molecular Diagnosis is not Always Straight Forward	Oral Presentation
Rosie	Nicholls	First Impressions Count! Comparison of Client Satisfaction and Distress Between Three Different Intake Processes to a Family Cancer Clinic	Poster Display
Mario	Nicola	SNP-Arrays for the Study of Plasma cell Myeloma	Oral Presentation
Stephanie	Oates	Mitochondrial Complex III Deficiency Associated with a Homozygous Mutation in TTC19	Poster Display
Sheridan	O'Donnell	The Importance of Confirming CFTR Mutation Genotypes for Affected Individuals and the Significance of This Information for Extended Family Members, Clinical Care and Future Reproductive Decision Making	Poster Display
Emma	Palmer	Changing Clinical Interpretation of Chromosomal Microarrays Over Time	Oral Presentation

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First Name	Last Name	Title	Presentation Type
Lina	Partis	The Differences Between Discovery and Diagnostic DNA Methylation Measurements May Impede Clinical Application	Oral Presentation
S.R.	Pasricha	Integration of Multicentre Data to Further Characterise the Phenotypes of Rare Clinically Significant Haemoglobinopathy Mutations	Poster Display
E.A.J.	Peeters	Pitfalls and Successes of Exome Sequencing: Mutations in a Novel Gene Cause Walker-Warburg Syndrome And Defective Glycosylation Of a-Dystrgoglycan	Oral Presentation
James	Pelekanos	Mutations in PRRT Cause Benign Familial Infantile Epilepsy and Infantile Convulsions and Choreoathetosis Syndrome	Oral Presentation
Gregory	Pelka	Deletion of a Novel Phosphatase Results in a Rett Syndrome-Like Phenotype	Oral Presentation
Gregory	Pelka	Mitochondrial Defects In RETT Syndrome	Oral Presentation
Heidi	Peters	Receiving Enzyme Replacement Therapy for a Lysosomal Storage Disorder; Exploring the Experiences of Young Patients and Their Families	Oral Presentation
Greg	Peters	Genome-Wide Analysis Using Next-Generation Sequencing For Disease-Gene Identification In Genetically Heterogeneous Eye Diseases	Oral Presentation
Greg	Peters	PAX6 Mutation Spectrum in an Australian and New Zealand Aniridia Cohort	Oral Presentation
Gregory	Peters	14q13 Microdeletion: a Recognizable Contiguous Gene Syndrome Encompassing Brain/Thyroid/Lung and Dental Phenotype	Oral Presentation
Madelyn	Peterson	Assessment of Patient and Relatives' Understanding of 'No Mutation Found' and 'Unclassified Variant' BRCA1/BRCA2 Gene Test Results and Effect on Health Behaviours	Poster Display
D.	Petković Ramadža	Pitfalls and Successes of Exome Sequencing: Mutations in a Novel Gene Cause Walker-Warburg Syndrome And Defective Glycosylation Of a-Dystrgoglycan	Oral Presentation
Vicki	Petrou	Population-Based Carrier Screening for Cystic Fibrosis in Victoria: The Past 6 Years Experience	Oral Presentation
M.	Petrou	Integration of Multicentre Data to Further Characterise the Phenotypes of Rare Clinically Significant Haemoglobinopathy Mutations	Poster Display
R	Pfundt	Pitfalls and Successes of Exome Sequencing: Mutations in a Novel Gene Cause Walker-Warburg Syndrome And Defective Glycosylation Of a-Dystrgoglycan	Oral Presentation
B	Phipson	Proteomic and Metabolic Analysis of a Mitochondrial Complex I Deficiency Mouse Model Obtained by Retroviral Insertion In The NDUFS4 Gene	Oral Presentation
R	Phua	Fish Reveals Hidden Complexity of Copy Number Variants	Oral Presentation
E	Piirinen	Pathomechanisms and Experimental Therapies of Mitochondrial Disease	Oral Presentation
James	Pitt	Implementing a Two-Stage Written Consent Process for Newborn Screening in Victoria	Oral Presentation
J.J.	Pitt	Proteomic and Metabolic Analysis of a Mitochondrial Complex I Deficiency Mouse Model Obtained by Retroviral Insertion In The NDUFS4 Gene	Oral Presentation
james	Pitt	Biochemical Screening for Antiquitin Deficiency: Australasian Experience	Oral Presentation
David	Power	A Genetic Approach to the Problem of Monitoring Kidney Transplant Rejection	Oral Presentation
Gareth	Price	Complex IV Deficiency in a Girl with 22Q11.2 Deletion and Adjacent Segmental Uniparental Disomy	Poster Display
Claire	Pridmore	Epilepsy with Cognitive Deficit and Autism Spectrum Disorders: Prospective Diagnosis by Array CGH	Oral Presentation
