
Lab Dept: Anatomic Pathology

Test Name: ACHONDROPLASIA OR HYPOCHONDROPLASIA (FGR3) REFLEX TO REST OF GENE SEQUENCING

General Information

Lab Order Codes: ACHRF

Synonyms: FRFR3 Gene Sequencing (reflex to remaining exons)

CPT Codes: 81479 – Unlisted molecular pathology procedure

Test Includes: Sequencing of the FGFR3 gene for remaining gene specific exons not previously tested in first level testing. First level testing previously performed included 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 frontal bossing. A small foramen magnum may cause generalized hypotonia, hyperreflexia 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

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours

Turnaround Time: Approximately 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 Reflex to remaining exons) 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: 4 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: DNA Sequencing

References: [CTGT Web Page](#) January 2013
Phone: 484-244-2900 Fax: 484-244-2904