Prader-Willi Syndrome Association New Zealand

APPWS 2024 Conference Summary

The 6th Asia Pacific PWS Conference was held in Sydney and hosted by PWS Australia and PWRFA (Prader-Willi Research Foundation Australia). This summary is a compilation of reports, key points and takeaways by attendees. With special thanks to Lori Grinter for all the comprehensive day 1 research summaries on pages 1—4, and to Hayley Arnott for the summary of Mental Health and Behaviour on day 2, and for a summary of her presentation on Communication in Children.

Developing a comprehensive understanding of PWS: some important knowns and significant unknowns —Prof Tony Holland

- Future possibilities of brain scans to understand PWS further (e.g hunger, anxiety etc) - to be able to focus on individuals treatment options.
- Research showing PWS symptoms/experiences on brain scan (showed they were able to satiate - however with 3 x the amount of calories), and became hungry again quickly.
- Referred to S. Brown (2022) study - for further reading on this topic: In vivo neuroimaging evidence of hypothalamic alteration in Prader–Willi syndrome | Brain Communications | Oxford Academic (oup.com)
- Vagal nerve stimulation (VNS) study - didn't help the hyperphagia, but improved behaviours. This is now external VNS.
- The behavioural problems in PWS stemming from:

  Cognitively: less able to evaluate what is around (whether friend or foe) - re: flight or flight systems. This leads to anxiety.

  Neuropsychological: false inferences, therefore evaluation of 'the world' is incomplete, therefore response is inappropriate (too much/over the top or not enough response to things around them)

  Impaired emotional regulation: more work is needed (e.g. biomarkers) for this. Because they're always hungry, they're in "hunting mode" - referring back to flight or fight

- If we can treat hunger, we can likely treat some of the other behavioural issues in PWS: may remove them from the "hunting mode". (L. Grinter)

Day 1– Research

Elucidating the causes and consequences of sleep disturbances in individuals with PWS —Assoc Prof Olivia Veatch, University of Kansas

- Daytime sleepiness may be an issue with the circadian (daily) rhythm.
- Brains build connections during sleep (plasticity) - a reason why good quality sleep is important.
- Quality/quantity/time of sleep important for neurodevelopment (re: behavioural problems) and metabolism (sleeping less = increased BMI in non-PWS children)

- Issues with breathing and sleep apnoea seem to be more prevalent with the deletion subtype of PWS.
- Children with deletion had lower BMI (in general) than children with UPD in the healthcare data that was referenced.
- No difference in OSA (obstructive sleep apnoea) and CSA (central sleep apnoea) between PWS genetic subtypes.
- Small difference (however not statistically significant) that the deletion subtype has less sleep insomnia than UPD. (L. Grinter)

Integrating computational and *in vitro* approaches to achieve drug re-positioning for PWS —Prof Anthony White, QIMR

- Finding drugs that are already on the market to have applications/therapy for people with PWS (different approach than trying to bring new drugs to the market all the time).
- Drug repurposing allows drugs to be made available - bypassing funding/approval hoops (they've already gone through this process).
- This has been successful in other areas (including motor neuron disease, batten disease)
- Focusing on hyperphagia and neurological disorders. (L. Grinter)

The AI toolbox; building blocks to rethink your problem—Mark Cameron, Alyve

- Presented the idea of using AI to help pull papers and language (e.g. medical jargon) together to help put everything into one place.
- Idea is to help support parents to have more meaningful (and the right) conversations with health providers.
- Stef Cola (Ligantics) was also introduced during this presentation to introduce the digital health assistant they are formulating. Currently in a 'prototype'/learning model. Idea is to have a chat style platform that is being mediated by carers and clinicians to approve the answers to peoples questions in the chat. (L. Grinter)

Centre of Expertise

We are looking to connect more of our specialists in New Zealand with the CoE in Australia and have one of our leading endocrinologists connected already.
If you are a medical professional interested in this, or if you are a parent who would like to recommend a specialist, please contact us.

The National PWS Centre Of Expertise; a platform to accelerate research and translation—Assoc Prof Honey Heussler, QCH

- To develop national, clinical knowledge platform for standard setting to improve PWS patient care.
- Full lifespan focused (from infancy through to adulthood).
- Align internationally.
- Looking to include:
  - Integrate research and clinical trials
  - Advocacy / care model
  - "Starting early" - connect as soon as PWS diagnosis confirmed.
  - Standards of care - guides for families, and guide for clinicians
- However, need to ensure data talks to each other (e.g. between systems, input from clinicians etc)

(L. Grinter)

How can we build expertise? Connection, Literature, Clinical Experience.

