

Angelman Syndrome Methylation

DESCRIPTION:

- Angelman syndrome is a disorder which manifests with severe developmental delays, absence of speech, ataxia, seizures, and inappropriate bouts of laughter. The disorder is caused by lack of expression maternally-derived genes in the q11 segment of chromosome 15.
- More than 70% of patients lack a maternal-specific pattern of methylation (imprint) at chromosome 15q11-q13 due to deletion, paternal uniparental disomy, or imprinting mutation.
- Remaining patients may have mutation of the UBE3A gene (Matsuura et al., *Nat Gen* 1997, 15:74-77).

REASONS FOR REFERRAL:

- Methylation testing can confirm a diagnosis of Angelman syndrome in a child with consistent symptoms and signs as described above.
- 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)...most identifiable cases of Angelman syndrome are ... ruled out."

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 paternal band supports a diagnosis of Angelman syndrome.
- Results are reported within 3 weeks of receipt of sample.
EXPEDITED CLINICAL 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.