*Rev. 4.0.6 03.03.2023*

# International Prader-Willi Syndrome Diagnostic Testing Initiative

**Clinical data collection form**

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

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| --- | --- | --- |
| **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 |

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| **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 | | | |

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| **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

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| --- | --- |
| 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 2023 Mar 9]. In: Adam MP, Mirzaa GM, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2023. Available from:

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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.