



genetic alliance
australia

OFFICIAL LAUNCH OF GENETIC ALLIANCE AUSTRALIA'S

24TH GENETIC DISORDERS AWARENESS WEEK

GDAW 2017 GENETICS, THE SCIENCE AND THE IMPACT

TUESDAY 19TH SEPTEMBER 2017. 5.45PM – 8.30PM

VENUE:

STRANGERS' DINING ROOM, PARLIAMENT OF NSW
6 MACQUARIE STREET, SYDNEY 2000

GENETIC ALLIANCE AUSTRALIA

MAKING THE RIGHT CONNECTIONS SINCE 1988



Genetic Alliance (GA) Australia's Genetic Awareness Week seeks to educate the public about the impact of genetic conditions on individuals and families.

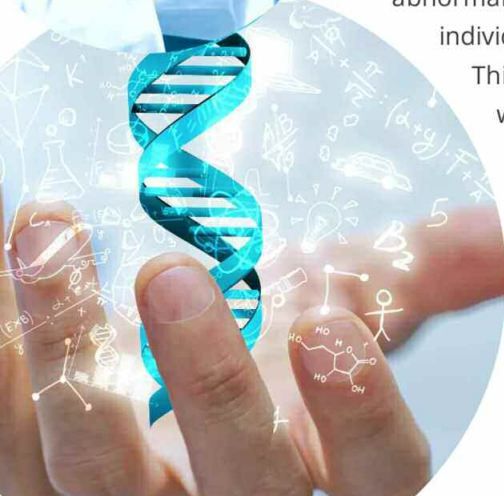
There are over 11,500 known hereditary single gene disorders and hundreds of syndromes known to be due to chromosomal abnormality. The number of genetic conditions identified due to an interaction of a genetic predisposition and environmental factors, many of which cause common health problems, increases every year e.g. cardiovascular, neurological, cancer, metabolic and mental illness.



It is estimated that in the first twenty years of life, about 5% of the Australasian population will be affected by an illness, impairment or disability either wholly or partly due to their inherited information. Approximately 30% of patients in paediatric hospitals have a genetic component to their illness and at least 28% of all infant deaths result from genetic factors.

At least 50% of all miscarriages are caused by chromosomal abnormalities. GA provides support and information for individuals and families affected by genetic conditions.

This is especially important for rare conditions, which may not have a support group.



GA runs specialised seminars on genetic conditions bringing together families, individuals and professionals in an informal setting. GA has been making the right connections for people affected by genetic conditions since 1988.

PROGRAM

- 5.45pm REGISTRATION
- DRINKS AND CANAPES
- 6.55 pm EVERYONE SEATED
- 7.00 pm CHAIR: DR MICHAEL BRYDON
Chief Executive of the Sydney Children's
Hospitals Network
- 7.05pm WELCOME – DIANNE PETRIE OAM
Executive Director, Genetic Alliance Australia
- 7.10pm OFFICIAL LAUNCH – THE HON BRAD HAZZARD MP
NSW Minister for Health,
and Minister for Medical Research
- 7.15pm "Two sides of a genetic mystery"
TANSEL ERSAVAS
Principal Software Engineer
Deep Learning Initiative, Garvan Institute of
Medical Research
- 7.25pm "Williams syndrome - A Sibling's Perspective"
JODIE MOK - Sibling Story
- 7.35pm "A Vision of Hope"
TOMMASINA OWENS
Adult Sibling
- 7.45pm "A Lifetime Impact of Intellectual Disability"
RADHIKA RAJKUMAR - Genetic Counsellor
- 7.50pm "RNF12 Joins the Quigley's"
LYN QUIGLEY
Family Impact
- 8.00 pm COFFEE & TEA
- 8.30pm CLOSE



SPEAKERS



THE HON. BRAD HAZZARD MP

Brad Hazzard is a senior Minister in the NSW Government and in 2017 was appointed as Minister for Health, and Minister for Medical Research.