Multi-disciplinary care creates better health outcomes and we have some main centre paediatric PWS clinics that work in this way in New Zealand. The connection of multiple disciplines can facilitate professional networking and learning, and the CoE also has this objective, except with a larger database to draw learning from. The CoE is a steadily growing, integrated virtual centre inspired by the Care Coordination Service in Pittsburgh and the work of Dr Jessica Duis in Colorado. The CoE is led by a steering committee, clinical advisory, employs a nurse navigator, and there are monthly complex case meetings. Ongoing data collection informs the development of care models.

Novel patterns of expression at 15q11-q13 in the brain and blood of individuals with Prader-Willi Syndrome—Assoc Prof David Godler, MCRI

Note: This session was quite heavy in genetics - apologies for my limited understanding of this session. If you're interested, search "David Godler PWS" for further reading on the impressive work he has - and is - doing for PWS and related genetic disorders.

- A/Prof Godler presented a study looking at mapping genes in the prefrontal cortex of 15q11-q13 protein coding genes for deletion and non deletion subtypes of PWS. Ultimately, this work has identified genetic differences between the subtypes of PWS, and hopes to use this knowledge with further research to support PWS subtype-specific therapeutics.
- Silencing of genes in 15q11-q13 is thought to be primary cause of PWS.
- Gene expression at 15q11-q13 in different cell types has not been mapped in the human brain in PWS.
- Study used prefrontal cortex (brain) samples

  Prefrontal cortex part of brain was used because they were available, and it has been reported to have lower grey matter in PWS compared to controls, and deletion compared to non-deletion subtypes (link added to this referenced research if interested.)
- Mapped 33 genes in PWS locus coding for proteins.
- Summary slide notes (from slides):

  Gain of expression observed for 5 non-imprinted genes (ATP10A, GABRA5, GOLGA5L7, and OCA2) in PWS silenced brain tissues of controls.

  GABRB3 standout in non-neuronal cell types e.g. in microglia from 5% of cell in controls increase to 43% in non-del, and 80% in del groups.

  GABRB3 major inhibitory neurotransmitter of the mammalian nervous system associated with the pathogenesis of Angelman syndrome, Prader-WIlli syndrome, nonsyndromic orofacial clefts, epilepsy, and autism.

  GOLGA6L7 standout for excitatory neurons - from 0% of cells in controls increase to 14% in the deletion group.

  The increase in UBE3A mRNA from ddPCR analysis, observed in non-deletion.

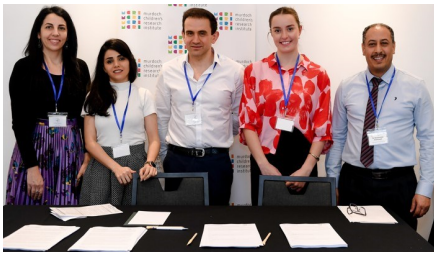
  This increase appears to be caused by increase in the number of cells expressing UBE3A at low level rather than increase transcription in individual cells (as would be expected for a paternally imprinted gene).

  This decrease in the number of UBE3A transcripts expressed in each cell was significantly associated with increase in proportion of cells expressing these UBE3A transcripts.

  These findings shed new light on molecular causes of PWS at the 15q11-q13 locus.

  If confirmed in other brain regions and larger cohorts, these results also have implications on the development of subtype specific therapeutics. (L. Grinter)

David Godler and the team from MCRI, Melbourne, Australia.



HERC2 and UBE3A gene expression in blood and brain tissues of individuals with Prader-Willi Syndrome—Shokouh Shahrokhi

Note: Shokouh is David Godler's phD student, presenting in-depth genetics beyond my understanding for parts of the presentation.

- pHD work looking at UBE3A and HERC2 - PWS subtypes (deletion and non-deletion), and linking severity of PWS:

  Adaptive behaviours, intellectual function, emotional/behavioural, food seeking, PWS behaviours and mental health
- Summary slide notes (from slides):

  Expression of UBE3A and HERC2 was increased in the non-deletion subtype (including UPD).

  Expression of both genes were related to phenotype severity in PWS.

  Increased UBE3A mRNA levels in PBMCs of children with non-deletion subtype were associated with enhanced autism features, and milder behavioural challenges.

