Diagnostic Criteria for Prader-Willi Syndrome

The following criteria for a diagnosis of Prader-Willi Syndrome are based on Holm et al. (Pediatrics 91, 398, 1993). Because infants and young children have fewer symptoms than older children and adults with PWS, the scoring system differs by age group.

Major Criteria

(Count as 1 *point each)*

____ 1. Neonatal and infantile central hypotonia with poor suck, gradually improving with age.

____ 2. Feeding problems in infancy with need for special feeding techniques and poor weight gain/failure to thrive.

____ 3. Excessive (crossing two centile channels) or rapid weight gain on weight-for- length chart after 12 months and before age 6; central obesity in the absence of intervention.

____ 4. Characteristic facial features with dolichocephaly in infancy, narrow face or bifrontal diameter, almond-shaped eyes, small-appearing mouth with thin upper lip, downturned corners of the mouth (three or more of these characteristics required).

____ 5. Hypogonadism-includes any of the following, depending on age:

a) Genital hypoplasia (in males: scrotal hypoplasia, undescended testes, small penis and/or testes; in females: absence or severe hypoplasia of labia minora and/or clitoris).

b) Delayed or incomplete gonadal maturation with delayed pubertal signs after age 16 (in males: small gonads, decreased facial and body hair, lack of voice change; in females: no or infrequent menses).

____ 6. Global developmental delay in a child younger than 6 years; mild to moderate mental retardation or learning problems in older children.

____ 7. Hyperphagia (excessive appetite)/food foraging/obsession with food.

____ 8. Deletion 15q 11-13 (>650 bands, preferably confirmed by fluorescence in situ hybridization) or other appropriate molecular abnormality in this chromosome region, including maternal disomy.

Sum of Major Criteria Points:_____

Diagnostic Criteria for Prader-Willi Syndrome, continued …..

Minor Criteria
(Count as 1/2 *point each.)*

____ 1. Decreased fetal movement or infantile lethargy or weak cry in infancy, improving with age.

____ 2. Characteristic behavior problems, temper tantrums, violent outbursts, and obsessive/compulsive behavior; tendency to be argumentative, oppositional, rigid, manipulative, possessive, and stubborn; perseverating, stealing, and lying (five or more of these symptoms required).

____ 3. Sleep disturbance or sleep apnea.

____ 4. Short stature for genetic background by age 15 (in absence of growth hormone intervention)

____ 5. Hypopigmentation-fair skin and hair compared with other family members.

____ 6. Small hands (less than 25th percentile) and/or feet (less than 10th percentile) for height age.

____ 7. Narrow hands with straight ulnar border (outer edge of hand).

____ 8. Eye abnormalities (esotropia, myopia).

____ 9. Thick, viscous saliva with crusting at corners of the mouth.

____ 10. Speech articulation defects.

____ 11. Skin picking.

Sum of Minor Criteria Points:_____

Sum of Major and Minor Criteria Points: _____

Requirements for a **Diagnosis of PWS:**

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

Supportive Findings
(The following are not scored but increase the certainty of a diagnosis of PWS.)

1. High pain threshold.
2. Decreased vomiting.
3. Temperature instability in infancy or altered temperature sensitivity in older children and adults.
4. Scoliosis or kyphosis (curvature of the spine).
5. Early adrenarche (pubic or axillary hair before age 8).
6. Osteoporosis (demineralization, or thinning, of the bones).
7. Unusual skill with jigsaw puzzles.
8. Normal neuromuscular studies.

Reference: Holm, V.A., Cassidy, S.B., Butler, M.G., Hanchett, J.M., Greenswag, L.R., Whitman, B. Y., & Greenberg, F. (1993). Prader. Willi Syndrome: Concensus diagnostic criteria. Pediatrics, 91,398-402.