2023 Prader-Willi Research Symposium Australia

TIME	PROGRAM	SPEAKER
8:45	**Registration Desk Opens**	
9:00	**Opening Address**	Kathlene Jones, PWRFA
	PWRFA Research Roadmap	Dr Diane Webster, PWRFA
	Invited Speaker: Co-design in research	Prof Cathy Vaughan, MISCH, Melbourne University
	Family Panel: Life with PWS	
10:35	**MORNING TEA**	
11:00	**Plenary Speaker:** An odyssey in PWS research: from imprinting, genes, animal & cell models, to genetic therapies	Prof Rob Nicholls, University of Pittsburgh
	Invited Speaker: Innovation & drug discovery in the 'RNA Era'	Prof Sue Fletcher, Murdoch University
	Invited Speaker: PWS Centre of Expertise	A/Prof Honey Heussler, Children's Health, Qld Hospital and Health Service
	Invited Speaker: Generative AI in research & healthcare	Mark Cameron
12:45	**LUNCH**	
13:30	Do incretins have a place in the management of people with PWS?	A/Prof Tania Markovic, Royal Prince Alfred Hospital, Sydney
	Early Sleep Interventions to Improve Outcomes in Children with Neurodisability	Dr Moya Vandeleur, Royal Children's Hospital, Melbourne
	Understanding ventilatory control in children with Prader-Willi Syndrome	Okkes Patoglu, Monash University
	Investigating the epigenetic regulator smchD1 as a potential therapeutic target for the treatment of PWS	Meg Iminitoff, Walter and Eliza Hall Institute
	Mapping Cell-Type-Specific Transcriptomic Signatures of the Brain in Prader-Willi Syndrome	Shokouh Sabzevar, Murdoch Children's Research Institute
	Brain organoids for the study of PWS	Dr Pratibha Tripathi, Monash University
15:00	**AFTERNOON TEA**	
15:20	Quantitative gait analysis in children who have Prader-Willi Syndrome using a wearable sensor	Dr Claudine Kraan, Murdoch Children's Research Institute
	A qualitative exploration of parents' views about wearable devices for research and treatment monitoring of children with Fragile X, Angelman and Prader-Willi Syndrome	Rachel Xifaras, University of Technology Sydney
	Strength training for people with Prader-Willi syndrome: update from the PRESTO trial	Prof Nora Shields, La Trobe University
	The role of vasopressin in PWS behaviours	Dr Lauren Rice, Uni Sydney
16:15	Round table discussion – Breakthroughs in PWS research	
16:50	Grant round announcement and closing remarks	

Plenary Speaker

Prof Rob Nicholls

Professor, Division of Genetic and Genomic Medicine, Department of Pediatrics

UPMC Children's Hospital of Pittsburgh, and University of Pittsburgh

Dr. Rob Nicholls obtained his B.Sc. (Hons) undergraduate degree at the University of Melbourne (Australia), his doctorate (D.Phil.) at the University of Oxford (UK), and did his postdoctoral work at Boston Children's Hospital/Harvard University where in 1987 he began his research on Prader-Willi Syndrome (PWS).

In 1989, his work first identified a role for genomic imprinting in human disease with the discovery of uniparental disomy in PWS. Dr. Nicholls' career stops include the University of Florida (1990-1993), Case Western Reserve University
(1993-2000), the University of Pennsylvania (2000-2005), and since 2005 to Pittsburgh. Career achievements include a PWSA (USA) lifetime achievement award in 2013 for his work on PWS, and in late 2013 he cycled from the Pacific Ocean to the Atlantic Ocean (across the sourthern USA) in 28 days to raise awareness and funds for PWS research. His publications have 20,720 citations with a h-index = 71 (Google Scholar).

Additional major scientific discoveries in PWS include genetic subclasses, imprinted genes, DNA methylation, imprinting mechanisms, and establishment of cellular and animal models. The latter include β-cell and other in vitro models to dissect molecular and cellular mechanisms, in vivo mouse and swine models of PWS, and current assessment of novel approaches to potential gene therapy. Dr. Nicholls' lab also identified the OCA2 and SPG6 disease genes and mechanisms of DNA deletions in α-thalassemia and in PWS/Angelman syndrome.