Clair	Pridmore	Mutations in PRRT Cause Benign Familial Infantile Epilepsy and Infantile Convulsions and Choreoathetosis Syndrome	Oral Presentation
Peter	Procopis	Childhood Spastic Diplegia in a Novel Form of Non-Ketotic Hyperglycinaemia, Associated with Pyruvate Dehydrogenase Complex Deficiency, Caused by Glrx5 Deficiency of the Iron-Sulfur Cluster	Oral Presentation
H.	Prokisch	Identification and Characterization of New Mitochondrial Disease Genes	Oral Presentation
Ivan	Prokudin	Genome-Wide Analysis Using Next-Generation Sequencing For Disease-Gene Identification In Genetically Heterogeneous Eye Diseases	Oral Presentation
Belinda	Rahman	Oncology Health Professionals' Attitudes Towards Treatment Focused Genetic Testing	Poster Display
Charlene	Rapsey	The Australasian Clinic of Genomic and Personalised Medicine	Poster Display
David	Ravine	HDAC6 Inhibitors: A Novel Therapy for RETT Syndrome	Poster Display
Juergen	Reichardt	Complementation between Specific Phenylalanine Hydroxylase (Pah) Missense Mutations and Correlation to Clinical Phenotype in Phenylketonuria (PKU)	Oral Presentation
Marta	Ribases	Effects of LPHN3 on ADHD, Substance Use Disorder and Disruptive Behaviors	Poster Display
M	Riemersma	Pitfalls and Successes of Exome Sequencing: Mutations in a Novel Gene Cause Walker-Warburg Syndrome And Defective Glycosylation Of a-Dystrgoglycan	Oral Presentation
Lisa	Riley	The Whole Exome Sequencing Approach to Identify Novel Genes in Mitochondrial Respiratory Chain Disorders	Oral Presentation
William	Ritchie	Integrated Microrna and Mrna Profiling of the Mouse Ventricles during Development of Severe Hypertrophic Cardiomyopathy and Heart Failure	Oral Presentation

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First Name	Last Name	Title	Presentation Type
Dora	Rivera	GWAS Reveals New Recessive Loci Associated with Non-Syndromic Facial Clefting	Poster Display
Cynthia	Roberts	Public Funding Of PGD - A Family Health Care Priority	Oral Presentation
Robert	Robertson	Family Testing For a Microdeletion Found on Microarray CGH in a Prenatal Setting	Poster Display
R	Robertson	Adventures in Prenatal Testing	Oral Presentation
Brian	Robinson	Childhood Spastic Diplegia in a Novel Form of Non-Ketotic Hyperglycinaemia, Associated with Pyruvate Dehydrogenase Complex Deficiency, Caused by Glrx5 Deficiency of the Iron-Sulfur Cluster	Oral Presentation
Juliane	Roeper	Molybdenum Cofactor Deficiency due to MOCS2 Mutation	Poster Display
Carolyn	Rogers	Postnatal Mothers Attitudes to Newborn Screening for Fragile X Syndrome	Oral Presentation
Tony	Roscioli	Pitfalls and Successes of Exome Sequencing: Mutations in a Novel Gene Cause Walker-Warburg Syndrome And Defective Glycosylation Of a-Dystrgoglycan	Oral Presentation
R.T.	Ryan	Proteomic and Metabolic Analysis of a Mitochondrial Complex I Deficiency Mouse Model Obtained by Retroviral Insertion In The NDUFS4 Gene	Oral Presentation
Rani	Sachdev	14q13 Microdeletion: a Recognizable Contiguous Gene Syndrome Encompassing Brain/Thyroid/Lung and Dental Phenotype	Oral Presentation
Lynette	Sadleir	Mutations in PRRT Cause Benign Familial Infantile Epilepsy and Infantile Convulsions and Choreoathetosis Syndrome	Oral Presentation
Maragaret	Sahhar	Receiving Enzyme Replacement Therapy for a Lysosomal Storage Disorder; Exploring the Experiences of Young Patients and Their Families	Oral Presentation
Manju	Salaria	An Audit of patients with Hereditary Hemorrhagic Telangiectasia (HHT)	Poster Display
Mona	Saleh	Predictors of Genetic Information Disclosure by CF Obligate Carriers to At-Risk Relatives	Poster Display
Mona	Saleh	Challenges Faced by Health Care Interpreters Practicing in Genetic Counselling Services	Poster Display
Mona	Saleh	Providing Written Information for Consumers on Rare Genetic Conditions - Attitudes Towards and the Use of Genetics Information Sheets	Poster Display
Wendy	Salter	Enzyme Diagnosis of Fumarase Deficiency in Non-Identical Twins	Oral Presentation
Don	Samaranayake	The Australasian Clinic of Genomic and Personalised Medicine	Poster Display
Louisa	Sanchez	Sequence Analysis oftThe Nemo Gene in a Cohort of Patients Without the Common Nemo?4–10 Deletion	Poster Display
