Prader Willi Syndrome (PWS)

Description:

Prader Willi syndrome (PWS) is a rare genetic disorder that affects growth and development. The prevalence of PWS is approximately 1 in 15,000 births and affects children of both sexes and of all races. Infants with PWS are often first recognized by their low muscle tone, feeding difficulties, and failure to thrive. Older children and adults with PWS typically have unusual eating habits including hyperphagia that can result in morbid obesity. In addition, they often have hypogonadism, short stature, small hands/feet, mild to moderate mental retardation, as well as difficult behaviors including obsessive-compulsive disorder. PWS results from loss or disruption of genetic material on the paternally inherited chromosome 15. About 70% of cases of PWS are due to a chromosome 15 deletion. Nearly 30% of cases of PWS are due to the inheritance of maternal uniparental disomy of chromosome 15 (inheritance of two copies of maternal chromosome 15 with no paternal contribution). In about 1-2% of cases, defects in the imprinting center of the paternally inherited chromosome 15 account for PWS.

Indications:

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

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 Prader Willi syndrome (PWS) 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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