

Preventing unexpected death in Prader-Willi syndrome:

Recognising risk, responding early and learning from families.

Jacob's Story

by Jo Davies

PWS PRADER-WILLI SYNDROME ASSOCIATION NEW ZEALAND

IPWSO International Prader-Willi Syndrome Organisation

Previously healthy

Jacob was born in February 2003 and diagnosed with PWS by deletion. He started GHT around 2.5 years and maintained a healthy weight most of his life.
He died in May 2023 at just 20 years old.





LEFT: age 9
ABOVE: 16th birthday
RIGHT: age 17
FAR RIGHT: 18th birthday





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Background

Slight weight gain after leaving school and becoming more independent, but this was managed. Mental health needs remained unaddressed – a contributing factor to the situation Jacob was in when he died.



- Lack of support during transition period.
- Intermittent, atypical mental health symptoms.
- Sertraline
- Discharged from mental health services.
- Sudden escalation in challenging behaviour.
- Disturbed sleep
- Police involvement

➢ Compulsory placement in an inexperienced and unprepared home.

Jan 2022
2 months before his 19th birthday.
Approx. 4 months before having to leave home.

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6 months later, ambulance called…

Jacob had been living at the compulsory care placement home for approx. 6 months.
He had gained 20kg, was diagnosed with prediabetes, had developed undiagnosed diabetes.

- Health advisor evening prior – prn laxative authorised.
- Unwell the next day: did not join BBQ outing – no lunch, remained in bed. Called out for help late afternoon. Dizzy, unable to stand, very thirsty, unable to swallow solid food – given soft weetbix. 6pm declined dinner.
- Noted at 8pm: hot, red and 'rash-like' skin, a 'really veiny' stomach, slurred speech, dry lips, dark circles around eyes, urinary incontinence. Ambulance called 8.48pm.
- Welfare guardians (parents) informed 10.56pm.
- Ambulance report:

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Pt felt unwell with abdominal pain 1/7 days ago
Pt states ongoing dizziness last 2/7
Pt states nausea and vomiting 1/7 days ago, none today
Fast radial pulse, Febrile, Hyperglycaemic
Lower abdomen pain, Firm abdomen not normal for pt
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LEFT: Jan 2023
ABOVE: Feb 2023, 20th birthday

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ED presentation and 2 days in ICU



- Severe diabetic ketoacidosis (DKA) – glucose 70.9mmol/L, ketones 5.6, AG 39
- Severe oliguric acute kidney injury (anuric in ED) & hypernatremia – Na 176
- Persistently febrile, high lactate - 4.0 (low O^2 to organs). Suspected sepsis.
- Marked vasopressor support required (to raise low blood pressure)
- CT chest and abdomen - small amount of free fluid within abdomen, possible pneumonitis in lungs.
- Standard DKA protocols followed using IV therapies including insulin.
- Broad spectrum antibiotic (tazocin), blood cultures
- Improving as DKA resolving. Then increasingly unstable with severe hyperthermia, increased circulatory support requirements.
- Chest x-ray – gas within biliary tree (bile duct system)
- Laparotomy surgery to rule out bowel ischaemia (blockage / restricted blood flow) - mildly 'unwell' looking bowel, but no lesion found.
- Multi-organ failure

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Official cause of death (post-mortem report)

1a. Diabetic ketoacidosis complicating intra-abdominal sepsis

2. Prader-Willi syndrome

Pathologist findings:

- Liver & kidney findings consistent with organ failure.
- Abscess found in the upper abdomen, in the area behind the stomach, situated in the small gap between the lesser sac, pancreas and duodenum. (5 x 2 x 0.3cm)
- Cause of abscess suspected to be a perforation of the small intestine by ingested object, subsequently passed through. Supported by inflammation extending through intestinal wall, likely representing the perforation tract.
- Bacteria detected shown to be resistant to multiple antibiotics, including tazocin. Susceptible to others.

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Early signs missed & overlooked vulnerabilities

Hospital admission - Saturday 27th May. During the previous week, the following events were logged in support records but not followed up, and signs of being unwell were missed.