Sarah	Sandaradura	An Additional Chime Syndrome Case Due to Compound Heterozygous Mutations in PIGL	Poster Display
Jose	Santamaria	Molybdenum Cofactor Deficiency due to MOCS2 Mutation	Poster Display
Christobel	Saunders	Oncology Health Professionals' Attitudes Towards Treatment Focused Genetic Testing	Poster Display
Ravi	Savarirayan	A Guide to the Prenatal Evaluation of Suspected Skeletal Dysplasias	Oral Presentation
Alka	Saxena	HDAC6 Inhibitors: A Novel Therapy for RETT Syndrome	Poster Display
Thomas	Scerri	Whole Exome Sequencing Identifies a Novel Missense Mutation in Two Affected Children with a Suspected Rare Mendelian Disorder	Oral Presentation
Gunter	Scharer	Childhood Spastic Diplegia in a Novel Form of Non-Ketotic Hyperglycinaemia, Associated with Pyruvate Dehydrogenase Complex Deficiency, Caused by Glrx5 Deficiency of the Iron-Sulfur Cluster	Oral Presentation
I	Scheffer	Mutations in PRRT Cause Benign Familial Infantile Epilepsy and Infantile Convulsions and Choreoathetosis Syndrome	Oral Presentation
P	Schofield	Exploring Beliefs about Causes and Heritability of Major Depressive Disorder among Chinese Australians	Oral Presentation
M	Schraders	Pitfalls and Successes of Exome Sequencing: Mutations in a Novel Gene Cause Walker-Warburg Syndrome And Defective Glycosylation Of a-Dystrgoglycan	Oral Presentation
Guenter	Schwarz	Molybdenum Cofactor Deficiency due to MOCS2 Mutation	Poster Display
Ulrike	Schwarze	OI Type III-FKBP10 in Non-Consanguineous Australian Families	Oral Presentation
Hamish	Scott	Haemoglobin E and Co-Existing Alpha Thalassaemia in the South Australian Population	Poster Display
H.S.	Scott	Proteomic and Metabolic Analysis of a Mitochondrial Complex I Deficiency Mouse Model Obtained by Retroviral Insertion In The NDUFS4 Gene	Oral Presentation
Rodney	Scott	SPN Array Technology Identifies Novel Candidate Loci for Hnpcc/Lynch Syndrome	Oral Presentation
Rodney	Scott	When Size Does Matter - Little Things Mean a Lot	Poster Display
Christopher	Semsarian	Integrated Microrna and Mrna Profiling of the Mouse Ventricles during Development of Severe Hypertrophic Cardiomyopathy and Heart Failure	Oral Presentation

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Christopher	Semsarian	Psychological Impairment Particularly in Mothers Following Sudden Cardiac Death in the Young	Oral Presentation
Christopher	Semsarian	Case Presentation - 'Getting Over the Shock of it' the Impact of Implantable Defibrillators'	Oral Presentation
Christopher	Semsarian	The Australian National Genetic Heart Disease Registry	Oral Presentation
T	Shaikh	Childhood Spastic Diplegia in a Novel Form of Non-Ketotic Hyperglycinaemia, Associated with Pyruvate Dehydrogenase Complex Deficiency, Caused by Glrx5 Deficiency of the Iron-Sulfur Cluster	Oral Presentation
Carolyn	Shalhoub	Predictors of Genetic Information Disclosure by CF Obligate Carriers to At-Risk Relatives	Poster Display
Leslie	Sheffield	Is it Too Late for One Family to be Able to Utilise Prenatal Testing for Tuberous Sclerosis And Have A Healthy Baby?	Poster Display
Marwa	Shehab	In Vitro Study of the Anti Oxidant Effect of Astaxanthin and Vitamin E on Cultured Lymphocyts from Fanconi Anemia Patients	Poster Display
Yaying	Shen	Case Study: Characterisation and Implications of Ring Chromosome 21 in a Female Carrier	Poster Display
Rhian	Shephard	Integrated Microrna and Mrna Profiling of the Mouse Ventricles during Development of Severe Hypertrophic Cardiomyopathy and Heart Failure	Oral Presentation
Jill	Shergold	5 Year Follow Up of 2 Siblings with Krabbe Disease Transplanted at 4 and 5 Weeks of Age Following Postmortem Diagnosis in a Sibling	Oral Presentation
E	Sheridan	Pitfalls and Successes of Exome Sequencing: Mutations in a Novel Gene Cause Walker-Warburg Syndrome And Defective Glycosylation Of a-Dystrgoglycan	Oral Presentation
Elva	Shi	A Genetic Approach to the Problem of Monitoring Kidney Transplant Rejection	Oral Presentation
Shoshana	Shiloh	Partnership Between Genetic Counseling and Psychological Research: Implications for Both Partners	Oral Presentation
Yosef	Shiloh	The ATM-Mediated DNA Damage Response: Moving Between the Forest and the Trees	Oral Presentation