Invited Speaker

Prof Sue Fletcher

Prof Sue Fletcher is a molecular and cell biologist with expertise in RNA biology and antisense technology. She completed her first degree at the University of Zimbabwe, and a PhD at The University of Western Australia, where she worked for 25 years before moving to Murdoch University in 2013. She has been an integral member of the internationally recognized group that pioneered therapeutic antisense oligomer development. In a 26-year partnership with Professor Steve Wilton, she contributed to the development of the antisense drugs Exondys51, Vyondys53 and Amondys45 to treat Duchenne muscular dystrophy. More recently, she established a collaboration with Professor Fred Chen to develop novel genetic drugs to treat inherited blindness, with their compound to treat retinitis pigmentosa type 11 being developed by PYC Therapeutics and Lions Eye Institute. Recently retired, Sue continues working with collaborators and her team at Murdoch University to develop antisense therapeutics for rare diseases.

Innovation and drug discovery in the 'RNA Era'

Emeritus Professor, Centre for Molecular Medicine and Innovative Therapeutics, Murdoch University, Perth Western Australia

Treatment options for rare diseases have been limited, although novel gene and molecular therapeutics are now demonstrating potential in the treatment of both inherited and acquired conditions. RNA therapeutics in particular hold unique promise in these diseases; however, achieving safe and efficient delivery of molecular drugs to deep target tissues such as heart, skeletal muscle, central nervous system and the retina remains a significant obstacle to clinical application. Antisense oligomers are a well-established class of RNA therapeutic whose potential is yet to be fully realised due to this delivery challenge, and to recurrent efficacy and tolerability failures.

Evolving oligonucleotide chemistries, conjugates and delivery modalities are addressing these challenges and delivering safer drug molecules. However, we are working in an environment beset by recent failures in the clinic, a poor appetite for biotech investment and a troubled biopharmaceutical sector. Despite this currently unfavourable ecosystem, opportunities exist to deliver patient impact and demonstrate the utility of oligonucleotide drugs.

Invited Speaker

Prof Cathy Vaughan

Professor Cathy Vaughan is a public health researcher with expertise in gender, violence prevention and response, sexual and reproductive health, and health systems. She has over 25 years' experience working in Asia and the Pacific, as well as Australia.

Cathy has extensive experience using qualitative and participatory research methods to work with groups experiencing health inequalities, including migrant and refugee women, women with disability, faith communities and young people, to generate evidence about the social and structural underpinnings of health. Her work is underpinned by a commitment to participatory approaches to health research and to strengthening research capacity.

Her current research focuses on the impact of intersecting inequalities on health, and in particular on the ways intersecting inequalities shape experiences of gender-based violence and the effectiveness of violence prevention and response initiatives. This includes a particular focus on technology-facilitated gender-based violence in Asia and the Pacific.

She uses her expertise in co-design approaches to work with community groups, NGOs and government departments to strengthen the use of research in the development, implementation and evaluation of health programs in settings across Asia and the Pacific.

Cathy has taught into the Master of Public Health program at The University of Melbourne since 2011, coordinating subjects on participatory research methodologies and community engagement, and on women's health. She co-leads the 'kNOwVAWdata' course to strengthen capacity to conduct safe, ethical and rigorous research on violence against women in low- and middle-income countries; and leads the Co-Design and Implementation Effectiveness node of the Methods and Implementation Support for Clinical and Health research hub (MISCH) at the university.

Invited Speaker

A/Prof Honey Heussler

Medical Director, Child Development Program,

Children's Health Queensland Hospital and Health Service

Prof Heussler trained at UQ and the Mater Children's Hospital before spending time in Melbourne at the RWH and RCH (Murdoch Institute). Her Doctorate in the Behavioural and Attentional consequences of Adenotonsillectomy was completed through the University of Nottingham where she was a Lecturer in Community Paediatrics.