He oversees record investment in Health infrastructure and services which has resulted in a hospital building boom across the state and the funding of thousands of new frontline positions. NSW also has a

robust and innovative medical research knowledge economy which the NSW Government seeks to encourage and expand.

Brad started his professional life as a graduate science teacher at North Sydney Boys' High School. He later studied law at the University of NSW and was admitted as a solicitor in 1977. He also holds a Master of Laws from the University of Sydney.

He entered Parliament in 1991 as the State Member for Wakehurst, on Sydney's northern beaches. Brad has previously served as Minister for Family and Community Services and Minister for Social Housing, Attorney General and Minister for Justice and as Minister for Planning and Infrastructure as a Leader of the Legislative Assembly.



CHAIR: DR MICHAEL BRYDON

Dr Michael Brydon was appointed as the Chief Executive of the Sydney Children's Hospitals Network (SCHN) in July 2016, having held the acting role since February 2015. Prior to this, Michael has been a very active Paediatrician and clinician manager, initially at Sydney Children's Hospital, Randwick and then within SCHN. Michael's professional career spans over three decades and he has spent a lot of his daily

life advocating for safe and improved care for children, explaining the differences in their needs, and ensuring a fair allocation and effective use of resources. Michael holds tertiary qualification in Paediatrics and management and is on several health related Boards.



DIANNE PETRIE OAM

Dianne is the Executive Director of Genetic Alliance Australia has a BA majoring in Psychology and an Advanced Certificate in Counselling and has worked in the genetic community since 1983. Dianne's daughter Natasha has Williams syndrome and together with her husband Richard they started the Williams Syndrome support group. Dianne has successfully expanded Genetic Alliance Australia, achieving national and worldwide recognition. She established Genetic Alliance's

rare diseases contact register representing over 1,500 conditions and 3,500 contacts and in 2005 implemented a rural outreach program for carers. Dianne fields over 1,000 enquiries a year regarding the diagnosis of a genetic condition and has organised and conducted hundreds of genetic-specific seminars, bringing families and individuals together with professionals in a non-clinical setting.

Dianne has been asked to present both here and internationally at many conferences and events. She is a consumer representative on a number of committees - The NSW Newborn Screening Advisory, Genetics Network Executive Committee, Lymphoedema Alliance and Stepping Stones Triple P National Reference Group. Dianne has been involved in research projects with Macquarie and Monash Universities and through advocacy has helped to shape government policy. Dianne was awarded an OAM on the Queen's Birthday Honour's List 2006 in recognition of her work.



TANSEL ERSAVAS

Tansel is a computer scientist and the father of Alan who has a rare genetic condition which impacts his immune system. Before being challenged with a medical mystery that plagued his son, he was working on complex systems as a project leader, consultant, and trainer for several major companies and some start-ups.

Over his 27-year career, he worked around the globe and mastered a wide range of languages and systems, and various branches of Artificial intelligence (AI). He has been invited to international conferences as a speaker, or to run workshops and short courses. He is currently working at Garvan Institute of Medical Research to analyse bioinformatics data using cutting edge AI tools.

SPEAKERS



TOMMASINA OWENS

Tommasina is a sibling of two beautiful individuals with an intellectual disability. She is also a proud daughter of a mother with an intellectual disability. Tommasina has a Masters in Psychology (Educational and Developmental) and currently works as a school psychologist for the NSW Department of Education. She participates in reference groups for Siblings Australia and Carers Australia NSW. Tommasina has

worked in the disability sector for reputable government and non-government organisations. She is passionate about raising awareness on how disability impacts individuals and their family as well as exploring creative ways to promote inclusion.



JODIE MOK

Jodie completed her Diploma of Teaching and Bachelor of Education in Primary Education in 1993. She was a targeted graduate from the University of New South Wales. Jodie is currently teaching in a primary school situation, specialising in the areas of LaST and EaLD, working both with students experiencing learning difficulties and those from a non-English speaking

background. Jodie is the sole and younger sibling of Mathew Dunn, who has William's Syndrome.