Shoshana	Shiloh	Coping	Oral Presentation
A	Sickmann	Proteomic and Metabolic Analysis of a Mitochondrial Complex I Deficiency Mouse Model Obtained by Retroviral Insertion In The NDUFS4 Gene	Oral Presentation
Letitia	Silberbauer	First Impressions Count! Comparison of Three 'Family History Intake Processes'; Influence of Intake Process on Clinician and Genetic Counsellor Experience and Satisfaction	Poster Display
Tish	Silberbauer	First Impressions Count! Comparison of Client Satisfaction and Distress Between Three Different Intake Processes to a Family Cancer Clinic	Poster Display
David	Sillence	Whole Exome Sequencing Identifies a Novel Missense Mutation in Two Affected Children with a Suspected Rare Mendelian Disorder	Oral Presentation
David	Sillence	Mutations in Exons 41 and 42 of Fibrillin 1 Cause Short Stature of the Acromelic Type	Oral Presentation
David	Sillence	Teaching and Learning in Genetic Medicine	Oral Presentation
David	Sillence	OI Type III-FKBP10 in Non-Consanguineous Australian Families	Oral Presentation
David	Sillence	An Observational Study of Nine Adult Late-Onset Pompe Disease Patients Treated or Untreated with Acid Alpha-Glucosidase Enzyme Replacement Therapy	Oral Presentation
Keow	Sim	Quantitative Amino Acid Analysis: Comparison of UPLC Tandem Mass Spectrometry With Ion-Exchange Chromatography With Post Column Ninhydrin Detection	Oral Presentation
Skye	Simpson	The Australian Familial Pancreatic Cancer Cohort: Screening for Pancreatic Cancer in High Risk Australians	Poster Display
Ruth	Simpson	Assessment of Patient and Relatives' Understanding of 'No Mutation Found' and 'Unclassified Variant' BRCA1/BRCA2 Gene Test Results and Effect on Health Behaviours	Poster Display
Aygul	Simsek	Haemoglobin E and Co-Existing Alpha Thalassaemia in the South Australian Population	Poster Display
Aygul	Simsek	Haemophilia A: Molecular Diagnosis is not Always Straight Forward	Oral Presentation
Howard	Slater	A Genetic Approach to the Problem of Monitoring Kidney Transplant Rejection	Oral Presentation
K	Smith	Mutation in STXBP5L Associated with an Early Onset Neurodegenerative Disorder	Oral Presentation
G.K.	Smyth	Proteomic and Metabolic Analysis of a Mitochondrial Complex I Deficiency Mouse Model Obtained by Retroviral Insertion In The NDUFS4 Gene	Oral Presentation
Engy	Soliman	In Vitro Study of the Anti Oxidant Effect of Astaxanthin and Vitamin E on Cultured Lymphocyts from Fanconi Anemia Patients	Poster Display
Benjamin D	Solomon	GWAS Reveals New Recessive Loci Associated with Non-Syndromic Facial Clefting	Poster Display

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First Name	Last Name	Title	Presentation Type
E	Spector	Childhood Spastic Diplegia in a Novel Form of Non-Ketotic Hyperglycinaemia, Associated with Pyruvate Dehydrogenase Complex Deficiency, Caused by Glrx5 Deficiency of the Iron-Sulfur Cluster	Oral Presentation
Helen	Spiers	Changing Clinical Interpretation of Chromosomal Microarrays Over Time	Oral Presentation
Allan	Spigelman	The Australian Familial Pancreatic Cancer Cohort: Screening for Pancreatic Cancer in High Risk Australians	Poster Display
Catherine	Spinks	Experiences of Parents Considering Genetic Testing to Better Understand their Child(Ren)'S Risk of Developing Familial Hypertrophic Cardiomyopathy (HCM) or Long Qt Syndrome (LQTS)	Poster Display
Michelle	Sproule	Is it Too Late for One Family to be Able to Utilise Prenatal Testing for Tuberous Sclerosis And Have A Healthy Baby?	Poster Display
Pawel	Stankiewicz	Baylor Experience with Chromosomal Microarray Analysis in Neurodevelopmental and Neurobehavioral Disorders (Molecular Mechanisms and Clinical Consequences of CNVs)	Oral Presentation
D.L.	Stemple	Pitfalls and Successes of Exome Sequencing: Mutations in a Novel Gene Cause Walker-Warburg Syndrome And Defective Glycosylation Of a-Dystrgoglycan	Oral Presentation
Alina	Stoita	The Australian Familial Pancreatic Cancer Cohort: Screening for Pancreatic Cancer in High Risk Australians	Poster Display
Micheal	Stormon	Liver Transplantation from an Adult Donor with an Unrecognized Urea Cycle Disorder to a Paediatric and an Adult Recipient	Oral Presentation