- **FRI 19TH MAY**: Jacob's weekly shopping budget spent on an Indian take-away (onion bhaji, lamb curry, naan bread, mango lassi.) Afterwards, he *"took himself into the kitchen and drank all the milk."* Had he eaten something that got stuck? Did he consume other food in the kitchen? Excessive thirst?
- **SAT 20TH MAY**: Jacob opted out of BBQ outing, *"spent most of the day in his bedroom, staff have noticed Jacob making frequent visit to the toilet today, has hardly eaten. staff have asked Jacob if he is fine and replies he is ok."*
- **SUN 21ST MAY**: We visited during afternoon; took Jacob out. Unaware of the above. On return, Jacob skipped dinner, but it was assumed he ate with us, although we had never done that on afternoon visits.
- **MON 22ND MAY**: Jacob declined to attend the gym (usually enjoyed), *"has been in his room majority of the day."*
- **TUES 23RD MAY**: Another day spent mainly in his room *"for a rest."*
- **WEDS 24TH MAY**: Jacob declined to attend the gym again, *"due to be tired."*
  Notes recorded that he visited bathroom x3 during night.

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Early signs missed & communication breakdowns

- **THURS 25TH MAY**: Inconsistency between support note and verbal information provided. Note says Jacob *"exercised for a good hour"* at the pool, contradicting Friday phone call.
- **FRI 26TH MAY**: Phone call – line not working. Jacob reported difficulty swimming, trouble swallowing. Asked if he had a sore throat, replied 'not really'. Asked if he needed more swimming lessons, replied 'maybe'. Support note: *"Jacob in a calm and settled mood.... JD spoke to his mother this evening was happy about that."*
- Call to house mobile - key worker confirmed difficulties above. Gave impression something not right. I provided what I thought were reminders that illness can present atypically in PWS – dysregulated temperature, high pain threshold, unlikely to vomit or be 'off' food. Requested they monitor Jacob, keep me posted, but info not passed on. Also informed KW that 2 discontinued meds still on script (iron, cetirizine).
- KW called their health advisor around 8pm. Discussed 'refusal' to take medication. Incident report filed. KW also reported to HA that Jacob thought he had constipation. HA record: *"Jacob states has not passed a BM for a few days. No bowel charts completed for him. Has not had a dose recently. Authorised prn laxsol."*
- KW sent text advising Jacob settled, and HA notified about meds changes. No mention of constipation & prn.

SAT 27TH MAY: House mobile unanswered all 3 times I called to check on Jacob and arrange a visit.

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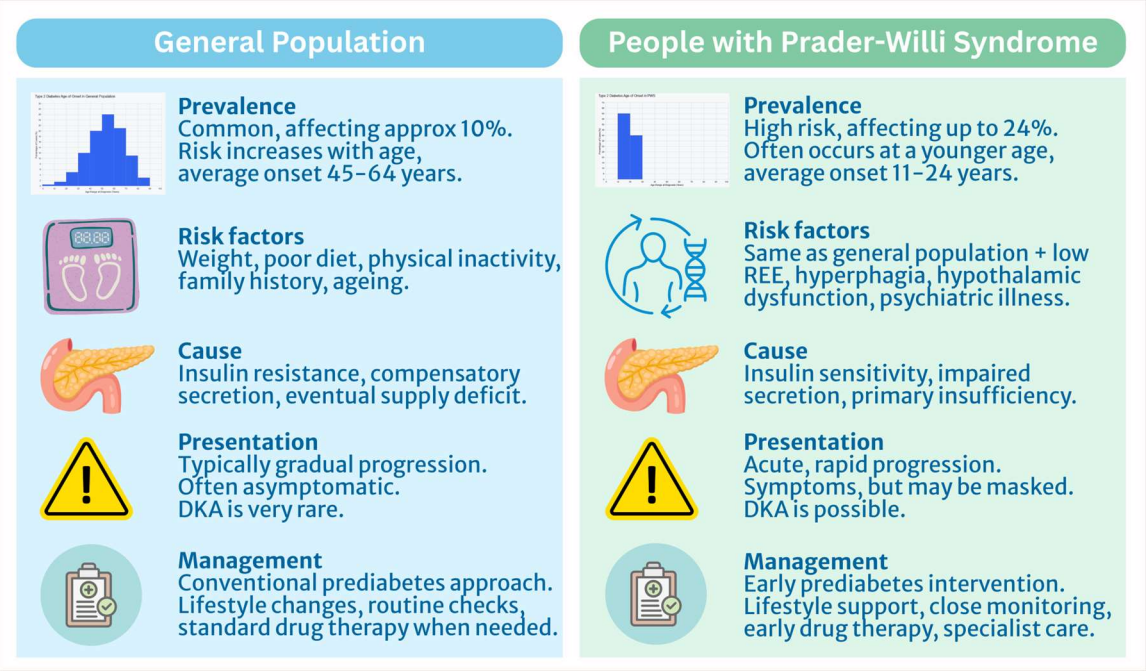
Underestimated diabetes risk