  Increased HERC2 levels in PBMCs of adults with deletion subtype were associated with better intellectual functioning, and milder behavioural challenges.
- More research is needed (larger study) to indicate whether these findings are reliable. If they are, this could offer new prognostic markers, and therapeutics. (L. Grinter)

Investigating the epigenetic regulator SMCHD1 as a potential therapeutic target for the treatment of PWS—Meg Iminitoff, WEHI. (Prof Marnie Blewitt Lab phD student)

- Presented research using mice to investigate the therapeutic approach for PWS of activating the maternally (silenced) genes.
- SMCHD1 is the epigenetic repressor enzyme (e.g. is a gene that makes protein, that regulates gene activity).
- Investigating deleting SMCHD1 to activate some of the maternally coded genes.

    Mouse model is being used for proof of concept.

    In vitro will be first step for human research.

    Research needs to investigate safety (SMCHD1 potentially has other roles in genetics), and the efficacy as a therapy option.
- Mouse research to this is indicating:

    No major genome effects in mouse data.

    Indications of improvement to phenotypes (locomotor defects, motor co-ordination and grip/strength testing).

    Improvement in weight.

    Mice are surviving and living to adulthood.
- Mouse research is exploring deleting SMCHD1 at day 10.5 of pregnancy (when PWS genetic issue occurs). Future research required to investigate this concept postnatally. (L. Grinter)

Delayed gastric emptying and the safety of long-acting GLP-1 receptor agonists in Prader-Willi Syndrome (Results from the ENGAGE PWS Study) - Assoc Prof Alexander Viardot, Garvan Inst.

Associate Professor Alexander Viardot was the supervisor of a PhD study looking at the safety of GLP-1 receptor agonists in PWS. This class of medications lower blood sugar and are sometimes used in PWS for patients who have diabetes, i.e. dulaglutide and liraglutide. There have been isolated case studies of people with PWS showing benefit from liraglutide treatment, but large-scale research found on the whole that liraglutide did not impact weight loss, although a small subgroup experienced some reduction in hyperphagia symptoms. This class of medication is fast developing and a more effective GLP-1RA diabetes and weight loss treatment, semaglutide (Ozempic and Wegovy), has become well known in the general population. There is also now Tirzepatide, a GIP and GLP-1 receptor agonist. However, whilst liraglutide was found to be generally well tolerated in PWS, the most common side effects were GI related, and there are concerns that these new medications (not yet tested in PWS) may potentially slow digestion further resulting in more severe side effects.

The ENGAGE PWS study aimed to look at how safe long-acting GLP-1 receptor agonists are and focused on gastric motility as a safety aspect. They also aimed to determine the prevalence rate of delayed gastric emptying in PWS and to examine effects of treatment on appetite, behaviour and weight loss. Using gastric endoscopy, the study found that 3/13 or 23% of participants had significantly delayed gastric emptying. The study treated participants with the GLP-1RA Exenatide and observed some degree of weight loss in 50% (less than the weight matched non-PWS control group), but no change in hyperphagia. They found the effect on gastric delay during treatment was persistent and did not disappear after a few weeks, as it often does in the general population.

Alexander Viardot emphasised that patients should be screened for delayed gastric emptying before treatment with GLP-1 agonists. The study recommends a personalised approach to prescribing long acting GLP-1RAs and careful monitoring during treatment.

DCCR for PWS—Dr Yoon Hi Cho (on behalf of Soleno Therapeutics)

DCCR activates potassium ion (KATP) channels in the hypothalamus, pancreas and fat tissue. This improves leptin pathways, a hormone which plays a role in satiety and energy balance. It reduces circulating leptin levels and leptin resistance. DCCR also reduces the secretion of appetite stimulatory neuropeptides and transmitters, e.g. NPY / agRP. Other action improves insulin sensitivity.

DCCR phase 3 trials experienced covid disruption and statistical strength issues. The FDA requested more data be collected through C602, Soleno's open label extension / random withdrawal study. This new data has been compared alongside PATH for PWS, a natural history cohort within FPWR's global PWS registry. Dr Yoon Hi Cho reported that there are currently 650 active participants in the ongoing C602 / PATH study.

The average age of participants is 13.5years. In addition to significant improvements in hyperphagia, positive benefits were also reported on anxiety, rigidity, compulsivity, irritability, aggression and depression. DCCR was well tolerated with no serious adverse events reported. Low grade emergent events included hypertrichosis, peripheral edema and hyperglycaemia.

