

GLI3 Gene Sequencing Grieg Cephalopolysyndactyly Syndrome Testing

Brief Description: Grieg Cephalopolysyndactyly Syndrome (GCPS) is defined by a spectrum of characteristic anomalies. These include preaxial polydactyly or mixed pre- and postaxial polydactyly, ocular hypertelorism and macrocephaly. Patients with more severe GCPS can have varying degrees of mental retardation and present with hydrocephalus and seizures.

Genetics: The diagnosis of GCPS is based on careful interpretation of the clinical findings and more recently on characterization of *GLI3* gene mutations. Currently, *GLI3* is the only gene known to be associated with GCPS. GCPS is inherited in an autosomal dominant manner. It is thought that mutations in the *GLI3* gene can be identified in perhaps 75% of GCPS patients. Children with a GCPS affected parent have a 50% chance of inheriting the syndrome. De novo gene mutation is also believed to contribute to the incidence of GCPS.

Sequencing for the known coding region of the *GLI3* gene is available at PreventionGenetics. Our mission is to deliver reliable results as rapidly as possible while keeping costs down. In addition we offer exon specific screening to confirm known familial mutations in additional family members and to confirm the results of research testing.

Sensitivity of Test: A positive result reports defining a *GLI3* gene mutation indicates only the potential for the development of GCPS. Further testing is required to confirm the diagnosis of GCPS in this patient.

It is important to note that a negative result for this assay does not rule out other rarer mutations either in other non coding regions of the *GLI3* gene or in other as yet non defined genes that may be contributory to GCPS.

Specimen Requirements

- Collect 2-5 ml of whole blood in EDTA (purple top tube) or ACD (yellow top tube). 5 ml is the preferred volume.
- Only one blood tube is required for multiple tests.
- Ship whole blood specimens at room temperature.
- Do not freeze blood.
- During hot weather, include a frozen ice pack in the shipping container. Do not allow the ice pack to come in direct contact with the specimen tube.
- In cold weather, include an unfrozen ice pack to help moderate extremes in temperature. The DNA in whole blood is stable for at least 48 hours at 21°C, 5-7 days at 4°C.

Sequence analysis of the *GLI3* gene

\$1,490.00

Molec Diag, Ascertainment 83890

Molec Diag, Isolation 83891

Molecular Diag, Amplif x19 83898

Mutat Id By Seq, Single Seg x19 83904

Molecular Diag, Separation 83894

Interpretation And Report 83912

Accreditation Info. CLIA ID #: 52D1027685 (expires 1/18/07) (CAP#: 7185561, AU ID: 1407125 expires 12/20/06)

Ship to:

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