Rachel	Susman	Assessment of Patient and Relatives' Understanding of 'No Mutation Found' and 'Unclassified Variant' BRCA1/BRCA2 Gene Test Results and Effect on Health Behaviours	Poster Display
Marleen	Susman	Should we be Offering Women a Choice of the types of Chromosome Conditions they want Prenatally Diagnosed?	Oral Presentation
Jeffrey	Suttle	SNP-Arrays for the Study of Plasma cell Myeloma	Oral Presentation
Bente	Talseth-Palmer	SPN Array Technology Identifies Novel Candidate Loci for Hnpcc/Lynch Syndrome	Oral Presentation
Patrick	Tam	Deletion of a Novel Phosphatase Results in a Rett Syndrome-Like Phenotype	Oral Presentation
Patrick	Tam	Mitochondrial Defects In RETT Syndrome	Oral Presentation
Melinda	Tam	Challenges In Supplying And Managing A Low Protein Diet For The Postpartum Period In Ornithine Transcarbamylase Deficiency Heterozygotes	Poster Display
Tiong	Tan	Mouse Models of Human Skeletal Dysmorphologies	Oral Presentation
G.M.B.	Tan-Sindhunata	Pitfalls and Successes of Exome Sequencing: Mutations in a Novel Gene Cause Walker-Warburg Syndrome And Defective Glycosylation Of a-Dystrgoglycan	Oral Presentation
Peter	Taylor	Changing Clinical Interpretation of Chromosomal Microarrays Over Time	Oral Presentation
Jessica	Taylor	An Audit of patients with Hereditary Hemorrhagic Telangiectasia (HHT)	Poster Display
Michel	Tchan	An Observational Study of Nine Adult Late-Onset Pompe Disease Patients Treated or Untreated with Acid Alpha-Glucosidase Enzyme Replacement Therapy	Oral Presentation
Michel	Tchan	Challenges In Supplying And Managing A Low Protein Diet For The Postpartum Period In Ornithine Transcarbamylase Deficiency Heterozygotes	Poster Display
Michel	Tchan	Review of Patients Attending for the First Twelve Months of an Expanded Adult Genetic Metabolic Disorders Service	Poster Display
Samia	Temtamy	In Vitro Study of the Anti Oxidant Effect of Astaxanthin and Vitamin E on Cultured Lymphocyts from Fanconi Anemia Patients	Poster Display
Elaine	Tham	A Family With an ARG134CYS Mutation In COl1A1 and Overlapping Phenotypes of Oteogenesis Imperfecta and Ehlers-Danlos Syndrome	Poster Display
David	Thomas	International Sarcoma Kindred Study (ISKS) - Knowledge of Genetics Amongst Patients, Families and Health Professionals	Oral Presentation
Suzanna	Thompson	Epilepsy with Cognitive Deficit and Autism Spectrum Disorders: Prospective Diagnosis by Array CGH	Oral Presentation
E	Thompson	Epilepsy with Cognitive Deficit and Autism Spectrum Disorders: Prospective Diagnosis by Array CGH	Oral Presentation
Susan	Thompson	OTC - Energy and Protein Requirements of a 2 Year Old Noumean Girl Presenting with Unstable Ammonium Levels	Poster Display
Susan	Thompson	Management of PKU in Australia - A Survey of Clinic Practices	Oral Presentation
Kate	Thompson	Developing a Youth Friendly Model of Genetic Counselling	Oral Presentation
Elizabeth	Thompson	A Family With an ARG134CYS Mutation In COl1A1 and Overlapping Phenotypes of Oteogenesis Imperfecta and Ehlers-Danlos Syndrome	Poster Display

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David	Thorburn	The Whole Exome Sequencing Approach to Identify Novel Genes in Mitochondrial Respiratory Chain Disorders	Oral Presentation
David	Thorburn	Novel Disorders of the Mitochondrial Respiratory Chain	Oral Presentation
D.R.	Thorburn	Proteomic and Metabolic Analysis of a Mitochondrial Complex I Deficiency Mouse Model Obtained by Retroviral Insertion In The NDUFS4 Gene	Oral Presentation
David	Thorburn	Enzyme Diagnosis of Fumarase Deficiency in Non-Identical Twins	Oral Presentation
David	Thorburn	Childhood Spastic Diplegia in a Novel Form of Non-Ketotic Hyperglycinaemia, Associated with Pyruvate Dehydrogenase Complex Deficiency, Caused by Glrx5 Deficiency of the Iron-Sulfur Cluster	Oral Presentation
Vanessa	Toh	Case Study: Characterisation and Implications of Ring Chromosome 21 in a Female Carrier	Poster Display
Bruce	Tonge	Changing Clinical Interpretation of Chromosomal Microarrays Over Time	Oral Presentation
Simone	Tregoning	Enzyme Diagnosis of Fumarase Deficiency in Non-Identical Twins	Oral Presentation
