Lab Dept: Anatomic Pathology

Test Name: CRANIODYSMORPHOLOGY (FGFR) FULL PANEL WITH TWIST SEQUENCING

General Information

Lab Order Codes: FGFRT

Synonyms: FGFR 1, 2, 3 genes and TWIST gene; Fibroblast Growth Factor Receptor gene family; Craniosystosis Panel

CPT Codes: 81400 – Molecular Pathology procedure, Level 1
81401 – Molecular Pathology procedure, Level 2
81404 x2 – Molecular Pathology procedure, Level 5

Test Includes: The Craniodysmorphology Syndrome's test panel combines the individual tests for FGFR 1, 2 and 3 along with examination of the TWIST gene into a panel.

Logistics

Test Indications: Several distinct syndromes, including Crouzon, Apert, Pfeiffer, Jackson-Weiss and the nonsyndromic syndromes, may be caused by different mutations within the Fibroblast Growth Factor Receptor gene family (FGFR 1, 2, and 3). Saethre-Chotzen syndrome is a craniosynostosis condition that is caused by mutations in the TWIST gene. These diseases represent the bulk of genetic conditions resulting in cranial dysmorphologies which are a result of premature closure of the sutures between the bones of the skull. If not surgically corrected, the brain continues to grow within a small skull causing protruding eyeballs, an altered head shape and possible brain damage. These syndromes may be either spontaneous or inherited in an autosomal dominant manner. In the inherited form, the disease is considered fully penetrant. Each disorder has differences in clinical symptoms, but the differences are often subtle and generally overlap.

Reasons for referral:

● To determine whether one of these syndromes is responsible for abnormalities of the skull or eyes.
● To discriminate between syndromes with overlapping clinical symptoms in an affected individual.
● To establish whether symptoms are spontaneous or inherited within a family.
● To evaluate the risk of having a child with one of these syndromes.
● Individuals at risk who wish prenatal diagnosis.

Lab Testing Sections: Anatomic Pathology - Sendouts

Referred to:	Center for Genetic Testing at Saint Francis, Tulsa, Oklahoma
Phone Numbers:	MIN Lab: 612-813-6280 STP Lab: 651-220-6550
Test Availability:	Daily, 24 hours
Turnaround Time:	15 days
Special Instructions:	A Molecular Genetics Laboratory Test Requisition must accompany the specimen. Contact the Molecular Laboratory at 866-846-0315 for more information.

Specimen

Specimen Type:	Whole blood
Container:	Lavender top (EDTA) tube
Draw Volume:	10 mL (Minimum: 0.7 mL) blood (2 tubes of 5 mL each preferred)
Processed Volume:	Same as Draw Volume
Collection:	Routine blood collection. Mix specimen thoroughly by gentle inversion.
Special Processing:	Lab Staff: **Do Not** centrifuge. Specimen should remain in original collection container. Send EDTA blood at room temperature via overnight courier. A Molecular Genetics Laboratory Test Requisition (http://www.sfh-lab.com/MOLREQFORM.pdf) must accompany the specimen. Contact the Center for Genetic Testing at Saint Francis at 866-846-0315 for further information. If blood will not be sent immediately, refrigerate until shipped.
Patient Preparation:	None
Sample Rejection:	Some specimens cannot be analyzed because of improper collection or improper handling in transit. Other specimens cannot be processed until patient information or clinical diagnostic information is received. Please follow collection and transport instructions carefully. Notification of rejected or problem specimens will be made on receipt.

Interpretive

Reference Range:	No mutations detected
Critical Values:	N/A

Limitations:	Deletions of the entire gene or mutations outside the tested regions of the genes will not be detected due to primer/probe site mutations or rare polymorphisms.
Methodology:	PCR and DNA sequencing of exon 7 and 9 (IIIa and IIIc) of the FGFR2 gene and the entire TWIST gene, and point mutation testing of the FGFR1 and FGFR3 genes.
References:	Center for Genetic Testing at Saint Francis, Tulsa, Oklahoma (June 2018) (866) 846-0315 Fax (918) 502-1723
Updates:	5/20/2004: Test previously performed at H.A. Chapman Institute. 2/5/2013: CPT update 6/19/2018: CPT update