Soleno were a generous platinum sponsor of APPWS2024 and have worked hard to develop DCCR for PWS, potentially the first therapeutic option for the treatment of hyperphagia in PWS. The FDA decision on DCCR is expected Dec 27th.

Restoring the gut-brain pathways of hunger control using oral ARD-101—Dr Tien Lee, Aardvark Therapeutics.

Dr Tien Lee explained that there are two main drivers behind hyperphagia: reward which drives appetite (affected by GLP-1 and MC4R neurons), and pain stimuli which drive hunger (affected by CCK, vagal affect neurons). He said that there was dysfunction of CCK secretion in PWS and that ARD-101 helps regulate this activity. ARD-101 is a bitter agonist which binds to bitter-sensing type 2 taste receptors (TAS2R) in the gut, activating and anatomically restricting where CCK affects.

During phase 2 trials, 11 out of 12 participants had reduced hyperphagia (four participants had near complete resolution of symptoms.) ARD-101 also had positive effects on inflammation. Data will be available soon on an expansion trial with increased dose (200mg to 800mg).

ARD-101 is an exciting new avenue of research specifically targeting only the hunger drive, a different action to other hyperphagia treatments in development. We look forward to hearing more and are very grateful to Aardvark for gold sponsorship of the conference.

Guanfacine XR for the reduction of aggression and self-injury in PWS: results from a double blind placebo controlled clinical trial—Dr Deepan Singh

Dr Singh has been prescribing guanfacine for PWS patients in his psychiatric practice and seen positive results on aggression, impulsivity and self-injurious behaviours. He had previously published a retrospective cohort study on these results in 2019.

In August this year, the results of his double blind placebo controlled trial were published which confirmed the significant improvements he was observing in clinical practice. Both studies above had enrolled patients who were experiencing moderate to severe difficulties in the areas being evaluated.

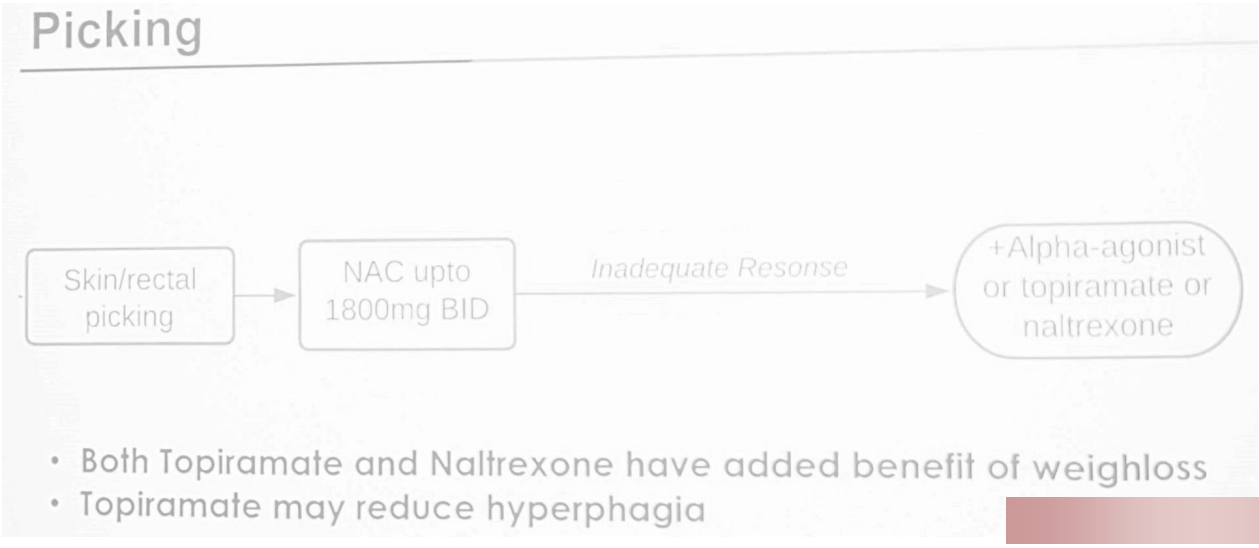
Dr Singh explained that guanfacine was well tolerated, although some people experience more fatigue on guanfacine. This sedation effect often reduces in a week, but for some people guanfacine is not suitable. He recommended starting treatment low and slow at 1-2mg dose. During the trial, the maximum dose was up to 4mg, but in clinical practice Dr Singh has used up to 7mg. Guanfacine should also not be used when there is evidence of psychosis.

