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MENTAL RETARDATION SYNDROMES

PRADER-WILLI SYNDROME: DIAGNOSTIC CRITERIA

Diagnostic criteria for Prader-Willi syndrome (PWS), developed by consensus of seven clinicians in consultation with experts, are reported from the University of Washington, Seattle, and six additional centers. Three categories evolved, scored on a weighted point system: 1) **Major criteria** (1 point) include neonatal hypotonia, feeding problems, excessive weight gain, characteristic narrow facies with almond-shaped eyes and small, down-turned corners of the mouth, hypogonadism, developmental delay with mild to moderate mental retardation or learning problems, hyperphagia and obsession with food, and chromosomal deletion 5q11-13. 2) **Minor criteria** (one half point) include weak cry, behavioral problems, sleep apnea, short stature, fair skin and hair, small hands and feet, myopia, viscous saliva, and speech defects. 3)**Supportive criteria** (unscored) include high pain threshold, temperature instability, scoliosis, osteoporosis, and expert at jigsaw puzzles. Symptoms change with age: 5 points (4 major) required for diagnosis at birth to 3 years; 8 points (5 major) in the 3 year to adult age groups. (Holm VA et al. Prader-Willi syndrome: consensus diagnostic criteria. Pediatrics Feb 1993; 91: 398-402). (Reprints: Vanja A Holm MD, Child Development and Mental Retardation Center, WJ-10, University of Washington, Seattle, WA 98195).

> COMMENT. Until a biological marker for PWS is available, the diagnosis is more an art than a science. These clinical criteria should ensure uniformity and alert the physician to a suspicion of PWS in hypotonic infants and in obese, mildly retarded and behaviorally disturbed adolescents and adults.

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