Condition Description

Tuberous sclerosis is inherited in an autosomal dominant pattern and affects 1 in 6,000 people. In approximately one-third of cases, an affected person inherits an altered TSC1 or TSC2 gene from a parent who has the disorder. The remaining two-thirds of cases are due to de novo mutations in the TSC1 or TSC2 gene. TSC1 mutations appear to be more common in familial cases of tuberous sclerosis complex, while mutations in the TSC2 gene occur more frequently in sporadic cases.

The TSC1 and TSC2 genes provide instructions for making the proteins hamartin and tuberin, respectively. Within cells, these two proteins likely work together to help regulate cell growth and size, and also act as tumor suppressors. Affected individuals are born with one mutated copy of the TSC1 or TSC2 gene in each cell. However, enough protein is usually produced from the other, normal copy of the gene to regulate cell growth effectively. For some types of tumors to develop, a second mutation involving the other copy of the TSC1 or TSC2 gene must occur in certain cells.

Tuberous sclerosis complex is characterized by the growth of numerous noncancerous tumors in many parts of the body. These tumors can occur in the skin, brain, eyes, heart, lungs, kidneys, and other organs, in some cases leading to significant health problems. The condition is extremely variable and is associated with seizures, mental retardation, behavior problems, and skin abnormalities (not only tubers, but also lesions).

Reference:
- GeneReviews.

Genes

TSC1, TSC2

Indications

The test is indicated for:
- Individuals with a clinical or suspected diagnosis of tuberous sclerosis.

Methodology

Deletion/Duplication Analysis: DNA isolated from peripheral blood is hybridized to a gene-targeted CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes that cover the entire genomic region.

Detection

Deletion/Duplication Analysis: Detection is limited to duplications and deletions. The CGH array will not detect point 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: 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.

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 unclootted 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.

**Related Tests**

- Tuberous Sclerosis: Sequencing Panel.