She has a significant role in teaching and assessment in the Paediatrics and Child Health rotation and has a strong interest in medical education. Her clinical work involves children with a variety of Developmental and Behavioural problems as well as a number of clinics that specialise in Sleep disorders for this population. She also runs a specialised clinics for some genetic disorders and has a number of research interests that reflect her clinical practice.

PWRFA are working alongside Prof Honey Heussler and Queensland Childrens Hospital to establish Australia's first dedicated PWS Nurse coordinator. Prader-Willi Research Foundation Australia (PWRFA) announced seed funding to pilot the first location of a new program to improve the standard of care for babies born with the syndrome. The Prader-Willi Syndrome Centre of Expertise (PWS CoE) aims to build deeper clinical expertise and centralise care for those diagnosed with PWS and their families, with a specific service for newly diagnosed newborns.

Abstracts - Research Talks

A/Prof Tania Markovic

Royal Prince Alfred Hospital, Sydney

Tania Markovic is a Senior Staff Specialist at Royal Prince Alfred Hospital and a clinical associate professor at the Charles Perkins Centre, University of Sydney. She is a recognised expert in obesity with contributions including the co-writing of the Australian Obesity Management Algorithm and guidelines for the use of very low energy diets in people with diabetes and those with kidney disease.

She is a member of the International Network for Research, Management & Education on adults with Prader-Willi Syndrome (INfoRMEd-PWS) and together with international collaborators is producing guidelines on the management of all aspects of care of adults with PWS.

She obtained her MBBS Hons degree from the University of New South Wales, and specialised in endocrinology at St Vincent's Hospitals, Sydney. With an NHMRC Postgraduate Scholarship, she undertook PhD research at the Garvan Institute, investigating the effects of weight loss on carbohydrate and lipid metabolism in people with obesity and type 2 diabetes.

After seminal publications in influential journals (Diabetes Care, Diabetologia), she joined Royal Prince Alfred Hospital where she is the director of the Metabolism & Obesity Services, a multidisciplinary tertiary referral weight management service.

At the Charles Perkins Centre she leads the Clinical Trials Unit, which runs both researcher and sponsor-initiated clinical trials. Her main research interests relate to the nutritional management of type 2 diabetes, pre-diabetes and gestational diabetes and the management of obesity in adolescents and adults, including those with psychiatric disorders, intellectual disability and PWS.

Quoted from: https://theconversation.com/profiles/tania-markovic-1354234

Okkes Patoglu
Monash University

UNDERSTANDING VENTILATORY CONTROL IN CHILDREN WITH PRADER-WILLI SYNDROME

Author/s: Okkes R Patoglu[1], Margot J Davey[2], Gillian M Nixon[1,2], Bradley A Edwards[3], Rosemary SC Horne[1]

[1]Department of Paediatrics, Monash University, Melbourne, Australia; [2]Melbourne Children's Sleep Centre, a Children's Hospital, Melbourne, Australia, Sleep and Circadian Medicine Laboratory; [3]Department of Physiology, School of Psychological Sciences and Turner Institute for Brain and Mental Health, Monash University, Melbourne Australia

Presenting Author: okkes.patoglu@monash.edu

Background:

Children with Prader-Willi syndrome (PWS) are at risk of both central and obstructive sleep apnoea (OSA), and have abnormal respiratory control during wakefulness. Many children with PWS are treated with growth hormone (GH) however, GH has been associated with the development of OSA. In this pilot study, we aimed to determine whether sleep and respiratory control (i.e. loop gain, LG) characteristics were altered by GH in children with PWS.

Methods:

Pre- and post-GH polysomnographic data in children (aged 0-18 years) were retrospectively collected. LG was quantified in periods of NREM sleep by fitting a model of ventilatory control to the respiratory pattern following spontaneous sighs.

Results:

Results (n=15, range 5-134 months, 46.7%F) were compared pre- and post-GH. No significant differences in total sleep time (474 [447, 498] vs 475 [451, 495.5]; p=0.81), percentage time in NREM and REM sleep, the obstructive apnoea-hypopnoea (OAHI) index (0.0 [0.0, 0.4] vs 0.3 [0.0, 0.7]; p=0.15) or the central apnoea-hypopnoea index (CAHI) (3.0 [0.7, 6.3] vs 2.8 [1.4, 4.6]; p=0.63) were found however, 1 child developed OSA. GH had no impact on LG (0.4 ± 0.1 vs 0.4 ± 0.1; p=0.99) and the change in OAHI/CAHI showed no correlation with the change in LG.

