Cincinnati Children's

Division of Human Genetics

Prader Willi Syndrome (PWS)

Molecular Genetics Laboratory
CLIA#: 36D0656333
Phone: (513) 636-4474
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Email: moleculargenetics@cchmc.org

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

Updated 1/2012

PWS is a rare genetic disorder that affects an individual's 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 an absence of genetic material on one of an individual's #15 chromosomes that should have been contributed by the individual's father at the time of conception. About 70% of cases of PWS are due to a deletion of a portion of the paternal #15 chromosome. Nearly 30% of cases of PWS are due to the inheritance of two maternal #15 chromosomes (with no paternal contribution). In about 1-2% of cases, defects in the imprinting center of the paternal chromosome #15 account for PWS.

INDICATIONS:

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

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 maternal-only pattern is diagnostic of PWS. Additional studies including fluorescent in situ hybridization (FISH) to identify large gene deletions and uniparental disomy (UPD) analysis should be considered to determine the specific genetic etiology of the disorder for genetic counseling purposes.

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:

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