



Australian Paediatric Surveillance Unit

1993 to 2018

*Celebrating 25 Years of research,
advocacy and care for children living
with rare conditions*



The APSU was founded in 1993 in recognition of the gap in knowledge about the epidemiology of rare childhood disorders in Australia. Through the APSU, paediatricians have made a significant contribution to closing that knowledge gap, and this has improved the health and well-being of children nationally.

In 25 years, over 300 researchers have used the APSU to conduct more than 60 studies on rare disorders including infections, genetic diseases and injuries. It has been a great pleasure to have been involved in the important work of the APSU. I thank all Staff, Study Investigators, Committee members and Paediatricians throughout Australia for their contributions to the success of the APSU and look forward to an on-going collaboration.

A handwritten signature in black ink, appearing to read 'Elizabeth Elliott'.

Professor Elizabeth Elliott

10 May 2018

Sunday 13th May 2018

**Charles Perkins Lecture Theatre,
The University of Sydney**

Session 1

Chair: Professor Paul Colditz

09:30am

Professor Carol Bower

Welcome

09:35am

Professor Elizabeth Elliott

Celebrating 25 years of APSU achievements

10:05am

A/Professor Yvonne Zurynski

Living with a rare disease

– experiences of families and clinicians

10:30am

Mr Steve Waugh

Meeting the needs of children living
with rare disease and their families

10:45am

Dr Sloane Madden

Early onset eating disorders in young children:
role of surveillance to support policy and care

11:00am

Morning Tea

Session 2

Chair: Professor Natasha Nassar

11:30am

Dr Marcel Zimmet

Fetal Alcohol Spectrum Disorder
– from surveillance to policy

11:45am

Professor Elizabeth Elliott

Female Genital Mutilation
– do Paediatricians see this rare injury?

12:00pm

Professor Heather Jeffery

Sudden Unexplained Death
in Australian infants

12:15pm

Professor Helen Leonard

RETT Syndrome: from surveillance to
genotype-phenotype characterisation

12:30pm

Awards

12:35pm

Lunch

Session 3

Chair: Professor John Christodoulou AM

1:30pm

Professor Cheryl Jones

Consequences of congenital infection:
HSV, Rubella, Varicella, HIV, cCMV and Zika

1:50pm

Dr Anne Durkan

Long-term outcomes after atypical HUS

2:05pm

A/Professor Julia Brotherton

Juvenile Onset Respiratory Papillomatosis

2:20pm

Professor Robert Booy

Severe Influenza in Australian Children

2:35pm

Dr Philip Britton

Encephalitis Study

2:50pm

Professor Carol Bower

Closing Remarks

3:00pm

Wine and cheese



Professor Carol Bower

Carol is the Senior Principal Research Fellow in Alcohol in Pregnancy and Fetal Alcohol Spectrum Disorder (FASD) at the Telethon Kids Institute and the Senior Principal Research Fellow at the University of WA. She has qualifications in medicine, epidemiology and public health. Her research has a strong focus

on investigating causes and effects of birth defects, translating research findings into public health policy and practice, and evaluating the effectiveness of that translation. Leading examples are the prevention of neural tube defects (promoting periconceptional folic acid supplement use and mandatory fortification of flour with folic acid) and research on prevention, diagnosis and management of FASD. She is Co-Director of an NHMRC Centre for Research Excellence, FASD Research Australia and previous Director of the WA Birth Defects Registry. She has long-term connections with the APSU – as a researcher (FASD, Rett Syndrome), Board member (Chair), and member of the Scientific Review Panel.



Professor Elizabeth Elliott AM FAHMS

Elizabeth is a Distinguished Professor in Paediatrics and Child Health at the University of Sydney; Consultant Paediatrician at the Sydney Children's Hospitals Network (Westmead); and an NHMRC Medical Research Futures Fund Practitioner Fellow. In 1993 she founded the Australian Paediatric Surveillance Unit for the study of rare childhood diseases and has since been its Director. She was involved in developing the International Network of Paediatric Surveillance Units (INOPSU) and in 2000 was elected its Convenor. She helped establish other surveillance units internationally and Paediatric Active Enhanced Disease Surveillance (PAEDS) for inpatient surveillance. Research interests include a range of rare diseases; Female Genital Mutilation; Fetal Alcohol Spectrum Disorder (FASD); and the health of remote-dwelling Aboriginal children, children in Immigration detention; and children in low-middle income countries including Vietnam. She is Co-Director of an NHMRC Centre for Research Excellence, FASD Research Australia, Head of the NSW FASD Assessment Service and lead in the development of a national FASD information website and a Register. She is a Fellow of the Australian Academy of Health and Medical Research, and recipient of the RACP's John Sands and Howard Williams' Medals.



