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**Lab Dept:**                   **Anatomic Pathology**

**Test Name:**               **HEREDITARY MULTIPLE EXOSTOSES (EXT1)  
SEQUENCING**

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***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.

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***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 osteoarthritis 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 multiple 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 Test: 1811)

**Phone Numbers:**       MIN Lab: 612-813-6280

STP Lab: 651-220-6550

**Test Availability:**       Daily, 24 hours

**Turnaround Time:**       4 weeks for new patients

**Special Instructions:** A GeneDx signed [request form](#) must be sent with any patient or specimen to the laboratory.

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### ***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, Monday through Saturday. Include a cool pack in warm weather. Samples drawn on Saturday or Sunday should be held at refrigerated temperatures for shipment on Monday. **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

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### ***Interpretive***

**Reference Range:** No mutations detected

**Critical Values:** N/A

**Limitations:** N/A

**Methodology:** Capillary sequencing and exon-array CGH

**References:** [GeneDx, Inc.](#) January 2018  
(301) 519-2100 Fax (301) 519-2892

**Updates:** 2/6/2013: CPT update