SKI– Shprintzen-Goldberg Syndrome

The SKI gene codes for a protein which is involved in the signaling pathway for transforming growth factor beta (TGF-β). This pathway helps regulate cell growth, proliferation, differentiation, motility, and apoptosis. The TGF-β pathway is also involved in activating the SMAD complex. Mutations in the SKI gene block the SMAD complex from entering the nucleus, thus targeted gene activation does not occur. Mutations in the SKI gene (all in exon 1) have been reported to be associated with Shprintzen-Goldberg syndrome (SGS). SGS is characterized by distinctive facial features, skeletal abnormalities, and intellectual disability. Cardiovascular findings associated with SGS include mitral valve prolapse and aortic root dilation. Mutations in the SKI gene are inherited in an autosomal dominant manner. SKI is a 7 exon gene located at 1p36.33.

Indication

SKI gene testing is utilized to confirm a diagnosis of SGS in patients with clinically evident disease.
Methodology:

All 7 exons of the SKI gene, as well as the exon/intron boundaries and portion of untranslated regions of the gene are amplified by PCR. Genomic DNA sequences from both forward and reverse directions are obtained by automatic fluorescent detection using an ABI PRISM® 3730 DNA Analyzer. Sequence variants different from National Center for Biotechnology Information GenBank references are further evaluated for genetic significance. If a mutation is identified, a known familial mutation analysis will be available for additional family members.

Sensitivity & Accuracy:

Greater than 99% of the mutations in exons 1-7 of SKI are detectable by sequence based methods. Sequencing does not detect deletions or duplications.

References:


Specimen:

Peripheral blood in EDTA tube
Adult: 3-5mL
Child: 3-5mL
Infant: 1-3mL
For other specimen types, please contact Amy Shikany at 513-803-3317

Turnaround Time:

Full Mutation Analysis 2-4 weeks
Known Mutation Analysis 1-2 weeks

CPT Codes:

Full Gene Sequencing 81479
Additional Family Members 81403