Associate Professor Yvonne Zurynski

Yvonne Zurynski was the APSU Assistant Director from 2005 to 2016 and APSU Director of Research from 2016 to 2018. Yvonne has a PhD in Medicine and conducts epidemiological and health services research into chronic and complex conditions. She recently started a new position at the Australian Institute

of Health Innovation, Macquarie University, as the Associate Professor of Health System Sustainability and leads the coordinating unit of the NHMRC Partnership Centre in Health System Sustainability. She is also an Honorary Associate Professor, Discipline of Child and Adolescent Health, Sydney Medical School, The University of Sydney.



Mr Steve Waugh AO

Steve, former Australian cricket captain and one of cricket's most legendary players is renowned for his sporting achievements, business acumen and commitment to philanthropy, which have earned him accolades, recognition and awards worldwide.

Steve says meeting humanitarian Mother Teresa was the inspiration and catalyst to devote his life to charity.

During Australian tours he felt overwhelmed and helpless but meeting Mother Teresa added a fresh perspective. Here was a tiny, frail lady, bent over with arthritis, slowly shuffling around, oblivious

to her own ills and championing the welfare of others. "There was a calming aura about her," Waugh said. "When you were in her presence you definitely felt more serene and peaceful." Steve's first charity work was with leprosy sufferers and their families at Udayan in India, an association that is now into its 20th year.

He then decided he wanted to help sick children who through no fault of their own, 'fall through the cracks' and with his wife Lynette founded the Steve Waugh Foundation (SWF) in 2004 to provide life changing support and champion the stories for children and young adults with rare diseases. It is the only standalone Foundation of its kind in Australia and funds raised go directly to life-changing, individual grants for medicine, specialised equipment, treatment and items to improve the quality of life for a child or young adult affected by a rare disease. The focus is to promote wellbeing, to help children not only thrive, but in many instances survive. The Steve Waugh Foundation is 'somewhere to turn'. The SWF has had a long association with the APSU and was a partner in the ARC Partnership grant to study the Impact of Rare Diseases on families.



Dr Sloane Madden

Dr Sloane Madden is a clinician and researcher in Child and Adolescent Psychiatry, recognised for his expertise in the management of child and adolescent eating disorders. He is the Eating Disorder Service Co-ordinator for the Sydney Children's Hospitals Network and Co-Director of The Eating Disorder Service at the Sydney Children's Hospitals Network (Westmead Campus), the largest public eating disorder service in NSW.

Dr Madden's research has three major foci; the treatment of anorexia nervosa, identification and management of early onset eating disorders and the neurobiology of eating disorders. He has led two APSU studies on early onset eating disorders.



Dr Marcel Zimmet

Dr Marcel Zimmet is a Developmental Paediatrician working at the Fetal Alcohol Spectrum Disorder Service at The Sydney Children's Hospitals Network (Westmead) and at Royal Far West in Manly. He is the Chief Investigator for the APSU surveillance study on FASD and was a member of the Expert Review Panel in the development of the Australian Guide to the Diagnosis of FASD, published in 2016. In 2016, Marcel received a Fulbright Professional Scholarship for study in the USA.



Professor Heather Jeffery AO

Heather is an Honorary Professor of International Maternal and Child Health, Sydney School of Public Health and an Honorary Paediatrician Neonatologist at the Royal Prince Alfred Hospital, Sydney; and Gates Foundation Clinical Trials Manager in the Faculty of Engineering at the University of Sydney. She has extensive experience in Maternal and Child Health in over 10 low-middle income countries.

Research interests include: Development of a low cost method to measure body composition in nutrition assessment from birth up to

2 years, using Near Infrared (NIR) light reflection; Prevention of neonatal infection and neonatal mortality and morbidity in four South East Asian countries; Educational methods to translate best evidence into practice; Sudden Unexpected Death in Infancy: future directions for prevention and research. In 2016 she received the Sir Edward “Weary’ Dunlop Asialink Medal for enhancing quality of life in Australia-Asia.

Dr Anne Durkan

Anne Durkan is a paediatric nephrologist at The Children’s Hospital at Westmead. She obtained both her medical degree and her doctorate in medicine from Edinburgh University. She has previously worked in the UK and in Canada but has worked in Australia for over 10 years. Anne has been involved in several clinical trials of treatments for atypical haemolytic uraemic syndrome (aHUS) and has published APSU data on aHUS. She has research interests in various aspects of renal transplantation.



Professor Helen Leonard

Professor Helen Leonard has qualifications in medicine and public health. She holds a National Health and Medical Research Council Senior Research Fellowship and is a Principal Research Fellow at the Telethon Kids Institute in Western Australia. For 20 years her clinical practice involved management of children and adults with intellectual disability and rare diseases. During this time the major need for epidemiological, clinical and multidisciplinary research in this area became obvious. She has taken a leading role in developing research infrastructure to study intellectual disability in general and specific disorders like Rett syndrome and Down

syndrome. In 1992 she established the internationally unique population-based Australian Rett Syndrome Database and has since maintained this. In 2002 she set up the International Rett Syndrome Foundation Phenotype Database, InterRett, which has now collected data on over 2000 individuals from 48 countries.

