Angelman Syndrome

Description:

Angelman syndrome (AS) is a rare genetic disorder that affects 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, absence of spoken language, seizures, gait abnormalities, specific behavioral differences including inappropriate laughter, and characteristic physical findings including prognathism. Angelman syndrome results from the loss or disruption of genetic material on the maternally inherited chromosome 15. About 70% of cases of AS are due to a chromosome 15 deletion, 2-3% of cases are due paternal uniparental disomy of chromosome 15 (inheritance of two copies of paternal chromosome 15 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 the *UBE3A* gene. The genetic cause of AS is currently unidentified in approximately 15% of children with a clinical diagnosis of AS.

Indications:

- Confirmation of diagnosis in an individual with clinical features of 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.

Testing Methodology:

Testing is performed using methylation-sensitive multiplex ligation-dependent probe amplification (MS-MLPA) to detect the presence of copy number changes (deletions/duplications) and/or methylation defects in or near the Angelman syndrome (AS) critical region of chromosome 15q11.

Turn-Around Time:

21 days

CPT Codes:

- 81331

Please call 1-866-450-4198 for current pricing, insurance preauthorization or with any billing questions.

Results:

Results will be reported to the referring physician or designee as specified on the requisition form.

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

Cincinnati Children's®
changing the outcome together

Laboratory of Genetics and Genomics
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