PWS NEW ZEALAND
Prader-Willi Syndrome Association NZ
ADVOCACY EDUCATION SUPPORT

March Newsletter

We hope everyone is remaining well during this Omicron outbreak. Data collected so far (French study and small IPWSO survey) has shown that PWS itself does not appear to be a risk factor for serious illness with COVID-19, and those infected have experienced more favourable outcomes than expected. However, there will be concerns about our more vulnerable members so we look forward to the arrival of oral anti-viral medications which will be available sometime from the end of April. We submitted to PHARMAC's consultation on the access criteria for these, which exclude some of our more vulnerable members as they are currently proposed. Immunocompromised patients would qualify, but those who are not would need to meet restrictive criteria. We explained our community need and asked that PHARMAC apply criteria that allow for assessed vulnerability and consider multiple comorbidities. We are waiting to hear a decision on this.

For those who use disability funded Carer Support, there is increased flexibility to help families through these difficult times. If you are a primary full-time carer, you can use your Carer Support to pay for resident family members to help you achieve a break. If carers are unwell and struggling to provide usual support to a family member, you are urged to contact your NASC and say that you need emergency help. If you have any questions, please contact Carers NZ: centre@carers.net.nz

Education Advocacy - Highest Needs Review

Earlier this month, we joined a meeting with representatives from other rare disorder groups to speak with Kirsty Christison from the Highest Needs Review Team about some of the barriers our children face. Following on from this, we have made a submission to the review. The review team are also keen to hear from young people and whānau, and invite you to share your thoughts via an online submission form which closes **March 31st**.
The Highest Needs Review is taking place as part of a commitment outlined in Priority 4 in the Learning Support Action Plan 2019-2025: Flexible Supports for Neurodiverse Children and Young People. The review team will develop a report for the Education Minister with recommendations for Cabinet in October 2022. The Highest Needs Review is looking at how we access supports, level and duration of supports, range and flexibility of supports, whether supports meet individual needs, transitions across settings or schools, relationships between educational settings and whether these allow access to expertise, plus the cohesiveness of the agencies we use (education and disability supports).

Fair for Rare NZ Campaign / Pae Ora Health Reform

Thank you to everyone who supported Rare Disease Day, February 28th. The international focus was ‘GLOBAL HEALTH EQUITY’ which provided an opportunity to ask that the Pae Ora Bill recognises our significant rare disorder population group. On Rare Disease Day, we shared a news post featuring a video made for the APPWS conference by one of our members. If you haven't watched this yet, please take a look and share across your social networks. Rare Disorders NZ also released their White Paper presenting the results of the 'Voice of Rare Disorders survey 2021'.

We're calling for

Fair

FOR RARE

Are you with us?
www.raredisorders.org.nz

Despite pleas by many, the Minister of Health seems intent on pushing the Pae Ora Health Reform through Parliament as soon as possible without establishing a framework for rare disorders. It is very disappointing that those living with rare disorders may not be recognised as a population with distinct health needs in this landmark legislation. A reform which cannot be described as innovative or 'world leading' when it falls further behind many other countries without implementing a specific health strategy for rare disorders.

Health Minister Andrew Little feels that the reform will improve things for all people, including those with rare disorders. We are told that the Disability Health Strategy will sufficiently cater for the needs of the rare disorder population group, but there can be many differences in terms of medical expertise required, medicines access, and care, treatment and support needs.
The current Bill also fails to make any changes to the way that PHARMAC operates and there has been no indication as yet that this will be revised. The Independent Review of PHARMAC is complete, but the Government are yet to release it's findings. This report cannot lead to improvements in our medicines funding system if the legislation that

governs it remains unchanged. There is a concern that the report will be quietly released when it can be conveniently swept under the carpet, as we know that the interim report raised issues around equity, transparency, timeliness, as well as historic underfunding.

What can you do?

As the Pae Ora Bill is a once-in-a-generation health reform which outlines core changes planned for our health system, we need to ask that rare disorder groups are consulted during the development of health strategies. We need to continue campaigning for recognition and a specific Rare Disorders Health Strategy which addresses inequities, improves health outcomes and aligns with international best practice.

- Visit your local MP to raise these issues (before the draft Bill is handed to the Minister in April / May and is put through Parliament). Please contact Kim at RDNZ who can post you an information kit to handover.
- Check out the campaign resources on RDNZ's website - find useful templates, share, tag and post away! #FairforRareNZ
- Forward the White Paper by RDNZ which provides fresh evidence for why health inequities for the rare disorder community must be addressed.

Scoliosis Care in New Zealand

We are aware that some children with scoliosis are not being diagnosed until their curve is quite severe. This is concerning because an earlier diagnosis can lead to treatments that reduce the curve or halt progression.