Simon	Troth	Providing Written Information for Consumers on Rare Genetic Conditions - Attitudes Towards and the Use of Genetics Information Sheets	Poster Display
Tatiana	Tsoutsman	Integrated Microrna and Mrna Profiling of the Mouse Ventricles during Development of Severe Hypertrophic Cardiomyopathy and Heart Failure	Oral Presentation
Kathy	Tucker	The Australian Familial Pancreatic Cancer Cohort: Screening for Pancreatic Cancer in High Risk Australians	Poster Display
Kathy	Tucker	Oncology Health Professionals' Attitudes Towards Treatment Focused Genetic Testing	Poster Display
Gillian	Turner	Changing Clinical Interpretation of Chromosomal Microarrays Over Time	Oral Presentation
Catherine	Turner	Postnatal Mothers Attitudes to Newborn Screening for Fragile X Syndrome	Oral Presentation
E	van Beusekom	Pitfalls and Successes of Exome Sequencing: Mutations in a Novel Gene Cause Walker-Warburg Syndrome And Defective Glycosylation Of a-Dystrgoglycan	Oral Presentation
Hans	van Bokhoven	Pitfalls and Successes of Exome Sequencing: Mutations in a Novel Gene Cause Walker-Warburg Syndrome And Defective Glycosylation Of a-Dystrgoglycan	Oral Presentation
C	van den Elzen	Pitfalls and Successes of Exome Sequencing: Mutations in a Novel Gene Cause Walker-Warburg Syndrome And Defective Glycosylation Of a-Dystrgoglycan	Oral Presentation
Mark	van der Hoek	SNP-Arrays for the Study of Plasma cell Myeloma	Oral Presentation
J	Van Hove	Childhood Spastic Diplegia in a Novel Form of Non-Ketotic Hyperglycinaemia, Associated with Pyruvate Dehydrogenase Complex Deficiency, Caused by Glrx5 Deficiency of the Iron-Sulfur Cluster	Oral Presentation
Jeroen	van Reeuwijk	Pitfalls and Successes of Exome Sequencing: Mutations in a Novel Gene Cause Walker-Warburg Syndrome And Defective Glycosylation Of a-Dystrgoglycan	Oral Presentation
Danya	Vears	Genetic Carrier Testing in Healthy Siblings: Investigation of Current Practice and Health Professionals' Views	Oral Presentation
Jorge	Velez	Pooling/Bootstrap-based GWAS (pbGWAS) Identifies New Loci Modifying the Age of Onset in PSEN1 E280A Alzheimer's Disease	Poster Display
Joris	Veltman	Pitfalls and Successes of Exome Sequencing: Mutations in a Novel Gene Cause Walker-Warburg Syndrome And Defective Glycosylation Of a-Dystrgoglycan	Oral Presentation
J.A.	Veltman	Mutation in STXBP5L Associated with an Early Onset Neurodegenerative Disorder	Oral Presentation
Douglas	Vernimmen	Genome-wide Analysis of Sequence Variation Underlying Tissue-Specific Transcription Factor Binding and Gene Expression	Oral Presentation
C	Viscomi	Pathomechanisms And Experimental Therapies Of Mitochondrial Disease	Oral Presentation
C	Viscomi	Identification and Characterization of New Mitochondrial Disease Genes	Oral Presentation
L.E.L.M.	Vissers	Pitfalls and Successes of Exome Sequencing: Mutations in a Novel Gene Cause Walker-Warburg Syndrome And Defective Glycosylation Of a-Dystrgoglycan	Oral Presentation
Ann	Voss	Proteomic and Metabolic Analysis of a Mitochondrial Complex I Deficiency Mouse Model Obtained by Retroviral Insertion In The NDUFS4 Gene	Oral Presentation
Claire	Wakefield	First Impressions Count! Comparison of Three 'Family History Intake Processes'; Influence of Intake Process on Clinician and Genetic Counsellor Experience and Satisfaction	Poster Display
Claire	Wakefield	First Impressions Count! Comparison of Client Satisfaction and Distress Between Three Different Intake Processes to a Family Cancer Clinic	Poster Display
Mark	Walters	An Exploratory Dysmorphometric Analysis to Define 3d Facial Phenotypic Signatures as a Foundation for Non-Invasive Monitoring of Lysosomal Storage Disorders	Oral Presentation
Wendy	Waters	Epilepsy with Cognitive Deficit and Autism Spectrum Disorders: Prospective Diagnosis by Array CGH	Oral Presentation
Sharon	Way	When Size Does Matter - Little Things Mean a Lot	Poster Display
Diane	Webster	Harmonisation of Newborn Screening Testing	Oral Presentation

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Anna	Wedell	Adenosine Kinase Deficiency, a Novel Cause of Hypermethioninemia Presenting as a Slowly Progressive Encephalopathy with Epilepsy, Myopathy and Liver Disease	Oral Presentation
