

Glucose Transporter Type 1 Deficiency Syndrome: *SLC2A1* Gene Sequencing

Test Code: SSLC2

Turnaround time: 2 weeks

CPT Codes: 81479 x1

Condition Description

Glucose transporter type 1 deficiency syndrome (Glut-1-DS) is characterized by infantile seizures that are resistant to treatment by anticonvulsants. This is followed by delays in mental and motor development, head growth retardation, ataxia, dysarthria, spasticity, and uncontrolled eye movement before meals. The most important laboratory observation in Glut-1-DS is a reduced cerebrospinal fluid glucose concentration. Affected infants appear normal at birth. Seizures typically begin between one and four months of age. The frequency, type, and severity of seizures vary among individuals with Glut-1-DS. Additionally, cognitive impairment can range from learning difficulties to severe intellectual disability.

Mutation of the *SLC2A1* (1p35-p31.3) gene cause Glut-1-DS. Glut-1-DS is inherited in an autosomal dominant manner with the majority of mutations occurring de novo. Affected parents have been reported with a mild or subclinical degree of impairment.

For patients with suspected Glut-1-DS, sequence analysis is recommended as the first step in mutation identification. For patients in whom mutations are not identified by full gene sequencing, deletion/duplication analysis is appropriate.

References:

- GeneReviews
- [OMIM #138140](#): *SLC2A1* gene
- [OMIM #606777](#): Glut-1-DS

Genes

[SLC2A1](#)

Indications

This test is indicated for:

- Confirmation of a clinical diagnosis of Glucose Transporter Type 1 Deficiency syndrome.
- Carrier testing in adults with a family history of Glucose Transporter Type 1 Deficiency syndrome.

Methodology

PCR amplification of 10 exons contained in the *SLC2A1* gene is performed on the patient's genomic DNA. Direct sequencing of amplification products is performed in both forward and reverse directions, using automated fluorescence dideoxy sequencing methods. The patient's gene sequences are then compared to a normal reference sequence. Sequence variations are classified as mutations, benign variants unrelated to disease, or variations of unknown clinical significance. Variants of unknown clinical significance may require further studies of the patient and/or family members. This assay does not interrogate the promoter region, deep intronic regions, or other regulatory elements, and does not detect large deletions.

Detection

Sequencing analysis detects 91% of mutations in individuals with Glut-1-DS. Mutations in the promoter region, some mutations in the introns and other regulatory element mutations cannot be detected by this analysis. Large deletions will not be detected by this analysis. Results of molecular analysis should be interpreted in the context of the patient's clinical and/or biochemical phenotype.

Analytical Sensitivity: ~99%

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: Saliva

Specimen Requirements:

Oragene™ Saliva Collection Kit

Oragene™ Saliva Collection Kit used according to manufacturer instructions. Please contact EGL for a Saliva Collection Kit for patients that cannot provide a blood sample.

Specimen Collection and Shipping:

Please do not refrigerate or freeze saliva sample. Please store and ship at room temperature.

Type: DNA, Isolated**Specimen Requirements:**

Microtainer

8µg

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.

Related Tests

- Deletion/duplication analysis of the *SLC2A1* gene by CGH array is available for those individuals in whom sequence analysis is negative.
- Custom diagnostic mutation analysis (KM) is available to family members if mutations are identified by targeted mutation testing or sequencing analysis.
- Prenatal testing is available only for known familial mutations to individuals who are confirmed carriers of mutations. Please contact the laboratory genetic counselor to discuss appropriate testing prior to collecting a prenatal specimen.