EGL Genetics | Eurofins Clinical Diagnostics
2460 Mountain Industrial Boulevard | Tucker, Georgia 30084
Phone: 470-378-2200 or 855-831-7447 | Fax: 470-378-2250
eglgenetics.com

Phenylketonuria (PKU): *PAH* Gene Deletion/Duplication

Test Code: DPAHX
Turnaround time: 2 weeks
CPT Codes: 81228 x1

Condition Description

Phenylketonuria (PKU) results in an inability to metabolize the amino acid phenylalanine to tryosine. If dietary phenylalanine is not metabolized, the amino acid accumulates to neurotoxic levels resulting. If untreated, the condition results in mental retardation, seizures, microcephaly and behavior abnormalities. PKU is among the disorders tested by newborn screening and treated by dietary restriction. The incidence of PKU is approximately 1 in 10,000 live births. PKU is autosomal recessive disorder and caused by mutations in the *PAH* gene (12q22-q24) leading to deficiency of phenylalanine hydroxylase. Disease severity, clinical phenotype, and effectiveness of treatment differs among the different *PAH* mutations and correlates with the level of *PAH* enzyme activity. Complete or near complete absence of enzyme activity results in classical phenylketouria (PKU), which requires strict dietary restraint of phenylalanine for life. Milder enzyme deficiencies can result in non-PKU hyperphenylalaninemia (non-PKU HPA) or variant PKU. Carriers of PKU are unaffected.

Visit www.ThinkGenetic.com for patient-friendly information on phenylketonuria.

References:

- GeneReviews Clinical Summary

Genes

PAH

Indications

This test is indicated for:

- Patients with a biochemical diagnosis of PKU.
- Individuals with biochemical test results indicating carrier status of PKU.
- Individuals who are at risk to be carriers of PKU, when the proband is unavailable for testing.

Sequencing is not appropriate for prenatal samples in which familial mutations have not been identified.

Methodology

DNA isolated from peripheral blood is hybridized to a CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes which cover the entire genomic region.

Detection

Detection is limited to duplications and deletions. Array CGH will not detect point mutations or intronic mutations. Results of molecular analysis must be interpreted in the context of the patient's clinical and/or biochemical phenotype.

Specimen Requirements

Submit only 1 of the following specimen types

Type: Whole Blood (EDTA)

Specimen Requirements:
EDTA (Purple Top)
Infants and Young Children (2 years of age to 10 years old: 3-5 ml
Older Children & Adults: 5-10 ml
Autopsy: 2-3 ml unclotted cord or cardiac blood

Specimen Collection and Shipping:
Ship sample at room temperature for receipt at EGL within 72 hours of collection. Do not freeze.

Type: DNA, Isolated

Specimen Requirements:
Microtainer
3µg
Isolation using the Perkin Elmer™Chemagen™ Chemagen™ Automated Extraction method or Qiagen™ Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping:
Refrigerate until time of shipment in 100 ng/µL in TE buffer. Ship sample at room temperature with overnight delivery.

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Special Instructions

Submit copies of diagnostic biochemical test results with the sample. Sequence analysis is required before deletion/duplication analysis by targeted CGH array. If sequencing is performed outside EGL Genetics, please submit a copy of the sequencing report with the test requisition. Contact the laboratory if further information is needed.

Related Tests

- Organic Acid Analysis (OA) and Plasma Amino Acid (AA) Analysis are used in the diagnoses of a patient with PKU.
- Prenatal testing is available to couples who are confirmed carriers of mutations. Please contact the laboratory genetic counselor to discuss appropriate testing prior to collecting a prenatal specimen.