Conclusions:

In this small cohort of children, the administration of GH had no impact on sleep and respiratory characteristics (including LG). A larger sample size will be required to gain a more comprehensive understanding of the impacts of GH on ventilatory control in children with PWS.

Funded by the Prader-Willi Research Foundation Australia.

Research Theme: **Sleep & respiratory**

Meg Iminitoff
Walter and Eliza Hall Institute

INVESTIGATING THE EPIGENETIC REGULATOR SMCHD1 AS A POTENTIAL THERAPEUTIC TARGET FOR THE TREATMENT OF PWS

Author/s: Megan Iminitoff[1,2], Tamara Beck[1,2], Andrew Keniry[1,2], Kelsey Breslin[1,2], James Murphy[1,2], Marnie Blewitt[1,2]

[1]WEHI, Melbourne, VIC, Australia, [2]The Department of Medical Biology, The University of Melbourne, VIC, Australia

Presenting author: iminitoff.m@wehi.edu.au

Abstract: Smchd1 is an epigenetic repressor, targetable by small molecules and known to play a role in silencing PWS cluster genes on the maternal allele in mice. We are working on proof-of-concept data for gene reactivation using SMCHD1 as a target for potential therapy.

Removal of Smchd1 in vitro has been shown to result in activation of the silent maternally inherited copy of PWS genes at the proximal end of the cluster. Using a reporter mouse model for Magel2 expression we have shown that in line with previous data deletion of Smchd1 can result in reactivation of maternally inherited PWS genes in vivo in the brain, specifically in the hypothalamus.

Furthermore, in hypothalamic tissue we see reactivation of PWS genes not only from the proximal end of the cluster but from the distal end as well, with seemingly no effect on Ube3a expression. To determine whether the level of reactivation we observe is sufficient to rescue disease phenotypes in the Magel2 paternal-null model we have begun behavioural and motor testing using mice that have had Smchd1 deleted in the brain, looking at features of PWS within the cohort.

From this we aim to increase understanding of the molecular mechanisms at work within the PWS cluster and to confirm viability of SMCHD1 as a target for epigenetic therapy of PWS.

Research Theme: **Gene Activation**

Shokouh Sabzevar
Murdoch Children's Research Institute

MAPPING CELL-TYPE-SPECIFIC TRANSCRIPTOMIC SIGNATURES OF THE BRAIN IN PRADER-WILLI SYNDROME

Author/s: Shokouh Shahrokhi Sabzevar[1,2], Candice Dyson[1,2], Mirana Ramialison[2,3,4], Michael See[3,5], Fernando J. Rossello[6], Anthony J. Hannan[7,8], Melissa C. Southey[9,10,11], Olivia Veatch[12], David J. Amor[1,2], Merlin G. Butler[12], David E. Godler[1,2]

[1]Murdoch Children's Research Institute, Parkville, VIC 3052, Australia, [2]Department of Paediatrics, University of Melbourne, Parkville, VIC 3052, Australia, [3]reNEW Novo Nordisk Foundation Centre for Stem Cell Medicine & Stem Cell Biology, Murdoch Children's Research Institute, Parkville, VIC 3052, Australia, [4]Australian Regenerative Medicine Institute, Monash University, Clayton, VIC 3800, Australia, [5]Monash Bioinformatics Platform, Monash University, Clayton, VIC, Australia, [6]Centre for Cancer Research and Department of Clinical Pathology, University of Melbourne, Parkville, VIC, Australia, [7]Florey Institute of Neuroscience and Mental Health, Parkville, VIC 3010, Australia, [8]Department of Anatomy and Physiology, School of Biomedical Sciences, University of Melbourne, Parkville, VIC 3010, Australia, [9]Precision Medicine, School of Clinical Science at Monash Health, Monash University, Clayton, VIC 3168, Australia, [10]Department of Clinical Pathology, The University of Melbourne, Melbourne, VIC 3010, Australia,[11]Cancer Epidemiology Division, Cancer Council Victoria, Melbourne, VIC 3004, Australia,[12]Department of Psychiatry and Behavioural Sciences, Medical Centre, University of Kansas, Kansas, KS, USA

