

IGHMBP2 gene

immunoglobulin mu DNA binding protein 2

Normal Function

The *IGHMBP2* gene provides instructions for making an enzyme called immunoglobulin mu DNA binding protein 2 (IGHMBP2). This enzyme functions as a helicase, which means that it attaches to particular regions of DNA and temporarily unwinds the two spiral strands of these molecules. When a cell prepares to divide to form two cells, the chromosomes are duplicated (replicated) so that each new cell will get a complete set of chromosomes. The replication process involves unwinding the DNA so that it can be copied. This mechanism is also involved in the production of RNA, a chemical cousin of DNA. Additionally, the IGHMBP2 protein is thought to be involved in the production of proteins from RNA through a process called translation. The IGHMBP2 protein is produced in cells throughout the body.

Health Conditions Related to Genetic Changes

Spinal muscular atrophy with respiratory distress type 1

More than 60 mutations in the *IGHMBP2* gene have been found to cause spinal muscular atrophy with respiratory distress type 1 (SMARD1). SMARD1 is an inherited condition that causes muscle weakness and respiratory failure typically beginning in infancy. Most mutations that cause this condition change single protein building blocks (amino acids) in the IGHMBP2 protein and disrupt the protein's ability to unwind DNA and RNA. The loss of helicase function impedes DNA replication and the production of RNA and proteins. These problems particularly affect alpha-motor neurons, which are specialized cells in the brainstem and spinal cord that control muscle movements. Altered IGHMBP2 proteins cause these neurons to become damaged and die over time, although the exact mechanism is unknown. The cumulative death of alpha-motor neurons leads to breathing problems and progressive muscle weakness in children with SMARD1.

Charcot-Marie-Tooth disease

MedlinePlus Genetics provides information about Charcot-Marie-Tooth disease

Other Names for This Gene

- cardiac transcription factor 1
- CATF1
- FLJ34220
- FLJ41171
- HCSA
- HMN6
- immunoglobulin mu binding protein 2
- SMARD1
- SMBP2_HUMAN
- SMUBP2

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of IGHMBP2 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3508\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3508[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28IGHMBP2%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2160+days%22%5Bdp%5D>)

Catalog of Genes and Diseases from OMIM

- IMMUNOGLOBULIN MU-BINDING PROTEIN 2; IGHMBP2 (<https://omim.org/entry/600502>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/3508>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=IGHMBP2\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=IGHMBP2[gene]))

References

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Genomic Location

The *IGHMBP2* gene is found on chromosome 11 (<https://medlineplus.gov/genetics/chromosome/11/>).

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