"MAURO BASCHIROTTO"
INSTITUTE FOR RARE DISEASES

International Prader-Willi Syndrome Diagnostic Testing Initiative

IPWSO International Prader-Willi Syndrome Organisation

Clinical data collection form

Patient:		
Last name: Last name		*First name:* First name
Date of birth: DD/MM/YYYY	*Sex:* ☐Male ☐Female	*Country:* Country

Referring medical doctor (the results will be sent to this address:		
Last name: Last name		*First name:* First name
Institution: Institution		
Address: Address		
Zip/Post code: Zip/Post code	*City:* City	*Country:* Country
Tel: Tel.		*E-mail:* E-mail

Clinical data:				
Current age: _____Years	*Current weight:* _____ kg	*Current weight percentile:* _____	*Current height:* _____ cm	*Current height percentile:* _____

☐ Hypotonia at birth with poor suck	Days/weeks of tube feeding _____
☐ Hyperphagia (excessive interest in food and eating)	Age when noted ____ Self control of food intake ☐Yes ☐No Age when weight gain became a problem ____
☐ Hypogonadism	Males: History or presence of cryptorchism ☐Yes ☐No Small genitals for age ☐Yes ☐No Delayed puberty ☐Yes ☐No (Note: early adrenarche is prepubertal) Females: Small labia as newborn ☐Yes ☐No Age at menarche ____ Menstruation monthly ☐Yes ☐No
☐ Global developmental delay	Brief description
☐ Cognitive impairment	Brief description
☐ Behavioural problems (tantrums, controlling)	Brief description
☐ Other symptoms	Brief description

Previous testing:	
☐ FISH analysis of the 15q11.2 region	Results
☐ DNA methylation assessment (MS-MLPA or MS-PCR)	Results
☐ Other tests (e.g. chromosomal microarray)	Results

Major findings in Prader-Willi syndrome:
Profound neonatal hypotonia, weak suck, weak cry, first weeks. Tube feeding needed
Hyperphagia starting at age 1-6 years, causing weight gain and obesity if food not controlled by others
Small genitals, pubertal delay
Short stature for family and no pubertal height spurt if not treated with growth hormone and/or sex steroids
Global developmental delay

Other frequent findings:
Decreased foetal movement
Infantile lethargy
Hip dysplasia
Behavioural problems after infancy
Skin picking
Sleep apnoea
Small hands and feet (if not treated with growth hormone)
Scoliosis
Thick viscous saliva
Esotropia, myopia
Speech articulation defects
Concrete thinking, strong visual memory

Age at assessment	**Features Sufficient to Prompt DNA Testing**
Birth to 2 years	Hypotonia **that gradually improves**, poor suck and appetite **the first weeks to months**
2-6 years	**Delayed motor milestones**, hypotonia decreasing, a history of poor suck
	Global developmental delay
6-12 years	Delayed motor milestones, a history of poor suck first weeks/months
	Global developmental delay
	Excessive eating (hyperphagia; obsession with food) with central obesity if uncontrolled
>13 years	Delayed muscle milestones, a history of hypotonia and poor suck first year
	Cognitive impairment, usually mild to moderate intellectual disability
	Excessive drive to eat (hyperphagia; obsession with food) with central obesity if uncontrolled
	Hypogonadism
	Behaviour problems (including temper tantrums and obsessive compulsive features)

"Table modified from Gunay-Aygun et al., 2001."

Please note that the features described in this table are not the only ones present at the various ages but in their absence the diagnosis of Prader-Willi syndrome is highly unlikely. The inclusion criteria are more relaxed for younger patients to ensure that all PWS cases are tested. As an example, hypothalamic hypogonadism is observed as genital hypoplasia at all ages in PWS but in younger individuals it is not included as a required criteria.

References:
Driscoll DJ, Miller JL, Cassidy SB. Prader-Willi Syndrome. [Updated 2024 Dec 5]. In: Adam MP, Mirzaa GM, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2025. Available from: https://www.ncbi.nlm.nih.gov/books/NBK1330/

Gunay-Aygun M, Schwartz S, Heeger S, O'Riordan MA, Cassidy SB. Pediatrics. 2001 Nov;108(5):E92. *The changing purpose of Prader-Willi syndrome clinical diagnostic criteria and proposed revised criteria.* doi: 10.1542/peds.108.5.e92. PMID: 11694676.