PRADER-WILLI SYNDROME ASSOCIATION NEW ZEALAND
ADVOCACY · EDUCATION · SUPPORT

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The healthcare of paediatric patients with PWS is usually overseen by an endocrinologist, or sometimes a paediatrician, depending on the region and clinics available. We are therefore writing to both endocrine and paediatric groups, as well as NZOA regarding our request for a review of scoliosis screening protocol for PWS, plus the development of consensus guidelines for the prescribing of growth hormone therapy during scoliosis treatment.

Request for Review of Scoliosis Screening for Patients with Prader-Willi Syndrome

The New Zealand PWS Association has recently been contacted by one of our members whose child was found to have a spinal curve of up to 50 degrees at age 7. Scoliosis had not been identified prior to this, so the severity of the curve was understandably quite a shock to the parents. We are also aware of other cases where scoliosis has been diagnosed later in childhood. The prevalence rate of scoliosis in Prader-Willi syndrome has been found to be very high [Crinò A, Armando M, Crostelli M, et al. 2022] and because it is known that earlier treatment can lessen the curve or halt progression resulting in a lower likelihood of future surgery, we believe there is a case for an early radiographic screening programme. We understand there may be a concern regarding the risks of radiation if screening x-rays were to be introduced, but we believe this is outweighed by scoliosis risk, considering new information about prevalence.

It is suggested that scoliosis can remain well hidden in PWS due to hypotonia and differences in how scoliosis can present in patients with PWS. Therefore, scoliosis may remain visually undetected at clinic appointments and curves may appear to be less significant. Current clinical practice to use the Adam's Forward Bend Test and refer for x-ray if there are concerns appears to result in cases being missed.

An early screening and treatment programme has the potential to prevent some future surgeries for which there is additional risk of complications due to the comorbidities associated with PWS [2, 3]. Furthermore, major surgery can be very disruptive and more

challenging to the lives of patients with PWS and their families due to the existing demands and difficulties of managing the syndrome.

PWSA NZ has looked to the recommendations of Dr Harold van Bosse, an orthopaedic surgeon who speaks internationally at PWS conferences on this subject and is highly regarded as a leading expert. We ask that his advice be considered in the development of scoliosis screening policy for PWS in New Zealand. Dr van Bosse believes data shows that almost a quarter of children with PWS will develop a spinal curve before their 4th birthday and he therefore recommends radiographic screening once a child is sitting unassisted, and then annually until around 4 years old. He states that spine casting can be very successful if a curve is detected at a young age. Dr van Bosse also recommends close observation from 10 years old when the risk of developing scoliosis increases again.

If similar screening practice were to be adopted in New Zealand, it would also be important to allow for screening checks on children currently older than 4 years who have not been previously screened for scoliosis.

We attach a copy of a recent paper by Dr van Bosse and Prof Merlin Butler:

> van Bosse HJP, Butler MG (2020) Clinical Observations and Treatment Approaches for Scoliosis in Prader–Willi Syndrome. [4]

Dr van Bosse has also kindly indicated to us that he is available to answer any questions around screening, or on his experience of scoliosis treatment in PWS.

On a different point, the family who recently contacted us had to wait for an MRI scan before they could see the specialist team that their child was being referred to. We do not have any information around whether this is usual practice for referrals in New Zealand, but when discussing this case with Dr van Bosse, he mentioned that he does not order MRIs on his patients with PWS unless, a.) a patient is having significant spine surgery, b.) a patient's curve is progressing extremely fast or otherwise acting unexpectedly, or c.) a patient is having unexplained symptoms such as headaches, backpain, leg weakness or numbness. He explained that normal orthopaedic practice is to order an MRI to rule out spinal cord abnormalities in curves that occur before 10 years old, but because 23% of children with PWS will develop curves before that age due to their PWS diagnosis alone (and hypotonia), an MRI and anaesthesia is usually unnecessary. He also mentioned that he does not recall any of his patients with PWS having an abnormal MRI.
Therefore, we question what the usual practice is in New Zealand for PWS patients and whether MRI referrals could be delaying treatment unnecessarily?

Growth Hormone Therapy and Scoliosis

We would also like to request the development of national consensus guidelines on the continuation of growth hormone therapy during scoliosis treatment in PWS. Our members are sometimes receiving differing advice on whether to pause GHT following a diagnosis of scoliosis and we would therefore like to ensure that national practice is standardised and reflects the latest research findings.

