Glucose Transporter Type 1 Deficiency Syndrome: \textit{SLC2A1} Gene Deletion/Duplication

| Test Code: DSCL2 |
| Turnaround time: 2 weeks |
| CPT Codes: 81228 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 \textit{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
- \textbf{OMIM \#138140:} \textit{SLC2A1} gene
- \textbf{OMIM \#606777:} Glut-1-DS

**Genes**

\textit{SLC2A1}

**Indications**

This test is indicated for:

- Confirmation of a clinical diagnosis of Glucose Transporter Type 1 Deficiency syndrome in an individual in whom sequence analysis was negative.
- Carrier testing in adults with a family history of Glucose Transporter Type 1 Deficiency syndrome in whom sequence analysis was negative.

**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. 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**

\textit{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 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.

**Special Instructions**

Sequence analysis is required before deletion/duplication analysis by targeted CGH array. If sequencing is performed outside of EGL Genetics, please submit a copy of the sequencing report with the test requisition.

**Related Tests**

- Sequence analysis of the *SLC2A1* gene is available and is required before deletion/duplication analysis.
- 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.