Dr Singh remarked that the most commonly used medications for behavioural management in PWS can all have undesirable side effects (SSRIs - psychosis, stimulants - increased skin picking, antipsychotics - weight gain) and there is a lack of studies investigating their use in PWS. Guanfacine provides an alternative safe option and Dr Singh would argue that in many cases it should be tried first.

He also outlined the treatments currently available for skin picking in PWS (see slide below), emphasising that few options exist and that guanfacine provides a useful additional, safe treatment option.

Guanfacine is a non-stimulant ADHD medication and Dr Singh remarked that all people with PWS would likely have an ADHD diagnosis or have sufficient symptoms to meet prescription criteria.

Guanfacine is currently only obtainable in NZ under Section 29 of the Medicines Act for unapproved medicines (imported from Australia.) We believe it could potentially be approved by Medsafe within the next year, but funding may take a while longer.



The role of the immune system in neurodevelopment—Prof Russell Dale

This was an interesting subject presented by Prof Russell Dale from the University of Sydney because it reinforced the idea that medicine usually aims to treat symptoms, and that potential treatable causes can be missed.

Professor Dale talked about how recurrent infections are more common in patients with neurodevelopmental disorders and how infections and stress can affect the neurodevelopment of children's brains. Parents often talk about a worsening of their child's symptoms after infection or stress. PANS was an example given of this (Paediatric-onset Neuropsychiatric Syndrome).

We need to learn more about neurological and immunological processes and how stress affects the immune system. Observing causes of worsening symptoms is important, and we need to understand more about the immune system in children with NDD compared to neurotypical children and the use of treatments which may help, such as anti-inflammatories. Epigenetic therapies are an area requiring further research.

Missed diagnoses and health problems in adults with PWS—Kirsten Davidse (on behalf of the de Graff lab, Erasmus, Netherlands)

This study revealed alarming statistics of undiagnosed and untreated health problems in adults with PWS. Out of the 115 patients screened (median age 29), 61% had undiagnosed health problems; 1 in 4 of those patients had multiple health problems.

All males and 93% of females had hypogonadism—it was previously undetected in 52% and 33% respectively, 74% had scoliosis (undetected in 20%), hypertension 18% (undetected in 3%), hypercholesterolemia 19% (undetected in 6%), T2DM 17% (undetected in 5%), hypothyroidism 17% (undetected in 2%). The study also found that 8% had undiagnosed vitamin D deficiency. 45 patients underwent DEXA scans which revealed 3 new cases of osteoporosis and 8 new cases of osteopenia. 40/115 met clinical criteria for EDS and all of those cases had either untreated vitamin D deficiency, untreated male hypogonadism, or another treatable cause such as sleep apnoea, narcolepsia, nycturia, or use of medications that cause sleepiness.

The study provided screening protocol for diagnosis and treatment with the aim of reducing early complications and early mortality. This recommended screening algorithm can be viewed here.

This prevalence of undiagnosed health problems is likely to be a concern in NZ with many older adults not regularly seeing an endocrinologist for checkups and the unavailability of multidisciplinary PWS clinics for adults living with PWS.

Dr Jenny Downs spoke about the Australian PWS database and the importance of pooling data to answer a question. Aims of the database include documenting natural history, understanding variation in treatments and outcomes, investigating service needs, and supporting community involvement in ethically approved studies.

Open Panel: clinical trials recruiting in Australia

- ### Gastrointestinal transit and safety of tirzepatide in people with PWS—Assoc Prof Tania Markovic

  Tania explained that weight loss with tirzepatide (GIP and GLP-1 receptor agonist) has been quite remarkable in the general population (Mounjaro for diabetes and Zepbound for obesity). This trial is looking at safety in PWS, with a focus on gastric emptying and bowel function, in addition to investigating effects on weight, appetite, food seeking behaviour and metabolism.

- ### TEMPO trial evaluating pitolisant for excessive daytime sleepiness (EDS) in PWS—Ann Adee, Harmony B.

  Harmony Biosciences are recruiting for the phase 3 trial of pitolisant at multi-national sites, including Sydney and Queensland Australia. Participants need to be age 6+, have EDS, and no evidence of uncontrolled sleep apnoea. The study will run for 11 weeks with a 1 year open label extension. Interested persons should contact clinicaltrials@harmoneybiosciences.com

  Pitolisant is a medication available in the US, EU and UK for the treatment of narcolepsy and cataplexy and is sometimes prescribed off-label in patients with PWS for the treatment of EDS. It has shown positive results in PWS for EDS, learning and behaviour.

