

Addressing the challenges of diagnosis for a rare disease: How IPWSO is supporting diagnosis for people with Prader-Willi syndrome

1. What is Prader-Willi syndrome (PWS)?

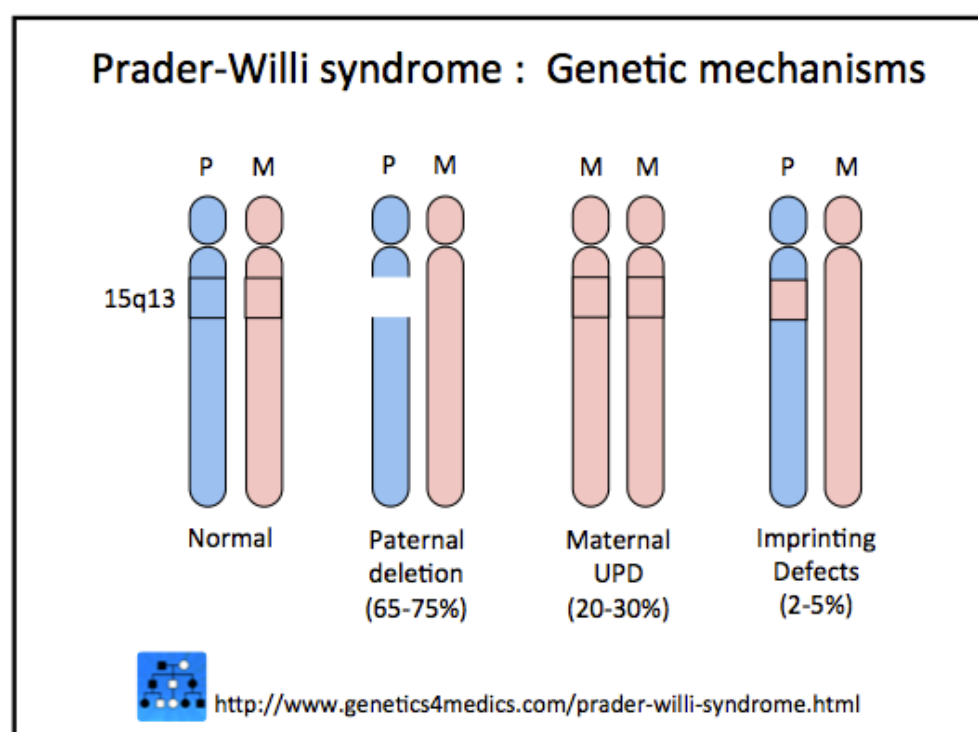
Prader-Willi syndrome is a complex and rare neurodevelopmental condition affecting between 1 in 15,000 to 25,000 births. PWS affects all races and both sexes equally.

What causes PWS?

A lack of expression of one or more genes in a specific region of chromosome 15.

What are the symptoms of PWS?

- Low muscle tone and failure to thrive at birth,
- later evidence of short stature,
- intellectual disabilities,
- behavioural and psychiatric challenges, and
- the development of a chronic feeling of hunger that can result in over-eating and in severe obesity and associated health problems.



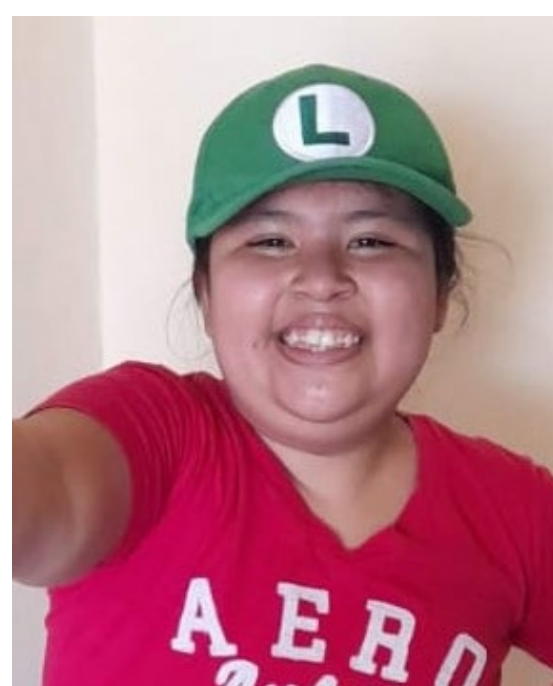
2. When and how are people diagnosed?

Diagnosis is ideally made following the observation of various symptoms in a young baby, in particular:

- severe hypotonia, and
- feeding difficulties.

A genetic test will confirm the diagnosis.