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RADHIKA RAJKUMAR

Radhika is an Associate Genetic Counsellor employed by the Garvan Institute of Medical Research to recruit participants from the Genetics of Learning Disability (GOLD) service into the “The Economic and Psychosocial Impacts of Caring for Families Affected by Intellectual Disability (EPIC-ID Study)”. The study is exploring the economic and psychosocial impacts of intellectual disability (ID) on families and the potential outcomes of genomic testing on families. Radhika has a Masters in Human Genetics from India and a Masters in Genetic counselling from the University of Sydney. Radhika worked in a Genetics clinic and was part of the Down syndrome clinic in Child Trust Hospital in Chennai, India. Radhika has genetic counselling experience in prenatal, paediatric and adult genetics from working in Liverpool Hospital and The Children’s Hospital Westmead.



LYN QUIGLEY

Lyn and her husband Brian live on a farm at Gregadoo, just out of Wagga Wagga. Together, they have four children; Matthew, Anthony, Anna and Claire. Their two sons are affected by a gene mutation, RNF12, which follows the X-linked inheritance pattern. They both have an intellectual disability and mental health disorders. However, their two daughters who carry the same mutation are university educated, both graduating with double degrees. Lyn and Brian have three beautiful granddaughters, two are “gene” free but the third carries the RNF12 mutation. Lyn completed her teacher training at Wagga Wagga Teachers College in 1967. In recent times Lyn has been the president of the local Northcott Fundraising committee, a long term member of Abbeyfield Australia and is currently chairperson of Koorringal Abbeyfield. This committee has been formed to develop a six unit complex as a housing choice for younger adults in her Local Government Area living with a mild/moderate intellectual disability. The family has been part of the Genetics of Learning disability (GOLD) for 30 years.

MISSION STATEMENT

To facilitate support for those affected directly
or indirectly by genetic conditions
throughout Australia.

OUR SUCCESS

GENETIC ALLIANCE WOULD LIKE TO RECOGNISE AND THANK THOSE PEOPLE WHO HAVE PRESENTED AT PREVIOUS GDAA LAUNCHES:

1997: "LIVING WITH A GENETIC DISORDER – THE TENACITY OF PARENTS, INDIVIDUALS AND SUPPORT GROUPS."

Dr Chris Green

1998: "KEEPING THE FAMILY HEALTH STORY"

Dr Andrew Wilson
Sue Pinkerton

1999: "RESEARCH AND REALITY"

Hon Jillian Skinner MP
Dr Grant Sutherland AC
Prof. David Sillence
Dianne Petrie
Ros Matthews
Marlene Brightwell

2000: "GENETICS OF LEARNING"

Prof John Dwyer
Assoc Prof Kathryn North
Stuart Purvis-Smith
Karen Neeves
Dianne Petrie
Michael Reid

2001: "GENETICS AND THE FAMILY"

Prof. John Dwyer
Dr Andrew Wilson
Prof. Bruce Dowton
Dr Michael Buckley
Dianne Petrie
Lauren Gatt
Darren & Kelly Fisher

2002: "GENE THERAPY. STEM CELL RESEARCH & FUTURE TREATMENTS- WHAT'S IN IT FOR YOU?"

Assoc. Prof. Bridget Wilcken
Dr Ian Alexander
Cheryl & Ian Emmerich
Dr Andrew Elefanty
Robert Dettmann
Prof. John Dwyer
Matthew Begg
Dianne Petrie

2003 "GENETICS AND THE HUMAN SPIRIT"

Prof. Jenny Graves
Prof. Ken Kian Yong Ho
Richard Petrie
Dianne Petrie
Kate Murray
Michael Daniel

2004: "GENETICS: AN ADULT'S PERSPECTIVE"

Her Excellency Professor Marie Bashir AC, Governor of New South Wales
Prof. Gillian Turner
Dr John Christodoulou
Matt Laffan
Melanie Porter
Marlene Brightwell
Seanne Lavender

