
Lab Dept: **Anatomic Pathology**

Test Name: **HEREDITARY MULTIPLE EXOSTOSES (EXT1)
KNOWN MUTATION**

General Information

Lab Order Codes: EX1K (Blood)

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

CPT Codes: 81479 – Molecular Pathology Unlisted procedure

Test Includes: Testing of a relative for a specific known mutation (carrier testing) where testing was previously done by GeneDx. Using genomic DNA, the exon or exons of interest are screened by bi-directional sequence analysis and/or by non-sequence methods such as heteroduplex analysis or restriction enzyme digestion. The previously tested proband DNA serves as a positive control.

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 multiples exostoses is the development of malignant chondrosarcoma, which in 1-5% of patients.

Carrier testing reasons for referral:

1. Testing parents of a child with a specific dominant mutation, in order to determine if the mutation in the child is new or inherited.
2. Carrier testing in parents of a child with apparently homozygous recessive mutations, to rule out the possibility that the child has one mutated allele and one allele that is deleted or refractory to amplification.
3. Carrier testing in the parents of a child with recessive mutations, to confirm that all four parental alleles can be detected prior to prenatal diagnosis.
4. Pre-symptomatic testing in siblings of the index case.
5. Carrier testing in the extended family.

Lab Testing Sections: Anatomic Pathology - Sendouts

Referred to: GeneDx, Inc. (GDx#: 9011, Specify gene/mutation)

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

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|------------------------------|---|
| Test Availability: | Daily, 24 hours |
| Turnaround Time: | 2 - 3 weeks |
| Special Instructions: | A GeneDx signed request form must be sent with any patient or specimen to the laboratory. |

Specimen

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| 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. Use a cool pack with the sample 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 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

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|-------------------------|--|
| Reference Range: | Interpretive report |
| Critical Values: | N/A |
| Limitations: | N/A |
| Methodology: | Capillary sequencing |
| References: | GeneDx, Inc. January 2018 (301) 519-2100 Fax (301) 519-2892 |

Updates:

1/11/2011: CPT update. Buccal specimens are no longer accepted for this testing. Blood is the only acceptable specimen.
2/6/2013: CPT update