- ### Investigating a new target treatment for PWS: the acamprosate trial—Dr Lauren Rice

  Acamprosate is a medication used to treat alcohol cravings. It works by modulating the GABA system, restoring balance between glutamate and GABA neurotransmission. Lauren Rice explained that lower GABA levels have been observed in PWS and this can cause higher emotional behaviours. In ASD, unique brain response to GABA modulators has been observed. This trial proposes to recruit individuals 18-30 years who will undergo brain MRI, be treated with acomprosate for 10 days, and then repeat MRI.

- ### Does CBD reduce severe behaviour problems in children and adolescents with ID? - Assoc Prof Daryl Efron

  Cannabidiol (CBD) is being investigated further via placebo controlled trial at MCRI, Melbourne. Participants need to be 6-18 years, have severe behaviour problems and live in Victoria or NSW. For further information: mctrials@mcri.edu.au

Dr Maryssa Portelli, psychiatrist at RPAH Sydney's PWS Clinic, Assoc Prof Tania Markovic, endocrinologist and Director of Metabolism and Obesity Service RPAH, Assoc Prof Honey Heussler, developmental / sleep paediatrician at QCH and Medical Director of Child and Youth Community Health Services, and Professor Anthony Holland, psychiatrist and Emeritus Professor at the University of Cambridge, IPWSO president.

Day 2– Family, Allied Health, Support Providers

Prof Duang Wattanasirichaigoon—Genetics Overview

Dr Deepan Singh—Mental Health and Behaviour

A/Prof Tania Markovic—Lifelong Medical Support

A/Prof Laura de Graaff—Is GHT worth it for adults living with PWS?

P Chuang & J Mc Dowell—Empowerment through Physical Activity

Hayley Arnott & Anica Jansson—Encouraging Communication

Cate Fox—Nutrition for Healthy Children

Dr Janet Franklin—Healthy Eating for Adolescents and Adults

Grace Kelly—Call for Sibling Support Group

Prof Brendon Yee—Sleep Disorders in People with PWS

Prof Tony Holland—Present & Future Treatments

Cindy Adams Vining—Transition from School

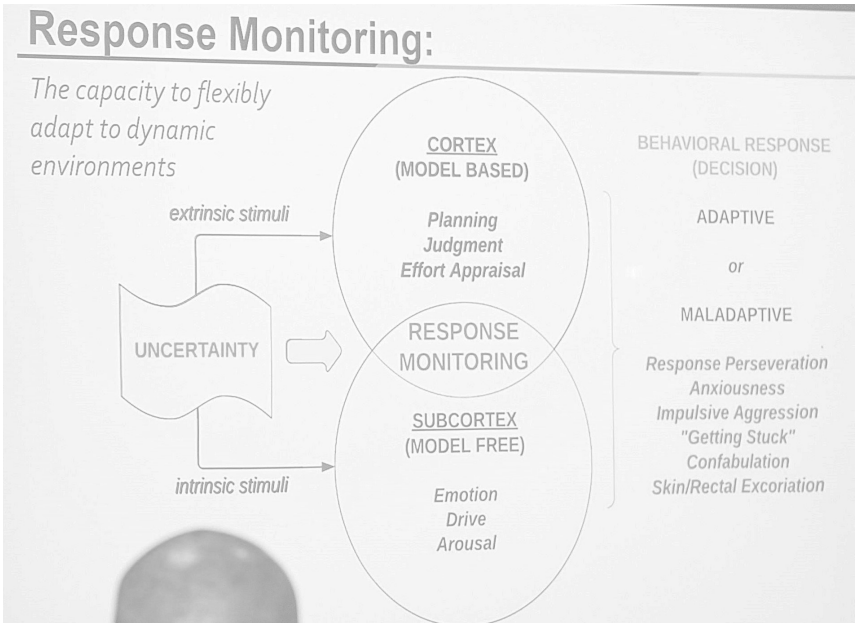
Diane Mangley—Nurturing Parental / Carer Wellbeing

Mental health and behaviour in PWS: a review for families —Dr Deepan Singh

Dr Singh first suggested that behaviour problems in PWS are a sign of success in other areas of treatment, meaning that people with PWS are living longer and fuller lives. He argued that this is a good problem to have.

Specific gene and neurobiological impairments (such as in the hypothalamus, grey matter and white matter of the brain) have been linked to specific behavioural manifestations – things like skin picking, aggression and impulsivity.