Requested a GP referral to dietetics to support the residential service. They ordered bloods and an in-person assessment (March 21st). Prediabetes was diagnosed; lifestyle changes were advised with 3-month repeat bloods scheduled. I believe this response was inadequate:



- Rapid weight gain – 16kg in 12 weeks
- Rapid increase in HbA1c (Dec 36 → March 48)
- Growth hormone therapy
- Antipsychotic (aripiprazole)
- No indication residential provider would be able to implement support for ‘lifestyle changes’
- Limited opportunity for exercise
- Typical age of onset
- PWS, reversible?

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The value of condition-specific specialist advice

ENGAGING PARENTS AS PARTNERS, RESPECTING ACCUMULATED KNOWLEDGE, LIVED EXPERTISE, AND RIGHTS TO BE INFORMED

- Ensuring timely notification and consultation with parents / guardians
- Preserving and protecting effective family advocacy
- Learning from previous incidents, i.e. inappropriate response to alcohol poisoning → GCS 9

EQUIPPING FRONTLINE HEALTH ADVISORS WITH SPECIALISED RESOURCES AND TRAINING

- Check credibility of information source
- Who trains the health advisors?
- Expanding the clinical care team

CONNECTING MEDICAL PRACTITIONERS WITH SEAMLESS ACCESS TO SPECIALIST EXPERTISE

- Improved clinical systems and digital tools
- Centres of expertise, disorder-specific care and screening guidelines

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Recognising risk in the residential care setting

SYSTEMIC AND CARE PLANNING RISKS

- Inadequate risk management and care planning: failure to incorporate PWS clinical complexity. Failure to use relevant information sources, check, and update information.
- Suitability of placement.
- Ineffective transition to service / lack of preparation.
- Absence of a guardianship policy.



STAFFING AND OPERATIONAL RISKS

- Deficits in team experience, awareness and training.
- Use of casual / agency staff, gaps in shift handovers.
- Superficial record keeping, lack of oversight – failing to 'connect the dots'.
- Poor accessibility and low visibility of critical information / alerts.
- Inconsistent food security, supervision, monitoring – failure to understand rapid weight gain & hyperphagia risks.
- Poor communication channels with family and the person with PWS. Restriction of communication tools.
- Delayed examination and sharing of support logs / records when things go wrong.

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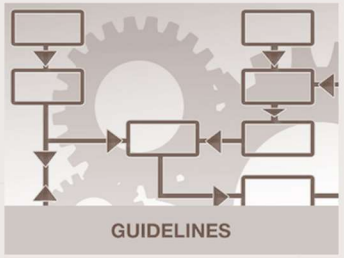
My recommendations:



1) Pharmacogenomic testing – did aripiprazole worsen weight gain and hyperglycaemia?



2) Adoption of standard clinical care guidelines / care pathways for PWS.

3) Linking PWS care guidelines to patient health records, which can activate 'red flag alerts'.



4) Mandatory PWS training for residential staff teams BEFORE supporting people living with PWS. (At least one trained person on shift at all times.)



5) Robust operational procedures that ensure illness in PWS is recognised early and promptly evaluated – with visible health alert reminders, and clear tracking systems to analyse support logs and catch trends.

6) A multidisciplinary care planning and support team, which includes the involvement of experienced health professionals and parents / guardians.

