Lab Dept: Anatomic Pathology

Test Name: HEREDITARY MULTIPLE EXOSTOSES (EXT1 & EXT2 CONCURRENTLY) SEQUENCING

General Information

Lab Order Codes: EX12

Synonyms: Exostoses 1 and 2; Hereditary multiple osteochondromata; Multiple cartilaginous exostoses; Diaphyseal aclasis; HME

CPT Codes: 81479 x4 – Molecular Pathology Unlisted procedure

Test Includes: Using genomic DNA obtained from the submitted specimen, bi-directional sequence of all 11 coding exons of the EXT1 gene is obtained and analyzed. Concurrently, targeted array CGH analysis with exon-level resolution (ExonArrayDx) is performed to evaluate for a deletion or duplication of one or more exons in both the EXT1 and EXT2 genes. Mutations found in the first person of a family to be tested are confirmed by repeat analysis using sequencing, restriction fragment analysis, or other appropriate method.

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 mutiples exostoses is the development of malignant chondrosarcoma, which in 1-5% of patients. Mutation in the EXT1 gene seems to be associated with a more severe disease and higher risk of developing chondrosarcoma than EXT2 mutations.

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

Lab Testing Sections: Anatomic Pathology - Sendouts

Referred to: GeneDx, Inc. (GDX#: 1813)

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:	4 weeks
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 blood collection, 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 Monday – Saturday. Samples drawn on Saturday should be held at refrigerated temperatures for shipment on Monday. Cool packs should be used in warm weather. **Do not** freeze. **Note:** Specimens may be stored at refrigerated temperature 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 brushes cannot be accepted for this testing.
Methodology:	Capillary sequencing, exon array CGH
References:	GeneDx, Inc. January 2018

Updates: 2/6/2013: CPT update
1/20/2018: CPT, shipping and method update.