2005: "GENETICS: CHALLENGES FOR CARERS IN THE FUTURE"

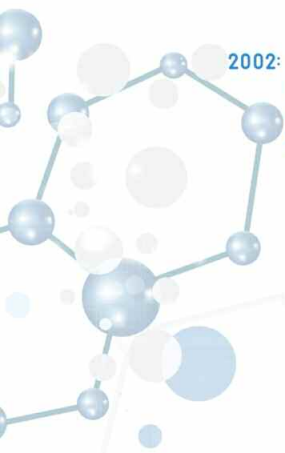
Michael Cori
Dr David Mowat
Laurie Taylor
Christine Regan
Felicity Maddison
Joan Hughes
Dr Jim Papadopoulos

2006: "GENETICS: ADULTS – BRIDGING THE GAP"

Mr. Peter Berner
Michael Cori
Dianne Petrie
Sue Pinkerton
Lynne Brodie
Debbie Colyer
Dr Jane Holmes-Walker

2007: "GENETICS: PAST, PRESENT AND FUTURE"

Prof Ron Trent
Michael Cori
Dianne Petrie
Lynne Foxall
Peter Allison



2008: "CELEBRATING 20 YEARS OF AGSA"

Dr Kris Barlow-Stewart
Sue Pinkerton
Ros Smith
Mandy O'Reilly
Glenn Fisher
Jan Cameron-Smith, Margo Latham
Dianne Petrie

2009 "CELEBRATING GREAT ACHIEVEMENTS"

James Castrission and Justin Jones,
Crossing the Ditch 2007
Kathy Secomb and Peter Secomb
(Williams syndrome)

2010 "THE GENETIC JOURNEY: RIGHTS, RESILIENCE & UNDERSTANDING"

Chair: Professor Les White AM
Alastair McEwin
Dr Alison Blatt
Tracey and Stephen Kable

2011 "FACING BARRIERS & OBSTACLES WITH INNOVATION & RESILIENCE"

Chair: Professor Les White AM,
Simon McKeon Australian of the Year
Jennifer Rollo OAM
Dr Peter Whiteman
Michael Cori
Laurie Taylor

2012 "CULTURAL DIMENSIONS IN GENETICS"

Donna Ingram
Dr Kevin Carpenter
Jennifer Edgar Genetic Alliance
Australia
Dr Emma Kowal
Libby Massey
Nancy Lucich
Mona Saleh

2013 "CELEBRATING 20 YEARS OF GENETIC DISORDERS AWARENESS WEEK: GENETICS IN 2013 – WHAT'S NEW"

Joanne Lind President, HGSA
Professor John Christodoulou AM
Kate Dunlop
Dianne Petrie OAM
Professor Graeme Morgan
Professor David Sillence AM

2014 "CHALLENGES IN HEALTHCARE"

John Sidoti MP – Member for
Drummoyne
Hon John Ajaka MLC NSW Minister for
Ageing, Minister for Disability Services,
and Minister for the Illawarra
Professor
Les White AM NSW Chief Paediatrician
Sean Murray CEO, Australian
Mitochondrial Disease Foundation
Megan Donnell Director, Sanfilippo
Children's Foundation
Professor David Thomas Director, The
Kingshorn Cancer Centre

2015 "MY GENOME – WHAT DOES IT ALL MEAN?"

The Hon. Jillian Skinner – Minister for
Health
Chair: Mary-Anne Young, Genetic
Counsellor GenomeOne
Professor John Mattick AO Executive
Director, Garvan Institute
of Medical Research
Stephan Damiani OAM – President,
Mission Massimo Foundation
Dr Cynthia Roberts Chief Risk Officer,
Genea

2016 "AUSTRALIAN PATIENTS & FAMILIES' PERSPECTIVES ON GENOME SEQUENCING: SURVEY RESULTS, TRANSLATION AND IMPLICATIONS"

