Diagnostic Criteria for Prader-Willi Syndrome

TABLE 3. Suggested New Criteria to Prompt DNA Testing for PWS

Age at Assessment	Features Sufficient to Prompt DNA Testing
Birth to 2 y	1. Hypotonia with poor suck.
2y–6 y	1. Hypotonia with history of poor suck. 2. Global developmental delay.
6y–12 y	1. History of hypotonia with poor suck (hypotonia often persists). 2. Global developmental delay. 3. Excessive eating (hyperphagia; preoccupation with food) with central obesity if uncontrolled.
13 y through adulthood	1. Cognitive disabilities; usually mild mental retardation. 2. Excessive eating (hyperphagia; preoccupation with food) with central obesity if uncontrolled. 3. Hypothalamic hypogonadism and/or typical behavior problems (including temper tantrums, perseverative and compulsive-like behaviors).

TABLE 2. Sensitivities and the Percentages of Documentation of the Published Criteria

	% Affected
Major criteria	
Neonatal hypotonia	88
Feeding problems in infancy	79
Excessive weight gain	67
Facial features	88
Hypogonadism	51
Developmental delay	99
Hyperphagia	84
Minor criteria	
Decreased fetal activity	62
Behavior problems	87
Sleep disturbance/sleep apnea	76
Short stature	63
Hypopigmentation	73
Small hands and/or feet	88
Narrow hands/straight ulnar borders	82
Eye abnormalities	68
Thick viscous saliva	89
Articulation defects	80
Skin-picking	83

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Reference:
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