Since commencing active research in 1998, she has co-authored over 250 journal articles and 12 book chapters. She is one of the most highly published authors on Rett syndrome over the last decade. Her strong collaborative approach has been evident throughout her career. She has previously used a Delphi approach to bring together from across the globe experts from different specialties to develop guidelines for clinical management. She has led two APSU studies providing novel data on Rett syndrome.



Professor Cheryl Jones

Professor Cheryl Jones is the Stevenson Chair of Paediatrics and Department Head, at the University of Melbourne, a world-renowned clinician-scientist, and the Executive Director of NHMRC Melbourne Academic Centre for Health (MACH). She is President of the Australasian Society for Infectious Diseases and, before her move to Melbourne in early 2017, held senior

executive roles with Sydney Medical School at the University of Sydney and was involved in Sydney's efforts to become an AHRTC. MACH is one of the two Victorian Advanced Health Research and Translation Centres (AHRTCs) designated by the NHMRC from a group of nine across Australia. It is a joint venture between 18 Victorian healthcare providers, research institutes and the University of Melbourne with a shared vision to better translate our

research into world-class healthcare delivery, more rapid commercialisation, and most importantly, improved patient and community outcomes. Cheryl is also an infectious diseases physician at Royal Children’s Hospital, Melbourne and an internationally recognised research leader in emerging viral infectious diseases with a PhD in Virology, particularly emerging infectious diseases of the brain. She has earned >\$25 million in peer reviewed funding including three NHRMC Centres for Research Excellence, most recently NHMRC CRE in protecting the public against emerging infectious diseases (CREID). She has led several APSU studies on congenital infections including Hepatitis C, Herpes Simplex Virus infection, Congenital Rubella syndrome and congenital cytomegalovirus infection.



Associate Professor Julia Brotherton

Associate Professor Brotherton is a public health physician and epidemiologist. As Medical Director of Registries and Research at the Victorian Cytology Service, she has substantial expertise and experience in the delivery and evaluation of population based screening programs and immunisation programs. She is a medical graduate

from the University of Newcastle, has a Masters degree in Public Health and a Doctorate of Philosophy from the University of Sydney, and holds a Fellowship in Public Health Medicine. She is an Honorary Principal Fellow at the School of Population and Global Health, University of Melbourne and a Visiting Professor, University of Malaya, Malaysia. For over ten years Julia has been involved in research and policy development informing the implementation

and evaluation of HPV vaccination programs in Australia. She is a lead investigator in Australian research which provides world-first evidence of dramatic declines in HPV infections and pre-cancerous cervical lesions in young women post-vaccination. She has 150 publications and is passionate about policy-relevant research. She is a chief investigator of the NHMRC Centre of Research Excellence in Cervical Cancer Control. She leads the APSU study on Juvenile Onset Recurrent Respiratory Papillomatosis.



Professor Robert Booy

Professor Robert Booy is the Head of Clinical Research at the National Centre for Immunisation Research and Surveillance of Vaccine Preventable Diseases (NCIRS). He holds an NHMRC Practitioner Fellowship. His research interests extend from understanding the genetic basis of susceptibility to, and severity of, infectious diseases, especially influenza, RSV and invasive disease caused by encapsulated organisms; the clinical, public health, and social burden of these diseases; and means by which to prevent or control serious infections through vaccines, drugs and non-pharmaceutical measures. Robert has led APSU studies on influenza, severe varicella infection and congenital varicella infection.



Dr Philip Britton

Dr Britton is a paediatrician and infectious diseases physician at the Sydney Children's Hospitals Network at Westmead, and a Senior Lecturer in the Discipline of Child and Adolescent Health with the University of Sydney. He has a Masters degree in Public Health and Tropical Medicine (James Cook University). He is an early career clinician

researcher with an interest in neurological infections, tropical infectious diseases, and international child health. His PhD investigated the clinical epidemiology of encephalitis in Australian children, using a national, active, hospital-based surveillance network (Paediatric Active Enhanced Disease Surveillance – PAEDS). He remains involved in active surveillance of childhood encephalitis, Acute Flaccid Paralysis (AFP – WHO polio surveillance), and influenza. He is the lead author on comprehensive guidelines for the investigation and management of encephalitis in Australia and New Zealand. His doctoral studies showed childhood encephalitis to be associated with epidemics of important emerging pathogens amongst children in Australia including EVA71 and HPeV3, and determined the magnitude of the contribution of influenza to this severe disease. He is a member the Australasian Society of Infectious Diseases (ASID), the Infectious Diseases Society of America (IDSA), ESCMID, the Public Health Association of Australia (PHAA) of the International Encephalitis Consortium.

Thank you to our supporters