A brain's job is to respond, or in other words decide what to do next. We take information in, assess that information and then respond accordingly.
"Response monitoring" is our brain's capacity to adapt to our ever-changing environment and is what causes us to consider possible consequences before acting.



Our behavioural response in any situation can either be adaptive (appropriate) or maladaptive. Maladaptive behaviours as seen in PWS are things like response perseveration, anxiousness, impulsive aggression, getting stuck, confabulation and skin/rectal picking.

Response perseveration is the inappropriate repetition of a response (behaviour) despite the absence of reward (or the presence of negative consequences). For example, in PWS, this can be repetitive questioning despite receiving a negative response from caregivers.
Response perseveration is a direct a result of a range neurobiological abnormalities present in PWS.

Depression is more common in individuals with a higher I.Q. and Deepan recommends prioritising community involvement from a young age (that is, engaging with the PWS community).

Dr Singh spoke about medication being a type of technology. It is ever-evolving, and each is so unique and different that we should not write off all medication if one type has not been effective. "If you've tried one type of medication you've tried one type of medication". Medication choices will depend on whether there is mania or psychosis present, OCD symptoms or ADHD symptoms (though Dr Singh would argue that most people with PWS exhibit ADHD features). Daytime sleepiness and skin picking will also alter medication choice as a stimulant for example can make skin picking worse. (Figure 12.1 in his book was broken down in his slides, it is a flow chart for various medications). Dr Singh speaks highly of guanfacine. A very small number of patients were unable to tolerate the drug due to overwhelming exhaustion, however this was not the case for most. Guanfacine is not currently available in NZ.
(H. Arnott)

Is GHT worth it for adults living with PWS?—Assoc Prof Laura de Graff, Erasmus

This presentation reinforced the understanding that growth hormone therapy significantly improves body composition in adults with PWS by reducing fat mass and increasing lean body mass. There is strong evidence for this from multiple studies.

It is assumed that improvements in body composition increase exercise capacity.

As poor body composition is closely linked to the high incidence of cardiovascular morbidity in adults with PWS, treatment with GHT has the potential to reduce cardiovascular complications in this patient group.

There have been mixed findings on other potential benefits of treatment, i.e. on cardiovascular endpoints such as lipids / cholesterol. No significant differences in bone mineral density (BMD) have been reported.

Fewer studies have looked at cognition and QoL (Quality of Life) and results are inconclusive. Some significant improvements in areas of cognition have been observed, but QoL has been hard to measure and results vary.

GHT is a safe treatment for adults living with PWS.

Move to thrive: empowerment through physical activity—Pauline Chuang and J McDowell

Some of the issues affecting physical activity in PWS:

hypotonia,
impaired secretion of growth hormone,
high body fat percentage—2-3 times higher body fat percentage (40-50%).

Hypotonia means that all muscle movement requires more effort. Regular exercise builds muscle strength which counteracts the effects of hypotonia. Shoulders, hips and core strength will have the biggest effect on the activities of daily living. Core strength helps improve posture and balance.

Exercise has a positive effect on GH secretion and enhances the effects of GH treatment.

How much exercise is needed? Daily exercise of moderate to vigorous intensity. The type of exercise needs to be enjoyable and the total daily amount recommended is 60 mins.

Imagine a 1– 10 scale of exercise effort (how hard you work). The recommendation is that people with PWS need to work at 6-9 range.

It is important to do strength training as well as aerobic conditioning.

Communication in all ages: Children —Hayley Arnott

- Infant feeding and communication - focus on what they miss out on (oral stimulation, skin to skin, rocking, etc).
  Appetite comes in time, but OT, SLT, swallow studies are helpful.
- First 1000 days - critical for cognitive development (talk to your child and respond to them as much as possible).
- Late talkers - encourage speech sound play, use AAC if necessary, i.e. communication boards.
  (Talk to your SLP about trialling different options.)
  Use visuals, i.e. checklists, first/then, picture/word charts
- Speech clarity issues:
  - childhood apraxia of speech (CAS)
    45% global PWS registry
    speech motor planning disorder
    affects both speech clarity and prosody (intonation)
  - speech sound delays
  - velopharyngeal incompetence (VPI) - can develop over time
  - orofacial myology (Kolby Kail 2018 video FPWR)
- Conversation and social skills - all children (and adults) can benefit. Can use resources such as We Thinkers 'Social Explorers Curriculum', 'Social Problem Solvers Curriculum', or 'Social Skill Activities for Kids' by Natasha Daniels.

