

## Pallister-Hall Syndrome Testing via *GLI3* Gene Sequencing

Pallister-Hall Syndrome (PHS) is defined by a spectrum of characteristic anomalies. These are polydactyly, asymptomatic bifid epiglottis, hypothalamic hamartoma with or without gelastic seizures, and laryngotracheal cleft with neonatal lethality. PHS can lead to pituitary insufficiency which may result in death as neonates from undiagnosed adrenal insufficiency.

The diagnosis of Pallister-Hall syndrome is based on clinical findings and more recently on characterization of *GLI3* gene mutations. *GLI3* is currently the only gene known to be associated with PHS. PHS is inherited in an autosomal dominant manner. Children with a PHS parent have a 50% chance of inheriting the syndrome. De novo gene mutation is also believed to contribute to the incidence of PHS.

Sequencing for all 14 *GLI3* exons is available at PreventionGenetics. This testing is described elsewhere. In addition, we offer exon specific screening to confirm known familial mutations in additional family members. Our mission is to deliver reliable results as rapidly as possible while keeping costs down.

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

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 PHS.

### 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.

Single exon Sequence analysis screening for the presence previously identified mutations in the *GLI3* gene **\$230.00.**

Molec Diag, Ascertainment	83890	
Molec Diag, Isolation	83891	
Molecular Diag, Amplif	83898	Turnaround time is 1~2 weeks
Mutat Id By Seq, Single Seg	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:

Attn: Diagnostics Lab  
PreventionGenetics LLC  
3700 Downwind Drive  
Marshfield, WI 54449 USA  
Contact: 715-387-0484  
FAX: 715-384-3661

### Contact for info:

Dr. Eric W. Johnson  
Director, Molecular Diagnostics and BioBanking  
715-387-0484  
eric.johnson@preventiongenetics.com