Presenting Author: shokouh.shahrokhi@mcri.edu.au

Abstract: Abnormal expression of genes at the 15q11-q13 locus causes Prader-Willi Syndrome (PWS). We used single nucleus RNA-sequencing (snRNA-seq) to establish for the first time a cell-type-specific map of 34 differentially expressed protein-coding genes at this locus in prefrontal cortex. Matched brain samples were used from 4 PWS individuals with deletion (ages 31 to 52 years), 4 individuals with non-deletion (ages 31 to 44 years), and 4 neurotypical controls (ages 32 to 42 years). Comparisons of normalized number of transcripts expressed from these genes were made between different cell types. The proportions of cells expressing specific genes in different cell types were also examined. Significant differences were observed between deletion and non-deletion groups in the proportion of non-neuronal cell types expressing UBE3A and HERC2 genes (-Log(P) = 1.5) and in the neuronal cell types for GABRB3 and GABRA5 (-Log(P) = 1.5). Of the non-imprinted genes, FAM189A1 and NSNMCE3 showed the greatest increase in the proportion of cells and decreased in normalized number of transcripts per cell expressing these genes in PWS as compared to control groups for most cell types (-Log(P) = 2.1). UBE3A expression in the non-deletion group was confirmed to be significantly elevated using droplet digital PCR (ddPCR) as compared to deletion (P=7.7e-4) and control (P=1.33e-6) groups. Understanding how dysregulation of these genes affects different cell types of the brain in different subtypes, may lead to development of new: (i) disease models for PWS to test novel therapies; (ii) prognostic biomarkers to enable earlier interventions and targeted treatments.

Research Theme: **Genetics**

Dr Claudine Kraan
Murdoch Children's Research Institute

QUANTITATIVE GAIT ANALYSIS IN CHILDREN WHO HAVE PRADER-WILLI SYNDROME USING A WEARABLE SENSOR

Author/s: Claudine Kraan[1,2], Laura O'Brien[1], Perrin Date[1] Emma K. Baker[1,2], Audrey Rattray[1], Morgan Sangeux[3], David J. Amor[1,2,4], and David E. Godler[1,2]

[1]Diagnosis and Development Research Group, Murdoch Children's Research Institute, Royal Children's Hospital, Melbourne, Victoria, Australia, [2]Department of Paediatrics, Faculty of Medicine, Dentistry and Health Sciences, University of Melbourne, Parkville, Victoria, Australia, [3]The University Hospital for Children, Basel, Switzerland, [4]Developmental Disability and Rehabilitation Research Group, Murdoch Children's Research Institute, Royal Children's Hospital, Melbourne, Victoria, Australia.

Presenting Author: claudine.kraan@mcri.edu.au

Background: Children with Prader-Willi syndrome (PWS) experience difficulties in motor and gait control that impact many areas of life, from cognition to physical fitness. Few studies have quantitatively described the PWS gait pattern.

Methods: A total of 10 children (6-16 years) with PWS (~60% female) performed a laboratory style short walk assessment and/or continuous "real-world" walking. Assessments were completed whilst wearing shoe-attached Physilog wearable inertial sensors. Data were compared against 28 age- and sex-matched typically developing (TD) controls completing the same walking assessments. Spatiotemporal data were extracted using a gait segmentation algorithm developed within the laboratory and corrected for leg length.

Results: Children with PWS were significantly slower than the TD group for leg length adjusted (LLadj) metrics of speed (stride time: $p<0.001$; cadence: $p<0.001$) and had shorter LLadj stride length ($p<0.009$). When compared to the TD group, children with PWS also spent more time in foot flat phase of the gait cycle ($p=0.0008$) and less time in push off phase ($p<0.0001$). PWS variability (an indicator of the overall smoothness of the gait) was increased compared to the TD group for multiple metrics (stride time: $p=0.0139$; cadence: $p<0.0001$; stance % cycle: $p=0.0052$; and stride length: $p=0.0185$).