K.M.	Weekes	Integration of Multicentre Data to Further Characterise the Phenotypes of Rare Clinically Significant Haemoglobinopathy Mutations	Poster Display
J	Wells	Integration of Multicentre Data to Further Characterise the Phenotypes of Rare Clinically Significant Haemoglobinopathy Mutations	Poster Display
Mary	Westbrook	Challenges In Supplying And Managing A Low Protein Diet For The Postpartum Period In Ornithine Transcarbamylase Deficiency Heterozygotes	Poster Display
Mary	Westbrook	Review of Patients Attending for the First Twelve Months of an Expanded Adult Genetic Metabolic Disorders Service	Poster Display
John	Whitehall	Molybdenum Cofactor Deficiency due to MOCS2 Mutation	Poster Display
John	Whitehall	Primary Ciliary Dyskinesia (PCD) Causing Idiopathic Tachypnoea in Newborn	Poster Display
Bruce	Whitehead	The Importance of Confirming CFTR Mutation Genotypes for Affected Individuals and the Significance of This Information for Extended Family Members, Clinical Care and Future Reproductive Decision Making	Poster Display
N	Wieskamp	Mutation in STXBP5L Associated with an Early Onset Neurodegenerative Disorder	Oral Presentation
Bridget	Wilcken	Predictive Value of Immunoreactive Trypsin for Detection of Cystic Fibrosis in NSW	Oral Presentation
Bridget	Wilcken	Current Trends in Newborn Screening Refusal	Poster Display
Bridget	Wilcken	Childhood Spastic Diplegia in a Novel Form of Non-Ketotic Hyperglycinaemia, Associated with Pyruvate Dehydrogenase Complex Deficiency, Caused by Glrx5 Deficiency of the Iron-Sulfur Cluster	Oral Presentation
A	Wilde	Exploring Beliefs about Causes and Heritability of Major Depressive Disorder among Chinese Australians	Oral Presentation
Veronica	Wiley	Newborn Screening for Congenital Adrenal Hyperplasia	Oral Presentation
V	Wiley	Postnatal Mothers Attitudes to Newborn Screening for Fragile X Syndrome	Oral Presentation
Veronica	Wiley	Predictive Value of Immunoreactive Trypsin for Detection of Cystic Fibrosis in NSW	Oral Presentation
Veronica	Wiley	Current Trends in Newborn Screening Refusal	Poster Display
Veronica	Wiley	Screening for the Other Known Cystic Fibrosis Mutations on the Lightscanner	Poster Display
M.A.	Willemsen	Pitfalls and Successes of Exome Sequencing: Mutations in a Novel Gene Cause Walker-Warburg Syndrome And Defective Glycosylation Of a-Dystrgoglycan	Oral Presentation
Mark	Williams	Genetic Testing for Maturity Onset Diabetes of the Young in Australia	Oral Presentation
Mark	Williams	Detection of Gene Mutations Causing Noonan Syndrome: Two Case Studies	Oral Presentation
David	Williams	The Australian Familial Pancreatic Cancer Cohort: Screening for Pancreatic Cancer in High Risk Australians	Poster Display
R	Williams	The Australian Familial Pancreatic Cancer Cohort: Screening for Pancreatic Cancer in High Risk Australians	Poster Display
Rachel	Williams	First Impressions Count! Comparison of Three 'Family History Intake Processes'; Influence of Intake Process on Clinician and Genetic Counsellor Experience and Satisfaction	Poster Display
Samantha	Williams	Niemann-Pick Disease Type C Caused By Maternal Uniparental Isodisomy of Chromosome 18	Oral Presentation
Rachel	Williams	First Impressions Count! Comparison of Client Satisfaction and Distress Between Three Different Intake Processes to a Family Cancer Clinic	Poster Display
Samantha	Williams	Unusual Radiographic Features of a 9 Year Old Girl with Attenuated Mucopolysaccharidosis Type VI	Poster Display
Sarah	Williamson	Deletion of a Novel Phosphatase Results in a Rett Syndrome-Like Phenotype	Oral Presentation
Meredith	Wilson	Whole Exome Sequencing Identifies a Novel Missense Mutation in Two Affected Children with a Suspected Rare Mendelian Disorder	Oral Presentation
John	Wilson	Attitudes of Adults with Cystic Fibrosis Towards Population-Based Cystic Fibrosis Carrier Screening	Oral Presentation
John	Wilson	A Retrospective Study Of Cancer In Australian And New Zealand Cystic Fibrosis (Cf) Patients	Poster Display
Meredith	Wilson	Experiences of Parents Considering Genetic Testing to Better Understand their Child(Ren)'S Risk of Developing Familial Hypertrophic Cardiomyopathy (HCM) or Long Qt Syndrome (LQTS)	Poster Display
