EXT1 gene
exostosin glycosyltransferase 1

Normal Function

The EXT1 gene provides instructions for producing a protein called exostosin-1. This protein is found in a cell structure called the Golgi apparatus, which modifies newly produced enzymes and other proteins. In the Golgi apparatus, exostosin-1 attaches (binds) to another protein, exostosin-2, to form a complex that modifies heparan sulfate. Heparan sulfate is a complex of sugar molecules (a polysaccharide) that is added to proteins to form proteoglycans, which are proteins attached to several sugars. Heparan sulfate is involved in regulating a variety of body processes including blood clotting and the formation of blood vessels (angiogenesis). It also has a role in the spreading (metastasis) of cancer cells.

Health Conditions Related to Genetic Changes

Hereditary multiple osteochondromas

About 480 mutations in the EXT1 gene have been identified in people with hereditary multiple osteochondromas type 1, a condition in which people develop multiple benign (noncancerous) bone tumors called osteochondromas. Most of these mutations are known as "loss-of-function" mutations because they prevent any functional exostosin-1 protein from being made. The loss of functional exostosin-1 protein prevents it from forming a complex with the exostosin-2 protein and adding heparan sulfate to proteins. It is unclear how this impairment leads to the signs and symptoms of hereditary multiple osteochondromas.

Trichorhinophalangeal syndrome type II

The EXT1 gene is located in a region of chromosome 8 that is deleted in people with trichorhinophalangeal syndrome type II (TRPS II). TRPS II is a condition that causes bone and joint malformations including multiple osteochondromas (described above); distinctive facial features; intellectual disability; and abnormalities of the skin, hair, teeth, sweat glands, and nails. As a result of this deletion, affected individuals are missing one copy of the EXT1 gene in each cell. A shortage of exostosin-1 protein causes the osteochondromas in people with TRPS II. The deletion of other genes near the EXT1 gene likely contributes to the additional features of this condition.
Chromosomal Location

Cytogenetic Location: 8q24.11, which is the long (q) arm of chromosome 8 at position 24.11

Molecular Location: base pairs 117,799,363 to 118,111,819 on chromosome 8 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI

Other Names for This Gene
• exostoses (multiple) 1
• exostosin 1
• EXT
• EXT1_HUMAN
• Glucuronosyl-N-acetylglucosaminyl-proteoglycan 4-alpha-N-acetylglucosaminyltransferase
• N-acetylglucosaminyl-proteoglycan 4-beta-glucuronosyltransferase

Additional Information & Resources

Clinical Information from GeneReviews
• Hereditary Multiple Osteochondromas
  https://www.ncbi.nlm.nih.gov/books/NBK1235
• Trichorhinophalangeal Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK425926

Scientific Articles on PubMed
• PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28EXT1%5BTIAB%5D%29+OR+%28exostoses+++1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D
Catalog of Genes and Diseases from OMIM

- EXOSTOSIN GLYCOSYLTRANSFERASE 1
  http://omim.org/entry/608177

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/EXT1ID212.html

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=EXT1%5Bgene%5D

- HGNC Gene Symbol Report

- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:2131

- NCBI Gene

- UniProt
  https://www.uniprot.org/uniprot/Q16394

Sources for This Summary

- OMIM: EXOSTOSIN GLYCOSYLTRANSFERASE 1
  http://omim.org/entry/608177

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11432960
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1757186/

  Determination of the mutation spectrum of the EXT1/EXT2 genes in British Caucasian patients with multiple osteochondromas, and exclusion of six candidate genes in EXT negative cases. Hum Mutat. 2006 Nov;27(11):1160.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17041877

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/28426188


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