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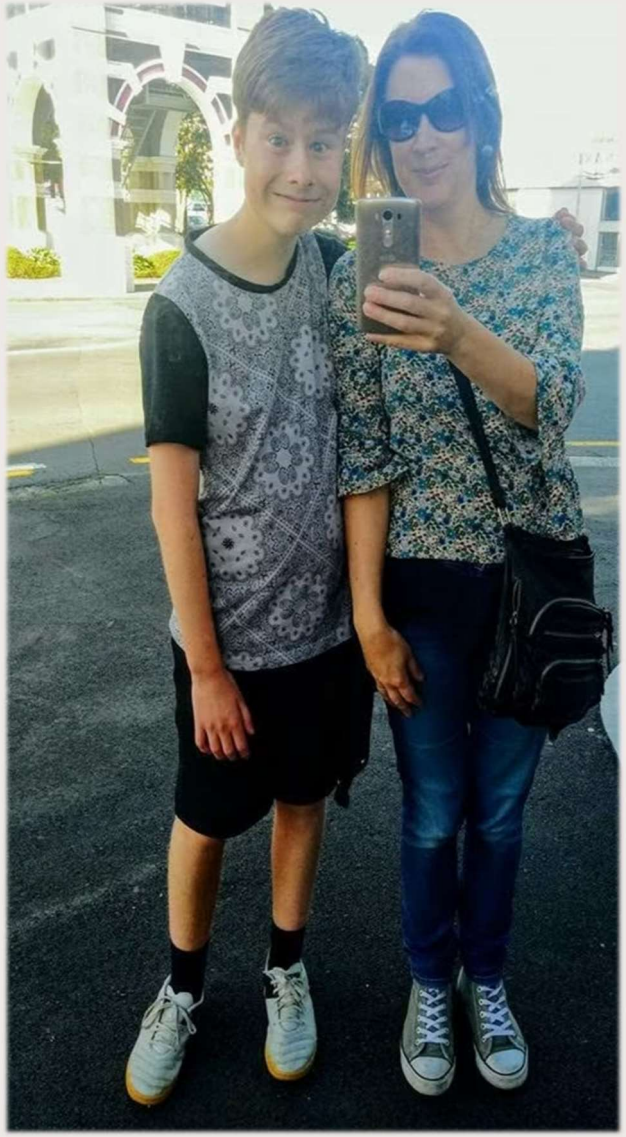

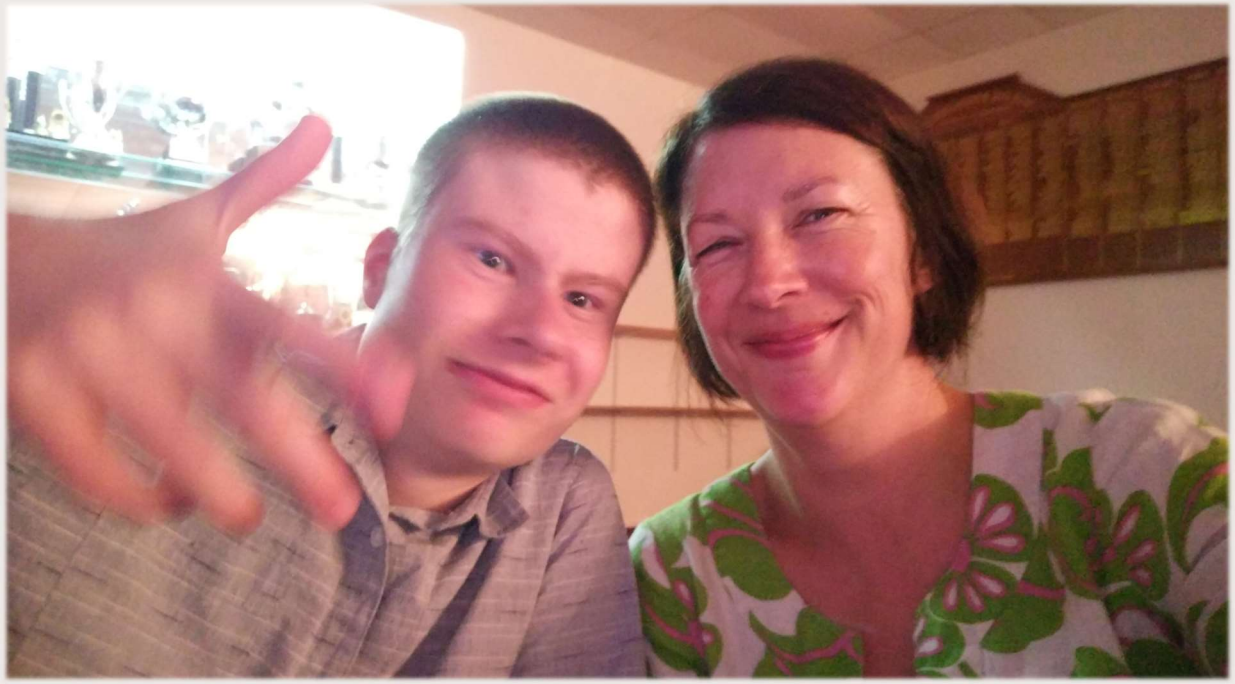

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