Conclusion: Children with PWS have a different gait profile to that of TD children. This profile is easily measurable with wearable sensors over a period of a few minutes. Gait metrics measured via wearable inertial sensors may be a simple, straightforward clinical biomarker approach to monitor functional outcomes in children with PWS.

Research Theme: **Motor Control**

Rachel Xifaras
University of Technology Sydney

A QUALITATIVE EXPLORATION OF PARENTS' VIEWS ABOUT WEARABLE DEVICES FOR RESEARCH AND TREATMENT MONITORING OF CHILDREN WITH FRAGILE X, ANGELMAN AND PRADER-WILLI SYNDROME

Author/s: Rachel Xifaras[1], David Amor[2,3], *Erin Turbitt[1] and *Claudine Kraan[3,4]

**Senior author, [1]Genetic Counselling Discipline, Graduate School of Health, University of Technology Sydney, Sydney, Australia, [2]Neurodisability and Rehabilitation Research, Murdoch Children's Research Institute, Melbourne, Australia, [3]Department of Paediatrics, The University of Melbourne, Melbourne, Australia, [4]Diagnosis and Development, Murdoch Children's Research Institute, Melbourne, Australia*

Presenting Author: rachel.a.xifaras@student.uts.edu.au

Background: Wearable devices are tools to monitor function in individuals with neurodevelopmental disorders (NDDs). These can relay real-time data and provide objective and reproducible measurements. A better understanding of the opinions of parents of children with NDDs such as Prader-Willi syndrome (PWS) is required to ensure that wearable devices are sensitive to the unique needs of this group. We explored experiences and views of parents regarding use of wearable devices.

Methods: Semi-structured interviews were conducted with parents (n=12) of children with genetic NDDs. The inclusion criteria were parents with at least one child with a diagnosis of Fragile X, Prader-Willi, or Angelman syndromes, who had experience with research using a wearable device. Thematic analysis was used to code and identify patterns across the transcribed dataset.

Results: Three themes were identified. First, parents placed value on how easily the technology could be incorporated into existing routines. Factors that incentivised parents to uptake wearable devices included feasibility, non-invasiveness, and ability for use in the home. Second, parents' decision-making processes involved reflection on positive and negative experiences in healthcare and research. Third, due to the unique features of the cohort, there is high value in open dialogue between parents and researchers in the early design and rollout of new technology.

Conclusions: There are unique factors associated with the behavioural phenotypes of children that impact uptake and success of wearable devices in the NDD population. Shared decision-making between researchers and parents is likely to facilitate the use of wearable devices for children with PWS.

Research Theme: **Qualitative research with parents, motor control**

Prof Nora Shields
La Trobe University

STRENGTH TRAINING FOR PEOPLE WITH PRADER-WILLI SYNDROME: UPDATE FROM THE PRESTO TRIAL

Author/s: Nora Shields,[1] Alesha Southby,[1] Tania Markovic[2,3] Viral Chikani,[4] Georgina Loughnan,[2] Janet Franklin,[2] Lauren Rice,[3] Susan Blair,[5] Luke Prendergast,[6] Jennifer J. Watts,[7] Kim Bennell,[8] Nicholas F. Taylor[9,10]

[1]Olga Tennison Autism Research Centre, La Trobe University, Melbourne, [2]Metabolism and Obesity Services, Royal Prince Alfred Hospital, Sydney, [3]Brain and Mind Centre, The University of Sydney, [4]Department of Endocrinology, Princess Alexandra Hospital, Brisbane, [5]Prader-Willi Research Foundation of Australia, [6]Department of Mathematics and Statistics, La Trobe University, Melbourne. [7]Deakin Health Economics, Deakin University, Melbourne, [8]Department of Physiotherapy, University of Melbourne, Melbourne, [9]Department of Physiotherapy, La Trobe University, Melbourne,[10]Allied Health Clinical Research Office, Eastern Health, Box Hill.

