Tuberous Sclerosis: Deletion/Duplication Panel

**Test Code:** MD430  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81405 x1, 81406 x1

### 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 \( \text{TSC1} \) or \( \text{TSC2} \) gene from a parent who has the disorder. The remaining two-thirds of cases are due to \textit{de novo} mutations in the \( \text{TSC1} \) or \( \text{TSC2} \) gene. \( \text{TSC1} \) mutations appear to be more common in familial cases of tuberous sclerosis complex, while mutations in the \( \text{TSC2} \) gene occur more frequently in sporadic cases.

The \( \text{TSC1} \) and \( \text{TSC2} \) 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 \( \text{TSC1} \) or \( \text{TSC2} \) 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 \( \text{TSC1} \) or \( \text{TSC2} \) 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

- \( \text{TSC1} \), \( \text{TSC2} \)

### 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. Please note that a "backbone" of probes across the entire genome are included on the array for analytical and quality control purposes. Rarely, off-target copy number variants causative of disease may be identified that may or may not be related to the patient's phenotype. Only known pathogenic off-target copy number variants will be reported. Off-target copy number variants of unknown clinical significance will not be reported.

**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: Whole Blood

Specimen Requirements:

In EDTA (purple top) tube:  
- Infants (2 years): 3-5 ml  
- Older Children & Adults: 5-10 ml

Specimen Collection and Shipping: Ship sample at room temperature with overnight delivery.

#### Type: Isolated DNA

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Specimen Requirements:

In microtainer: 10 ug

Isolation using the Qiagen™ Puregene kit for DNA extraction is recommended.

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

### Related Tests

- Tuberous Sclerosis: Sequencing Panel.