Cincinnati Children's

Division of Human Genetics

Angelman Syndrome

Molecular Genetics Laboratory
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Additional information and test requisitions are available at:
www.cincinnatichildrens.org/molecular-genetics

Helping you fit the pieces together

Shipping Instructions

Please enclose test requisition with sample. All information must be completed before sample can be processed.

Place samples in Styrofoam mailer and ship at room temperature by overnight Federal Express to arrive Monday through Friday

Ship to:
Cytogenetics and Molecular Genetics Laboratories
3333 Burnet Avenue NRB 1042
Cincinnati, OH 45229
513-636-4474

Angelman syndrome (AS) is a rare genetic disorder that affects an individual's growth and development. The estimated prevalence of AS is 1 in 15,000 births. AS affects children of both sexes and all races. Infants with AS often appear normal at birth, but typically develop feeding difficulties, slowed head growth, developmental delays, and seizures during infancy or early childhood. Older children and adults with AS typically have moderate to severe mental retardation, virtual absence of spoken language, seizures, gait abnormalities, specific behavioral differences including inappropriate laughter, and characteristic physical findings including prognathism.

Angelman syndrome often results from the absence of specific genetic material on one of an individual's #15 chromosomes that should have been contributed by the individual's mother at the time of conception. About 70% of cases of AS are due to a deletion of a portion of the maternal #15 chromosome, 2-3% of cases are due to inheritance of two paternal #15 chromosomes (with no maternal contribution) and 3-5% of cases have defects in the imprinting center of the maternal chromosome #15. About 5% of cases are due to mutations within a specific gene termed *UBE3A*. The genetic cause of AS is currently unidentified in approximately 15% of children with the clinical diagnosis of AS.

INDICATIONS:

Confirmation of a suspected diagnosis in an individual with the clinical features of AS.

METHODOLOGY:

Methylation specific polymerase chain reaction (PCR) is performed on sodium bisulfite treated DNA that allows specific amplification of the maternal (methylated) and paternal (un-methylated) *SNRPN* gene at 15q11-13.

SENSITIVITY:

A paternal-only pattern is diagnostic of AS. Additional studies including fluorescent in situ hybridization (FISH) to identify large gene deletions, uniparental disomy (UPD) study and *UBE3A* gene mutation analysis should be considered to determine the specific genetic etiology of the disorder. Currently, the molecular mechanism causing AS is not identified in approximately 15-20% of individuals with the diagnosis. Thus, a normal test result does not rule out AS in an individual with clinical symptoms consistent with AS.

SPECIMEN:

At least 2ml of whole blood in purple top (EDTA) tube. Label tube with patient's name, birth date, and date of collection. Phlebotomist must initial tube to verify patient's identity.

TURN-AROUND TIME:

2-3 weeks

COST:

Please call 1-866-450-4198 for pricing or with any billing questions.

CPT CODES:

81331

RESULTS:

Each test report includes a detailed interpretation of the genetic findings, the clinical significance of the result, and specific recommendations for clinical management and additional testing, if warranted. Results will be reported to the referring physician or health care provider as specified on the test requisition form.