Official Launch The Hon Pru Goward
MP, NSW Minister for Medical Research
and Assistant Minister for Health
Chair: Dr Michael Brydon Chief,
Executive of the Sydney Children's
Hospitals Network
Dianne Petrie OAM Executive Director
Genetic Alliance Australia
Dr Tony Roscioli Clinical Geneticist
/Genomicist,
Ayesha Wijesinghe Office Manager,
Genetic Alliance Australia
Tara Morrison Founder & President,
Homocystinuria Network Australia
Michelle O'Sullivan Occupational
Therapist & Advocate, Hypermobility
Connect

GENETIC ALLIANCE AUSTRALIA'S FINANCIAL SUPPORT GROUP & ORGANISATIONAL MEMBERS

(AS AT AUGUST 2017)

If you wish your organisation to appear on this list, please contact Genetic Alliance Australia or fill in the membership form and return to Genetic Alliance Australia with membership payment enclosed.

NB: THIS LIST REPRESENTS SUPPORT GROUPS AND ASSOCIATIONS ONLY. IN ADDITION TO THIS LIST OF MEMBERS, GA HAS ESTABLISHED A RARE DISEASES CONTACT REGISTER OF OVER 1500 GENETIC CONDITIONS REPRESENTING FAMILIES AND INDIVIDUALS SEEKING CONTACT.

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| ACT Genetics | Australia |
| ACI Transition Care | Children's Hospital at Westmead |
| ACT Muscular Dystrophy Association | Children's Medical Research Inst. |
| Alport Foundation of Australia | Children's Tumour Foundation of Australia (NF Australia) |
| Alzheimer's Australia ACT | CLIMB (Children Living with Inherited Metabolic Diseases UK) |
| AMEND | CJD Support Group Network |
| Angelman Syndrome Association Australia | Coeliac Society of NSW & ACT |
| Angelman Syndrome Association of WA | Congenital Adrenal Hyperplasia Support Group Australia |
| ARCAN – Australian Rare Chromosome Awareness Network | ConnecTed Foundation |
| Association for Children with a Disability | Contact a Family UK |
| Association for the Wellbeing of Children in Healthcare (AWCH) | ConnectGroups |
| Australian Galactasaemia Support Group | Cri Du Chat Support Group of Australia |
| Australia Kabuki Syndrome Network | Cystic Fibrosis NSW |
| Australian Leukodystrophy Support Group Inc | Cystic Fibrosis Victoria Inc. |
| Australian MPS and Related Diseases Society | Cystic Fibrosis Australia |
| Australian Pompe's Association | D.E.B.R.A. of NSW Inc. |
| Australian Thyroid Foundation Ltd | Darling Point Special School |
| Autoimmune Resource and Research Centre | Department of Clinical Genetics CHW |
| Batten Disease Support & Research Association (BDSRA) | Department of Clinical Genetics Liverpool |
| BEECH Australian Community INC | Disability South West Inc |
| Cancer Institute NSW | Down Syndrome NSW |
| Cardiomyopathy Association of Australia Ltd | Dystonia Network of Australia Inc. |
| Carer Support Program | Early Childhood Intervention Service Tasmania |
| CdLS Association Inc. | EURODIS (European Organisation for Rare Diseases) |
| Charcot-Marie-Tooth Association of Australia | Family and Community Services (FACS) |
| CHARGE Syndrome Association of Australasia | Family Insight |
| Child Development Service | Fragile X Association of Australia |
| Children's Eye Centre | FSHD Global Research Foundation |
| Children & Young People with Disability | Gaucher Association of Australia |
| | Genetic Alliance South Africa |
| | Genetic Alliance USA |
| | Genetic Alliance UK |



