

## Prader-Willi Methylation

---

### DESCRIPTION:

- Prader-Willi syndrome is a disorder which manifests with hypotonia, feeding problems and delayed development in infancy, and as hyperphagia with obesity, short stature, mental retardation, and behavior disorders in older individuals.
- 99% of patients lack a paternal-specific pattern of methylation (imprint) at chromosome 15q11-q13 due to deletion, maternal uniparental disomy, or imprinting mutation.

### REASONS FOR REFERRAL:

- According to "Diagnostic Testing for Prader-Willi and Angelman Syndromes" (*Am J Hum Genet*, 1996, 58:1085-1088), "If biparental inheritance is identified (by methylation), Prader-Willi syndrome is ruled out on the basis of all cases known to date".
- Confirm or rule out this diagnosis in hypotonic neonates, confirm or exclude a clinical diagnosis in older children.

### METHOD OF ANALYSIS:

- Upon Southern hybridization to a XbaI / NotI digest of genomic DNA, SNRPN probe (ATCC 95679) detects two bands for a normal biparental pattern.
  - Presence of only the maternal band supports a diagnosis of Prader-Willi syndrome.
  - Results are reported within 3 weeks of receipt of sample.
- PLEASE CALL THE LAB WHEN TESTING A HYPOTONIC NEONATE. EXPEDITED RESULTS CAN USUALLY BE PROVIDED IN LESS THAN ONE WEEK. PRENATAL TURNAROUND TIME IS TARGETED AT 14 DAYS OR LESS.

### SAMPLE REQUIREMENTS:

- For DNA testing, 5 to 10 milliliters of blood (minimum 1 ml) in EDTA (purple top) tubes should be sent by overnight carrier at room temperature.
- Prenatal testing: two (2) confluent T25 flasks of amniocytes or cultured CVS cells.

### TEST CPT CODES:

CPT 83890 DNA extraction highly purified  
CPT 83892 DNA enzymatic digestion X 2  
CPT 83894 DNA separation  
CPT 83896 DNA nucleic acid probe  
CPT 83897 DNA nucleic acid transfer  
CPT 83898 DNA amplification  
CPT 83912 DNA interpretation and report

*Discounts from list price are available for institutional billing under contractual arrangement with the laboratory. Contact Ellen Livers at 800-447-6614 ext 7523.*