Leeanda	Wilton	Preimplantation Genetic Diagnosis of Aneuploidy and Chromosomal Translocations Using Array Comparative Genomic Hybridisation	Oral Presentation
Andrew	Winnington	The Australasian Clinic of Genomic and Personalised Medicine	Poster Display
Ingrid	Winship	An Audit of patients with Hereditary Hemorrhagic Telangiectasia (HHT)	Poster Display

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J	Woenig	Mutation in STXBP5L Associated with an Early Onset Neurodegenerative Disorder	Oral Presentation
Flora	Wong	Molybdenum Cofactor Deficiency due to MOCS2 Mutation	Poster Display
Yee Chee	Wong	Case Study: Characterisation and Implications of Ring Chromosome 21 in a Female Carrier	Poster Display
M	Woontner	Childhood Spastic Diplegia in a Novel Form of Non-Ketotic Hyperglycinaemia, Associated with Pyruvate Dehydrogenase Complex Deficiency, Caused by Glrx5 Deficiency of the Iron-Sulfur Cluster	Oral Presentation
Tiffany	Wotton	Postnatal Mothers Attitudes to Newborn Screening for Fragile X Syndrome	Oral Presentation
Joyce	Wu	Genetic Testing for Maturity Onset Diabetes of the Young in Australia	Oral Presentation
Kathy	Wu	An Observational Study of Nine Adult Late-Onset Pompe Disease Patients Treated or Untreated with Acid Alpha-Glucosidase Enzyme Replacement Therapy	Oral Presentation
Joyce	Wu	Enzyme Diagnosis of Fumarase Deficiency in Non-Identical Twins	Oral Presentation
Tong	XingZhang	Towards the Development of a Genetically Modified Probiotic as a Novel Therapy for Phenylketonuria (PKU)	Oral Presentation
M	Xu	Exploring Beliefs about Causes and Heritability of Major Depressive Disorder among Chinese Australians	Oral Presentation
Laura	Yeates	Psychological Impairment Particularly in Mothers Following Sudden Cardiac Death in the Young	Oral Presentation
Laura	Yeates	Case Presentation - 'Getting Over the Shock of it' the Impact of Implantable Defibrillators'	Oral Presentation
Alison	Yeung	Is it Too Late for One Family to be Able to Utilise Prenatal Testing for Tuberous Sclerosis And Have A Healthy Baby?	Poster Display
Mary-Anne	Young	International Sarcoma Kindred Study (ISKS) - Knowledge of Genetics Amongst Patients, Families and Health Professionals	Oral Presentation
Mary-Anne	Young	Developing a Youth Friendly Model of Genetic Counselling	Oral Presentation
David B	Young	Genotype/Phenotype Classification of a Family with a Novel Missense Mutation in NOG, Surgical Implications for Stapedectomy	Poster Display
Saira	Yousoof	Genome-Wide Analysis Using Next-Generation Sequencing For Disease-Gene Identification In Genetically Heterogeneous Eye Diseases	Oral Presentation
S	Yu	Epilepsy with Cognitive Deficit and Autism Spectrum Disorders: Prospective Diagnosis by Array CGH	Oral Presentation
Sui	Yu	Mitochondrial Complex III Deficiency Associated with a Homozygous Mutation in TTC19	Poster Display
Sui	Yu	6p24.2 Microdeletion Involving TFAP2A Without Classic Features of Branchio-Oculo Facial Syndrome	Oral Presentation
R	Zahedi	Proteomic and Metabolic Analysis of a Mitochondrial Complex I Deficiency Mouse Model Obtained by Retroviral Insertion In The NDUFS4 Gene	Oral Presentation
Andreas	Zankl	Unusual Radiographic Features of a 9 Year Old Girl with Attenuated Mucopolysaccharidosis Type VI	Poster Display
Massimo	Zeviani	Markedly Effective Gene Therapy in an Ethylmalonic Encephalopathy Mouse Model	Oral Presentation
Massimo	Zeviani	Pathomechanisms and Experimental Therapies of Mitochondrial Disease	Oral Presentation
Massimo	Zeviani	Identification and Characterization of New Mitochondrial Disease Genes	Oral Presentation
H	Zhou	Pitfalls and Successes of Exome Sequencing: Mutations in a Novel Gene Cause Walker-Warburg Syndrome And Defective Glycosylation Of a-Dystrgoglycan	Oral Presentation
Lilian	Zou	Exploring Beliefs about Causes and Heritability of Major Depressive Disorder among Chinese Australians	Oral Presentation
Sameer	Zuberi	Mutations in PRRT Cause Benign Familial Infantile Epilepsy and Infantile Convulsions and Choreoathetosis Syndrome	Oral Presentation
Yvonne	Zurynski	Inborn Errors of Metabolism: Significant Impacts on Families and Health Services	Oral Presentation

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