Communication in all ages: Adults —Anica Jansson

- Changes and transitions—clear information needed.
- Behaviours of concern can be communication, i.e. I'm hungry, I'm bored, I don't like it, I don't understand.
- Communication is state dependent—harder to understand others and communicate wants / needs when distressed.
- AAC (Augmentative and Alternative Communication) - hard copy visual support, device, sign, gesture/body movement
- Use communication supports to involve a person in decision making, i.e. choice boards, talking mats.
- Communicating change, expectation / target behaviour—provide easy read information, video modelling.
- Communicating food security—visual / pictorial meal plan
- Communicating routine—first/then board, now/next/later board, visual schedule board, who is here today board
- Problem solving—emotions board (feelings thermometer), something's wrong board, requests board (what will help).
- Mealtime risks: overfilling mouth, eating quickly, ineffective chewing, reduced coordination, muscle tone, distractibility.
- Mealtime support: visual reminders to slow down, pacing plates, smaller spoon, sit upright, head/neck alignment, modify food as needed—soft, bite-sized, moist etc., and minimise distraction.

Call for Sibling Support

Would you or your child be interested in joining a sibling support group? Grace Kelly from Australia is the sister of an adolescent younger brother with PWS and is keen to establish a support group.

This idea has been discussed previously in New Zealand too, but a group is needed to get this project off the ground. The idea being that there is no parental involvement, creating a safe space for siblings to share freely. (Moderated by adult siblings.)

Get in touch if interested and we can connect you with Grace!

Nutrition for Healthy Children—Cate Fox

Healthy Eating for Adolescents and Adults—Dr Janet Franklin

These were two great presentations which contained too much information to be able to summarise here, much of which was supported by visuals. If there is interest in learning more, we could hopefully arrange a presenter webinar, which would also allow opportunity to ask questions.

A key takeaway for me from Cate Fox's presentation was the use of different hand measures for red meat, chicken and fish. There was also an emphasis on the importance of good nutrition and quality versus quantity.

Dr Franklin talked about low energy density foods and showed comparisons of 200Kcal in different foods. She discussed a recommended PWS plate model.

Sleep Disorders in People with PWS—Prof Brendon Yee

- Central Sleep Apnoea (CSA) - common in infants, mostly gets better on its own, usually no treatment needed.
- Obstructive Sleeo Apnoea (OSA) - more than 50% of children will have this, may cause EDS (excessive daytime sleepiness)
- Sleep related hypoventilation, or a combination.
- Medications:
  Modafanil—some success
  orexin agonists—potentially a promising emerging treatment for promoting wakefulness

Transition from School—Cindy Adams-Vining

Cindy's presentation reinforced that transition is different in PWS for several reasons, but a main issue being that transition to adulthood usually focuses on increasing independence and this means increased access to food in PWS if not managed well.

Other differences to consider might include anxiety, difficulties coping with change, inflexibility, challenging behaviour and a higher risk of mental health complications.

It's also important to understand that adolescence can be a notoriously difficult time for people living with PWS due to pubertal differences, widening peer gaps, relationship problems, potential cessation of GHT, and a loss of experienced support systems (school, paediatric health team). People with PWS have dreams and ambitions like everyone and these might be grandiose or unrealistic, which can lead to disappointment. A strong desire for independence that is not realised will also lead to frustration.

Any changes need to be well planned and managed. It is essential that support systems have good knowledge and understanding of the syndrome. Cindy stressed that parents need to stay involved and keep informing and advocating for their child.

Cindy recommended making use of transition services who work with individuals to make a transition plan based on goals and aspirations. They will help look for opportunities, and evaluate things like accessibility and support needs.

Some of the problems encountered are limited professional knowledge amongst adult supports, limited residential placement options, and limited employment / vocational opportunities. It may be necessary to think 'outside the box' and to create opportunities, i.e. micro enterprises.

When your child moves to live in a residential service, this can be a huge leap of faith for parents. Understand that the service provider may have limited skill level / experience at entry into service, don't assume others will read information provided or online, remember that you are the expert in your child and ensure that you are kept informed. Compulsory regular training is essential—ensure this happens. Ensure there are regular staff meetings and system reviews.

Parents may wish to look at guardianship laws and future plan financial arrangements. The key thing is to plan ahead (before a crisis occurs).

Prof Tony Holland on present and future treatments:

"This is the most exciting time for PWS research. The potential for new treatments that could be transformative for people with PWS is unprecedented. I feel more optimistic now than in any time in my career."