Scoliosis can remain well hidden in PWS due to altered tone and differences in how scoliosis presents in PWS compared to idiopathic scoliosis. This makes it hard to detect visually and curves can appear less significant. Screening practice in New Zealand is to use the Adam's Forward Bend Test and then to refer for x-ray if there are concerns. However, this can result in cases being missed and does not align with the recommendation of Dr Harold van Bosse, who is highly regarded internationally as a leading expert.

As studies have shown that almost a 1/4 of children with PWS will develop a curve before their 4th birthday, Dr. van Bosse recommends radiographic screening once a child is sitting unassisted, and then annually until about 4 years old. If a curve is detected at a young age, casting can be very successful. After 4 years, the risk of scoliosis developing then diminishes until increasing again in adolescence. We intend to call for changes to current screening practice, but in the meantime, we suggest you request your paediatric team follow Dr. van Bosse's recommendation if your child is under 4 years. After this time, monitoring with thorough spine exams is sufficient, but with close observation from 10 years old. Look carefully for any signs of asymmetry and request an x-ray if you have concerns. (Note: if your child did not receive spinal x-rays at a young age, consider that a curve could have been missed.) Several families have told us that their doctors blame visual appearance on poor posture or are reluctant to order an x-ray because they feel radiation risks outweigh scoliosis risk. However, risk of scoliosis in PWS has been found to be very high, with a prevalence rate of 60-70%. The prevalence rate also appears slightly higher in females and those with UPD.

On another note, there are often questions about whether growth hormone treatment should be paused when a curve is found. Expert advice is based on two studies (2021 and 2009) which found neither prevalence or progression to be affected by GHT. It is more likely that GHT is helpful as it improves lean muscle mass and bone density. Recent data has indicated that GHT helps to ward off scoliosis in infants.

For further information about scoliosis, expert presentations can be found in the orthopaedics playlist on our YouTube channel. Guidelines and research papers for your medical team can be found in the professional resources section of our website. Please ask if you can't find what you are looking for and we would be happy to help.

Listening to Voices of people living with PWS around the World

This is a reminder about the international study we are participating in by Kamila Lipiec and Kate Woodcock at the University of Birmingham, UK. This is the first study surveying people with PWS in order to capture their voice. It aims to understand how people with PWS feel they fit into society and what is important to them.

We hope to have strong participation from New Zealand as the results will provide us with greater insight and will help to inform how we support people with PWS moving forward.

Individuals with PWS aged 16 years or older have been invited to participate by completing an online survey, asking for short written answers that takes no more than about 45 minutes. The survey has 'easy read' support and caregivers can support with writing down answers, as long as the answers come from the person living with PWS. Please contact us if you would like us to resend details to you.

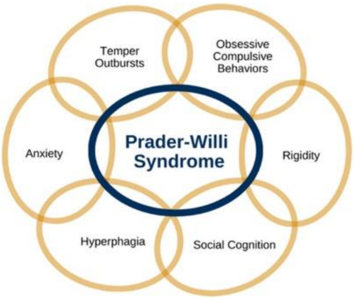
Board Member Vacancy

We are always keen to hear from members who have an interest in becoming more involved in the work of our organisation and if becoming a Board Member is something you might be considering, please get in touch with CEO Charlotte Roos to ask any questions and discuss further: ceo@pws.org.nz. We are very pleased to say that we have received interest in the Board Member / Treasurer role.

Zoom Chat Time (Tweens and Teens)

If you are keen to participate in Zoom Chat for Tweens and Teens, this is just a reminder to please complete the survey that was sent out to find the most suitable time for all, if you haven't already done so. Survey link: www.surveymonkey.com/r/JXWBTZP
Zoom Chat for Tweens & Teens will still be held on the 2nd weekend of the month.

PWS Research News Overview



This is just a brief overview with snippets of information about some of the latest research news. To read more, please follow the links to further information found on our Research News website page.
For information about different types of treatments being investigated, see our Potential Future Treatments page.

Carbetocin / LV101 Update (intranasal oxytocin-like treatment)

A New Drug Application for LV-101 was granted FDA Priority Review last year for the treatment of hyperphagia and behavioural symptoms in PWS, but ultimately, the review was unsuccessful. Following positive results observed on hyperphagia, anxiety and distress symptoms, the negative vote by the FDA advisory committee surprised many in the PWS community. In January 2022, the FDA explained that the efficacy data available for the proposed 3.2 mg dose was insufficient and they recommended an additional study be conducted to confirm results. Levo Therapeutics is currently in discussions with the FDA regarding the design of this new study. Read more >