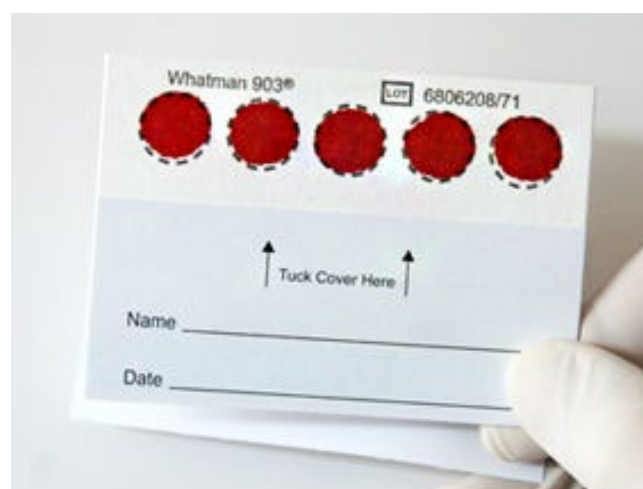
However, we hear of people being diagnosed in their teens, if not later, most recently a 17-year-old in Peru. We are also concerned that many people do not receive a diagnosis at all. There may also be high infant mortality amongst babies with PWS in parts of the world. **We are aware that diagnosis can be prohibitively expensive and/or simply not available for many families.**



3. What is IPWSO doing?

In the early 2000s IPWSO made contact with a laboratory in Italy to discuss establishing **a global base for PWS diagnostic testing**. Since 2003 IPWSO has funded the testing through the Baschirotto Institute for Rare Diseases (BIRD) of over 500 samples received from 49 countries.

The system is simple. The referring physician uses a widely available sampling method (dried blood spot on filter paper) and mails the sample to Italy. IPWSO covers the cost of analysing the test.



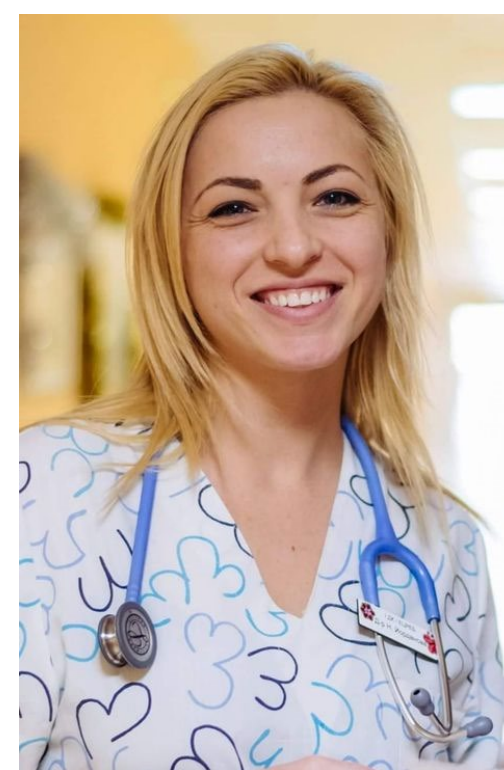
We have also provided training, advice and travel grants to clinicians interested in establishing diagnostic testing for PWS in their own countries.

4. Case study: Bulgaria

First Pediatric Clinic, University Hospital "Sveta Marina" in Varna, Bulgaria



"My name is Nikolinka Yordanova. I am a pediatric endocrinologist and core specialist of our PWS Multidisciplinary team. The head of our Department, Prof. Violeta Iotova has collaborated with the BIRD Foundation since the early 2000s. There are currently 20 patients (16 children and 4 adults) in our center, half of them are confirmed through testing at BIRD, which is funded by IPWSO. In Bulgaria MLPA (genetic testing) was introduced about 10 years ago, in Varna it has been available since the beginning of 2020. There is no reimbursement for this expensive genetic test and most of the families are not able to pay for it.



"Sending a sample of dried blood spot by post is a very simple and cost-effective way to confirm a diagnosis and to give the patients the chance to then start on other therapies such as Growth Hormone as early as possible. Our patients and staff are very thankful to BIRD and IPWSO for the excellent collaboration and timely diagnoses and we hope that we will continue to work together in the future!"

5. Why is diagnosis important?

During 2020, 20 tests were carried out on behalf of IPWSO by BIRD. The samples came from children ranging in age from infancy to 14 years.

Diagnosis is key to starting families off on the best path to access care, information, treatment and support. Supporting early diagnosis is vital in **promoting global equality, enabling patients to find a simple route to diagnosis and working together with professionals to share expertise.**

IPWSO funded tests, 2020

Country	Number
Bulgaria	8
Peru	5
Myanmar	2
Georgia	2
Kosovo	2
India	1

6. About IPWSO

We are an international, non-profit, parent-led, membership organisation supporting national Prader-Willi syndrome associations, as well as people with PWS, their families and the professionals who work with them. We have a membership of 40 national PWS associations and contacts in over 100 countries.

Find out more about our work at www.ipwso.org or follow us on social media.