General Information

Lab Order Codes:  ACHE

Synonyms:  FRFR3 Gene Sequencing

CPT Codes:  81479 – Molecular Pathology, Unlisted Procedure

Test Includes:  Sequencing of the FGFR3 gene for specific exons 3, 5, 7, 9, 10, 13, 15.

Logistics

Test Indications:  Investigation of symptoms characterized by Achondroplasia (ACH)/Hydrochondroplasia (HCH).

ACH represents the most common skeletal dysplasia in humans. It is characterized by rhizomelic dwarfism, trident hands and short fingers, midface hypoplasia with macrocephaly and fontal bossing. A small foramen magnum may cause generalized hypotonia, hyperrelexia or clonus in newborns. HCH is similar but milder than ACH. Patients are still rhizomelic dwarfs but are not as short, do not have trident hands and the facial manifestations are not as severe. Neurological complications in HCH are not constant findings. HCH is genetically heterogeneous. Not all patients diagnosed clinically with HCH have demonstrable mutations in FGFR3.

Lab Testing Sections:  Anatomic Pathology - Sendouts

Referred to:  CTGT (CTGT Test: 1008)

Phone Numbers:  MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability:  Daily, 24 hours

Turnaround Time:  1-2 weeks
Special Instructions: See Patient Preparation. Request special forms for patient consent, physician signature and clinical history from Children’s Laboratory or print Request form here. Select (FGFR3 Exons 3, 5, 7, 9, 10, 13, 15) under Achondroplasia (ACH)/Hypochondroplasia (HCH).

Note: These completed forms must accompany the specimen or come with the patient when they present at the lab to be drawn.

Specimen

Specimen Type: Whole blood

Container: Lavender top (EDTA) tube

Draw Volume: 6 mL (Minimum: 3 mL) blood

Processed Volume: Same as Draw Volume

Collection: Routine venipuncture

Special Processing: Lab Staff: Do Not process. Specimen should remain as whole blood in original collection container. Send Monday – Thursday via overnight shipping at refrigerated temperatures. Include patient consent and physician signature forms with the specimen. Forward promptly.

Note: For specimens collected Friday – Sunday (or on a holiday), they should be held in Children’s Laboratories at refrigerated temperatures and shipped on Monday (or the next business day Monday – Thursday).

Patient Preparation: None

Sample Rejection: Frozen specimens, mislabeled or unlabeled specimens

Interpretive

Reference Range: No mutations detected

Critical Values: N/A

Limitations: Not all patients diagnosed clinically with HCH have demonstrable mutations in FGFR3.

Methodology: Sanger DNA Sequencing

References: CTGT Web Page January 2013
Phone: 484-244-2900 Fax: 484-244-2904

Update: 7/10/2013: CPT change from 81405 to 81479. CTGT update.