**Picture2**

**International Prader-Willi Syndrome**

**Diagnostic Testing Initiative**

**Clinical data collection form**

|  |  |  |
| --- | --- | --- |
| **Patient:** | | |
| *Surname:*  Surname | | *Name:*  Name |
| *Date of birth:*  DD/MM/YYYY | *Sex:*  Male  Female | *Ethnic origin:*  Origin |

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Referring medical doctor (the results will be sent to this address):** | | | | |
| *Surname:*  Surname | | | | *Name:*  Name |
| *Institution:*  Institution | | | | |
| *Address:*  Address | | | | |
| *ZIP:*  ZIP | *City:*  City | | *Prov. / State:*  Prov. | *Nation:*  Nation |
| *Tel.:*  Tel. | | *Fax:*  Fax | | *E-mail:*  E-mail |

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **Clinical data:** | | | | | |
| *Weeks of gestation:*  Week | *Birth weight:*  kg | | *Birth weigth percentile:*  Perc. | *Birth length:*  cm | *Birth length percentile:*  Perc. |
| *Current age:*  Years | *Current weight:*  kg | | *Current weight percentile:*  Perc. | *Current height:*  cm | *Current height percentile:*  Perc. |
| Hypotonia at birth with poor suck | | Brief description | | | |
| Hyperphagia (excessive interest in food) | | Brief description | | | |
| Hypogonadism | | Brief description | | | |
| Global developmental delay | | Brief description | | | |
| Cognitive impairment | | Brief description | | | |
| Behavioral problems (tantrums, controlling) | | Brief description | | | |
| Other | | Brief description | | | |

|  |  |
| --- | --- |
| **Previous testing:** | |
| FISH analysis of the 15q11.2 region | Results |
| Methylation assessment of the PWCR (MS-MLPA kit ME028 or MS-PCR) | Results |
| Other | Brief description and results |

|  |  |  |  |
| --- | --- | --- | --- |
| **Consensus clinical diagnostic criteria for Prader-Willi syndrome** | | | |
| **Major criteria** *(1 point each)* | | **Minor criteria** *(1/2 point each)* | |
|  | *Neonatal/infantile hypotonia and poor suck* |  | *Decreased fetal movement and infantile lethargy* |
|  | *Feeding problems and failure to thrive as infant* |  | *Typical behavior problems* |
|  | *Weight gain at 1–6 years; obesity; hyperphagia* |  | *Sleep apnea* |
|  | *Characteristic dysmorphic facial features* |  | *Short stature for family by 15 years* |
|  | *Small genitalia; pubertal delay and insufficiency* |  | *Hypopigmentation for the family* |
|  | *Developmental delay/ intellectual disability* |  | *Small hands and feet for height* |
|  |  |  | *Narrow hands, straight ulnar border* |
|  |  |  | *Esotropia, myopia* |
|  |  |  | *Thick, viscous saliva* |
|  |  |  | *Speech articulation defects* |
|  |  |  | *Skin picking* |
| N° | **Total points major criteria** | N° | **Total points minor criteria** |
| N° | **Total points major and minor criteria** | | |

Requirements for the clinical diagnosis of PWS:

* From birth to 3 years of age: Five (5) total points are required, of which four (4) must be from the major criteria list.
* From 3 years of age to Adulthood: Eight (8) total points are required, including at least five (5) from the major criteria list.

| Age at assessment | **Features Sufficient to Prompt DNA Testing** |
| --- | --- |
| Birth to 2 years | Hypotonia with poor suck |
| 2 – 6 years | Hypotonia with history of poor suck |
| Global developmental delay |
| 6 – 12 years | Hypotonia with history of poor suck |
| Global developmental delay |
| Excessive eating (hyperphagia; obsession with food) with central obesity if uncontrolled |
| >13 years | Hypotonia with history of poor suck |
| Cognitive impairment; usually mild intellectual disability |
| Excessive drive to eat (hyperphagia; obsession with food) with central obesity if uncontrolled |
| Hypothalamic hypogonadism |
| Behavior problems (including temper tantrums and obsessive-compulsive features) |

Please notice 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 criterion.

**References:**

Driscoll DJ, Miller JL, Schwartz S, et al. Prader-Willi Syndrome. 1998 Oct 6 [Updated 2017 Dec 14]. In: Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2019. 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.*