Dr van Bosse summarises the argument for continuing GH treatment following a scoliosis diagnosis in the paper attached [4]. It is a common concern that GHT may contribute to or worsen a curve's progression, however, GHT has been found to have no effect on the development of scoliosis in PWS [5, 6, 7] and it is believed to be more likely that it is helpful due to improving lean muscle mass. In the systematic review of research by 43 international experts and stakeholders in the Growth Hormone Research Society Workshop in 2013, consensus guidelines were produced which included the following recommendation in Table 1. VI. "Scoliosis should not be considered a contraindication to rhGH treatment in patients with PWS. (Recommendation level A; level of evidence 2)" [8]

In his own practice, Dr van Bosse only stops GH treatment in patients who are having surgery on the day of their surgery, and restarts the following day, unless otherwise directed by a patient's endocrinologist.

We would be very grateful if these issues could be discussed and shared via the relevant professional network communications. Thank you for your time in reading and considering our letter.

Yours sincerely,

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References

1. Crinò, A., Armando, M., Crostelli, M., Mazza, O., Bruzzese, D., Convertino, A., Fintini, D., Bocchini, S., Ciccone, S., Sartorio, A., & Grugni, G. (2022).
High Prevalence of Scoliosis in a Large Cohort of Patients with Prader-Willi Syndrome.
J Clin Med. 2022;11(6):1574. Published 2022 Mar 13. doi: 10.3390/jcm11061574

2. McQuivey, K. S., Chung, A. S., Jones, M. R., Makovicka, J. L., Christopher, Z. K., Brinkman, J. C., & Belthur, M. (2022).
Hospital outcomes in pediatric patients with Prader-Willi syndrome (PWS) undergoing orthopedic surgery: A 12-year analysis of national trends in surgical management and inpatient hospital outcomes.
Orthop Sci. 2022;27(6):1304-1308. doi: 10.1016/j.jos.2021.08.005

3. McQuivey, K. S., Sheridan, J. R., Chung, A., Mayfield, C., Gulbrandsen, M., Brinkman, J. C., & Belthur, M. V. (2021).
Hospital outcomes of scoliosis surgery in children with Prader-Willi Syndrome: comparison with adolescent idiopathic scoliosis.
Spine Deform. 2021;9(6):1641-1647. doi: 10.1007/s43390-021-00359-7 [PubMed]

4. van Bosse HJP, Butler MG (2020).
Clinical Observations and Treatment Approaches for Scoliosis in Prader-Willi Syndrome.
Genes (Basel). 2020;11(3):260. Published 2020 Feb 28. doi: 10.3390/genes11030260

5. de Lind van Wijngaarden R.F., de Klerk L.W., Festen D.A., Duivenvoorden H.J., Otten B.J., Hokken-Koelega A.C. (2009)
Randomized controlled trial to investigate the effects of growth hormone treatment on scoliosis in children with Prader-Willi syndrome.
J. Clin. Endocrinol. Metab. 2009;94(4):1274–1280. doi: 10.1210/jc.2008-1844

6. Nakamura Y, Murakami N, Iida T, Asano S, Ozeki S, Nagai T. (2014)
Growth hormone treatment for osteoporosis in patients with scoliosis of Prader-Willi syndrome.
J Orthop Sci. 2014;19(6):877-882. doi: 10.1007/s00776-014-0641-0

7. Grootjen, L. N., Rutges, J. P. H. J., Damen, L., Donze, S. H., Juriaans, A. F., Kerkhof, G. F., & Hokken-Koelega, A. C. S. (2021)
Effects of 8 years of growth hormone treatment on scoliosis in children with Prader-Willi syndrome.
Eur J Endocrinol. 2021;185(1):47-55. doi: 10.1530/EJE-21-0211 [PubMed]

8. Deal, C. L., Tony, M., Höybye, C., Allen, D. B., Tauber, M., Christiansen, J. S., & the 2011 Growth Hormone in Prader-Willi Syndrome Clinical Care Guidelines Workshop Participants. (2013)
Growth Hormone Research Society workshop summary: consensus guidelines for recombinant human growth hormone therapy in Prader-Willi syndrome.
J Clin Endocrinol Metab. 2013;98(6):E1072-E1087. doi: 10.1210/jc.2012-3888