DCCR Update

Last year we reported that results of the phase 3 trial (DESTINY PWS) were negatively skewed by the impact of Covid19, so Soleno submitted additional analyses to the FDA showing differences in pre-pandemic data. The FDA requested an additional trial to support a New Drug Application, but an ongoing extension study, C602, was already underway. Soleno, PWSA(USA) and FPWR petitioned and questioned the need for another trial, supported by the collection of compiled experiences of patients and caregivers during the completed DESTINY PWS trial and the C602 open-label study. The FDA did not rule out the need for an additional trial, but agreed to evaluate supplementary data and to allow the inclusion of patient voice in discussions. In January 2022, the FDA indicated they were receptive to a study design involving participants currently enrolled in C602 to generate additional control data required to support an NDA. Soleno is submitting a study proposal and if acceptable, this study will be initiated soon. Read more >

New area of the Brain Identified for Theapeutic Targets

The hypothalamus and hindbrain have been implicated in appetite regulation, but drugs targeting these areas have not been as successful as hoped. Drs. Betley and Chen speculated that the cerebellum could play a role in hunger suppression and re-examined MRI scans, looking for differences in how the brains of PWS patients responded to food compared to a control group. They identified a subset of glutamatergic neurons in the anterior deep cerebellar nuclei (aDCN) that were associated with promoting satiety. They found that activating these aDCN neurons in mice led them to dramatically curb their meal size and that this process increased striatal dopamine levels. Dopamine reward pathways are known to influence eating behaviour. Read more >

Consensus Paper on Behavioural Features of PWS

The International PWS Clinical Trials Consortium have produced consensus definitions and descriptions of the key behavioural features of PWS, with the aim of providing standardised measures for treatment studies. Follow the link to view a useful infographic to print summarising the 6 definitions. Read more >

Pharmacodynamic Gene Testing

Side effects of psychotropic medications are often atypical in PWS compared to the general population, so starting low and slow is recommended and introducing only one new treatment at a time. Potential for the worsening of symptoms can result in patients spending years trying to find the right balance of treatments, but pharmacogenomics can now be used to interpret how genes affect a person’s response to drugs and find appropriate treatments without trialing lots of medications first. Drs. Janice Forster, Jessica Duis and Merlin Butler explored gene alterations in PWS and response to psychotropic medications. Trends were observed that indicated differences between the genetic subtypes of PWS. Amongst other key findings, they found greater potential for clinical response to lower doses of SSRIs, and mood activation at typical doses. Read more >

Accelerated Ageing in Adults with PWS

It has been observed that physical appearance can change earlier in PWS, and that problems associated with older age in the general population can also emerge earlier. Two new studies investigated brain-age and evidence for accelerated biological ageing in PWS. They both concluded that premature ageing occurs in PWS. Read more >

International PWS Conference

If you are planning some international travel with our border reopening soon, you might want to consider Ireland in your travel itinerary!
Registration for the 11th International PWS Conference is open now with Early bird registration closing on April 7th.
Visit the IPWSO conference webpage for further details and registration.
WHEN: 6th – 10th July, 2022
WHERE: University of Limerick

Notices / Events

Dates for your Diary

Virtual Social Evening for Parents of 0 - 5 Years - Sun 27th March, 7.30pm
Virtual Social Evening for Parents of Teens/Adults - Sun 24th April, 7.30pm
Zoom Chat for Adults with PWS - Saturday 2nd April, 4pm
Zoom Chat for Tweens and Teens - Saturday 9th or Sunday 10th April, TBC
Transition Expo 2022, Auckland - Wednesday 7th September 2022
PWSA(NZ) Young Families Weekend - Sat 17th - Sun 18th Sept 2022, Wellington
PWSA(NZ) Family Camp - 10th-12th March 2023

Annual Transition Expo 2022

2nd PPT date: Weds Sept 7th, 9am-2pm - Auckland Netball Centre, St Johns
by Disability Connect, supported by MSD and Geneva Healthcare
Opportunities and advice from Transition, Vocational, Recreation, Activity and Information Providers. For all students with a disability and those involved in the disability sector.
Click here for more information >

Many regional events and workshops are being presented online at the moment. The following websites can be checked for future dates.

Imagine Better Workshops - i.e. Making Individualised Funding Work, and other workshops for families, service providers or people with disabilities

Care Matters Workshops - i.e. Explore-Learn-Connect, Transitions

Parent to Parent Courses and Workshops - i.e. Navigating the System, Renew-Connect-Reflect

Disability Connect Workshops (Auckland) - i.e. Education Legal Issues, IF, Transition, Planning for Adulthood, Living Options, WINZ, *($20 per family)*

Parent to Parent branches for local support group meetings

Sibling Support Programmes - SibShops and Camps are advertised on the Care Matters and Parent to Parent calendars.