Presenting Author: n.shields@latrobe.edu.au

Objective: To present an overview of participation in the PRESTO trial by people with Prader-Willi Syndrome (PWS).

Design: A multi-site randomised controlled trial

Participants and Methods: We recruited people with PWS from across Australia aged 13-60 years. Participants were randomised to receive either progressive resistance training (experimental) or non-progressive exercise (placebo-control). Both groups trained twice weekly for 24 weeks (48 sessions) at a community gymnasium with an exercise professional using pin-loaded weight machines. Assessments were completed at baseline (week 0), post-intervention (week 25) and 6 months post-intervention (week 52). Participant characteristics and attendance at the intervention are described.

Results: The trial commenced in February 2020 and was delayed due to COVID-19 pandemic restrictions. Recruitment closed in September 2022 with 58 enrolments from across seven states and territories. Participants (31 female, mean age 24 years) reported their type of PWS as paternal deletion (n=34), maternal uniparental disomy (n=18) or unknown (n=6). Twenty-three participants were receiving growth hormone at recruitment. Forty participants lived with family and 18 in assisted-living residences. Participants attended an average of 32 ± 1 gym sessions between week 0 and week 25 (66% attendance rate). COVID-19 mitigation measures (e.g. lockdowns) disrupted the intervention phase for 13 participants. All remaining assessments will be completed by September 2023.

Conclusion: The PRESTO trial met its recruitment target despite the challenges of the COVID-19 pandemic. On completion, it will be the largest trial internationally investigating efficacy of an exercise intervention for people with PWS.

Research Theme: **Exercise**

Dr Lauren Rice

University of Sydney

Dr Rice is a Research Fellow at the Brain and Mind Centre, University of Sydney and The Children's Hospital Westmead Clinical School. She is an investigator on a number of important PWS research projects.

Dr Rice and her team were awarded a 2019 research grant by the US-based Foundation for Prader-Willi Research. The funded study will provide fundamental information about PWS brain connectivity and evaluate a potential treatment for challenging PWS behaviour.

Dr Diane Webster

Research Director,
Prader-Willi Research Foundation Australia

Dr Diane Webster (She/Her) is a PhD-qualified biomedical science professional. She is the Research Director for the Prader Willi Research Foundation Australia (PWRFA), where she has developed and maintained a pipeline of research projects which focus on improving outcomes for people with Prader Willi Syndrome (PWS). Her career includes extensive experience in the Academic and Not-for-Profit sectors.

She also has expertise in biosafety, risk management and gene technology.
Her research is multidisciplinary and collaborative in nature, and has encompassed molecular biology, plant biotechnology, virology, vaccine development, and Prader Willi Syndrome (PWS), including epigenetics, genomics, neuroscience and community-driven person-focused research partnerships.

About Us

The Prader-Willi Research Foundation exists to improve clinical outcomes and deliver better treatments for people living with Prader-Willi Syndrome. We fund cutting-edge research which will help people with Prader-Willi Syndrome live independent lives, free from the most debilitating aspects of the condition.

PWRFA was founded in 2015 by our now CEO Kathlene Jones, mother to Chloe, who was diagnosed with PWS at 19 days old. A renowned scientist commented that Kath need not worry for Chloe's future as Prader-Willi is a prime candidate for gene therapy. After she discovered the limited Australian research into treatments for Prader-Willi Syndrome, PWRFA was born.

Prader Willi Syndrome (PWS) is a randomly occurring genetic condition affecting as many as 1 in 8000 babies. Due to some inactive genes on chromosome 15, people with PWS have complex medical needs, global developmental delays, challenging behaviours, mental illness and, the hallmark feature of the condition, a relentless feeling of starvation. The strength, resilience and courage people with Prader-Willi Syndrome show to do things others take for granted is remarkable.

Connect

Connect our PWS community, clinicians and scientists to drive cutting edge research to deliver life-changing treatments for our loved ones.

Empower

Empowering people with PWS and their families to raise their voice and drive research into life-changing treatments

Change

Fund research focusing on treatments for the most debilitating symptoms of PWS and the inactive genes.

For more information, visit: www.praderwilli.org.au