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Possible link between multiple hereditary osteochrondromas and EXT3.

Multiple Hereditary Osteochrondroma/Exostoses (MHE) induces benign bone tumors at the ends of long bones and often leads to skeletal deformities and tumor formations at the end of these bones. Recent research suggests an under-laying cause associated with glycosyltransferases involved in the synthesis of heparin sulfates. Two genes designated EXT 1 and EXT 2 have been mapped and linked to MHE. Although phenotypes have been associated with both genes, there seems an additional possible link between the third EXT (EXT3). Little is known of the contributions from glycosyltransferases 3. Although the gene has been cloned and mapped there is little information associated with MHE's. We suggest the possibility that several of the distinct bone deformities are associated with mutations in this genetic variant. In this study, we examine possible gene variants in EXT3 through PCR. Patient samples included affected individuals with varied phenotypes as well as control samples. Two patients with unilateral metatarsal deformations were examined as suggest a possible phenotypic link to EXT3.