Lab Dept: Anatomic Pathology

Test Name: HEREDITARY MULTIPLE EXOSTOSES (EXT1) SEQUENCING

General Information

Lab Order Codes: EXT1

Synonyms: Exostosin 1; Hereditary multiple osteochondromata; Multiple cartilaginous exostoses; Diaphyseal aclasis; HME

CPT Codes: 81479 – Molecular Pathology Unlisted procedure

Test Includes: Tier 1 analysis for HME includes full gene sequencing of the EXT1 gene and deletion/duplication testing for both the EXT1 and EXT2 genes. Analysis is performed bi-directional sequencing of all coding exons of the EXT1 gene. Mutations found in the first member of a family to be tested are confirmed by repeat analysis using sequencing, heteroduplex or restriction fragment analysis.

Logistics

Test Indications: Individuals with HME often develop benign cartilage-capped tumors (exostoses) at the ends of the long bones or the surface of flat bones. Exostoses develop prior to skeletal maturity only. Bony deformity, bowing of the long bones, limited range of motion, and premature osteoarthrosis may be associated with hereditary multiple exostoses (HME). Exostoses also may cause complications by putting pressure on nearby tissues, nerves or blood vessels. A rare but severe risk in patients with mutiple exostoses is the development of malignant chondrosarcoma, which occurs in 1-5% of patients.

Reasons for referral:
1. Confirmation of a clinical diagnosis
2. Genetic counseling

Lab Testing Sections: Anatomic Pathology - Sendouts

Referred to: GeneDx, Inc. (GDX#: 1811)

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours (Preferred draws are Sunday - Thursday as specimens can only be received at the reference lab Monday - Friday. Specimens collected Friday or Saturday will be held for shipment on Monday.)

Turnaround Time:	6 weeks for new patients
Special Instructions:	A GeneDx signed request form must be sent with any patient or specimen to the laboratory.

Specimen

Specimen Type:	Whole blood
Container:	Lavender top (EDTA) tube
Draw Volume:	1 - 5 mL blood
Processed Volume:	Same as Draw Volume
Collection:	Routine venipuncture for blood specimens, invert gently to mix
Special Processing:	Lab Staff: Send whole blood in original collection container labeled with patient name, date of birth and medical record number, including signed consent form and requisition, with a cool pack, via overnight or second-day courier so that the sample will arrive at GeneDx, Inc. on a weekday (Monday through Friday). Include a cool pack in warm weather. Samples drawn on Friday or Saturday should be held at refrigerated temperatures for shipment on Sunday. **Do not** freeze. **Note:** Specimens may be stored at refrigerated temperatures for up to 7 days prior to shipping.
Patient Preparation:	None
Sample Rejection:	Unrefrigerated specimens older than 48 hours; clotted or hemolyzed for blood; frozen specimens; mislabeled or unlabeled specimens

Interpretive

Reference Range:	No mutations detected
Critical Values:	N/A
Limitations:	Buccal swabs are not allowable for this testing.
Methodology:	Bi-directional sequence analysis and targeted CGH analysis with exon-level resolution
References:	GeneDx, Inc. February 2012 (301) 519-2100 Fax (301) 519-2892
Updates:	2/6/2013: CPT update