Genetic & Rare Disease Network (GaRDN – WA)
 Genetic Support Network of Victoria (GSNV)
 Haemochromatosis Australia
 Haemophilia Foundation NSW
 Health Consumers' Council of WA
 Hunter Genetics
 Homocystinuria Network of Australia Inc.
 HSP Research Foundation
 Hunter New England Research Ethics and Governance Unit
 Huntington's Disease Association (NSW)
 Huntington's Disease Association (NZ)
 Huntington's Disease Association (QLD)
 Hypermobility Connect
 IDEAS
 Immune Deficiencies Foundation Australia
 Indian Organisation for Rare Diseases
 KU Children's Services
 Lara Jean Association Inc.
 Leukodystrophy Australia
 Lifestart Nepean
 Lowes Syndrome Association
 Lucas Gardens School
 Lupus Association of NSW Inc.
 Machado Joseph Disease Foundation
 Malaysian Rare Disorders Society
 Marfan Support and Info in NSW
 Metabolic Dietary Disorders Association (MDDA)
 Motor Neurone Disease Association of NSW
 Murdoch Children's Research Institute
 Muscular Dystrophy Association of NSW
 Muscular Dystrophy Association of NZ
 NORD (National Organisation for Rare Diseases)
 Northcott
 Northern Sydney Central Coast Area Health Service
 NSW & ACT Hereditary Cancer Registry
 NZORD (New Zealand Organisation for Rare Disorders)
 Osteogenesis Imperfecta Society of Australia
 OzE - Ectodermal Dysplasia Support Group
 Parent to Parent (NZ)
 Physical Disability Council of NSW
 Physical Disability Council of WA
 Pierre Robin Australia
 PKU Association of NSW
 Prader-Willi Syndrome Association of Victoria
 Prader-Willi Syndrome Association of NSW
 Psuedoxanthoma Elasticum (PXE) Support Group
 RareConnect
 Rare Diseases UK
 Rare Voices Australia
 Retina Australia (NSW) Inc.
 Royal Melbourne Hospital Genetics
 Royal North Shore Department of Clinical Genetics
 SANDS
 Sanfilippo Children's Foundation
 Sanofi Genzyme
 Sarcoidosis Lyme Australia
 Self Help Qld Inc
 Short Statured People of Australia
 Special Needs Support Group
 Spinal Muscular Atrophy Association of Australia
 St Paul's College Library
 State Library of Victoria Serials
 Sutherland Shire Libraries
 Sydney Ultrasound for Women
 Tasmanian Clinical Genetics Service
 Taiwan Foundation of Rare Disorders
 Teen Time
 Thalassaemia Society of NSW
 The Australian Arthrogyposis Group (TAAG)
 The Australian Thyroid Foundation Limited
 The Centre for Genetics Education
 The Children's Hospital at Westmead
 The Children's Tumour Foundation of Australia - NF Australia
 The Chromosome 18 Registry & Research Society (Australia) Inc.
 The Sydney Children's Hospital
 Tuberous Sclerosis Australia
 University Western Sydney Journals/Ward Library
 Unique UK
 VCFS and 22q11 Foundation
 Villa Maria (Disability, Aged Care and Education Vision Australia)
 Wellington Huntington's Disease Assoc. (Inc.) (NZ)
 Williams Syndrome Australia
 Williams Syndrome Family Support Group VIC
 Women with Disabilities Australia
 Zach Armstrong Fund Inc

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GENETIC ALLIANCE AUSTRALIA

- Member of the International Alliance of Patient Organisations (IAPO)
- Orphanet
- European Organisation of Rare Diseases (EURORDIS)
- Health Consumers NSW

GENETIC ALLIANCE IS A CONSUMER REPRESENTATIVE ON:

- Genetics Network Executive Committee
- NSW Newborn Screening Advisory Committee
- Consumer Health Forum - Voting Member
- Stepping Stones Triple P (SSTP) National reference Group

GENETIC ALLIANCE AUSTRALIA BOARD MEMBERS

Richard Petrie – Treasurer, Brenda Phillis – Secretary, Wendy Bruce, Anne Cutler, Kate Dunlop – Chair, Jane Fleming, Carolyn Shalhoub and Ann Mulder – Rural Outreach Representative

www.geneticalliance.org.au

FUNDED BY



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