

HIVEP2 gene

HIVEP zinc finger 2

Normal Function

The *HIVEP2* gene provides instructions for making a protein that functions as a transcription factor. Transcription factors attach (bind) to specific regions of DNA and help control the activity (expression) of particular genes. The HIVEP2 protein is most abundant in the brain, where it controls the expression of multiple genes, many of which are involved in brain growth and development. This protein may also play a role in the function of immune system cells and the process of bone remodeling, in which old bone is broken down and new bone is created to replace it. It may also be involved in other body processes; however these additional roles are not completely understood.

Health Conditions Related to Genetic Changes

HIVEP2-related intellectual disability

At least nine mutations in the *HIVEP2* gene have been found in individuals with a neurological disorder called *HIVEP2*-related intellectual disability. This condition is characterized by delayed development of speech and walking, moderate to severe intellectual disability, mild physical abnormalities (dysmorphic features), and weak muscle tone (hypotonia), among other features. The *HIVEP2* gene mutations are thought to lead to a shortage of functional HIVEP2 protein. It is unclear how these genetic changes result in the features associated with *HIVEP2*-related intellectual disability, although researchers speculate that a shortage of the HIVEP2 protein alters the expression of several genes involved in brain growth and development. Abnormalities in the growth and development of the brain likely underlie the cognitive problems and other neurological features of *HIVEP2*-related intellectual disability. It is unclear how *HIVEP2* gene mutations contribute to the unusual physical features and health problems that can occur with this condition.

Other Names for This Gene

- c-myc intron binding protein 1
- HIV-EP2
- MBP-2
- MHC binding protein-2

- MIBP1
- MRD43
- Schnurri-2
- SHN2
- ZAS2
- ZNF40B

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of HIVEP2 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3097\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3097[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28HIVEP2%5BTIAB%5D%29+OR+%28human+immunodeficiency+virus+type+I+enhancer+binding+protein+2%5BTIAB%5D%29%29+OR+%28%28HIV-EP2%5BTIAB%5D%29+OR+%28MBP-2%5BTIAB%5D%29+OR+%28MHC+binding+protein-2%5BTIAB%5D%29+OR+%28MIBP1%5BTIAB%5D%29+OR+%28SHN2%5BTIAB%5D%29+OR+%28Schnurri-2%5BTIAB%5D%29+OR+%28ZAS2%5BTIAB%5D%29+OR+%28c-myc+intron+binding+protein+1%5BTIAB%5D%29+OR+%28transcription+factor+HIVEP2%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D%29>)

Catalog of Genes and Diseases from OMIM

- HUMAN IMMUNODEFICIENCY VIRUS TYPE 1 ENHANCER-BINDING PROTEIN 2; HIVP2 (<https://omim.org/entry/143054>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/3097>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=HIVEP2\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=HIVEP2[gene]))

References

- Dorflinger U, Pscherer A, Moser M, Rummele P, Schule R, Buettner R. Activation of somatostatin receptor II expression by transcription factors MIBP1 and SEF-2 in the murine brain. *Mol Cell Biol.* 1999 May;19(5):3736-47. doi:10.1128/MCB.19.5.3736. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/10207097/>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC84194/>)

- Fukuda S, Yamasaki Y, Iwaki T, Kawasaki H, Akieda S, Fukuchi N, Tahira T, Hayashi K. Characterization of the biological functions of a transcription factor, c-myc intron binding protein 1 (MIBP1). *J Biochem*. 2002 Mar;131(3):349-57. doi: 10.1093/oxfordjournals.jbchem.a003109. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/11872163>)
- Iwashita Y, Fukuchi N, Waki M, Hayashi K, Tahira T. Genome-wide repression of NF-kappaB target genes by transcription factor MIBP1 and its modulation by O-linked beta-N-acetylglucosamine (O-GlcNAc) transferase. *J Biol Chem*. 2012 Mar 23; 287(13):9887-9900. doi: 10.1074/jbc.M111.298521. Epub 2012 Jan 31. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/22294689>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3323019/>)
- Jones DC, Schweitzer MN, Wein M, Sigrist K, Takagi T, Ishii S, Glimcher LH. Uncoupling of growth plate maturation and bone formation in mice lacking both Schnurri-2 and Schnurri-3. *Proc Natl Acad Sci U S A*. 2010 May 4;107(18):8254-8. doi: 10.1073/pnas.1003727107. Epub 2010 Apr 19. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/20404140>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2889532/>)
- Srivastava S, Engels H, Schanze I, Cremer K, Wieland T, Menzel M, Schubach M, Biskup S, Kreiss M, Ende S, Strom TM, Wieczorek D, Zenker M, Gupta S, Cohen J, Zink AM, Naidu S. Loss-of-function variants in HIVEP2 are a cause of intellectual disability. *Eur J Hum Genet*. 2016 Apr;24(4):556-61. doi: 10.1038/ejhg.2015.151. Epub 2015 Jul 8. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/26153216>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4929870/>)
- Staton TL, Lazarevic V, Jones DC, Lanser AJ, Takagi T, Ishii S, Glimcher LH. Dampening of death pathways by schnurri-2 is essential for T-cell development. *Nature*. 2011 Apr 7;472(7341):105-9. doi: 10.1038/nature09848. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/21475200>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3077958/>)
- Steinfeld H, Cho MT, Retterer K, Person R, Schaefer GB, Danylchuk N, Malik S, Wechsler SB, Wheeler PG, van Gassen KL, Terhal PA, Verhoeven VJ, van Slegtenhorst MA, Monaghan KG, Henderson LB, Chung WK. Mutations in HIVEP2 are associated with developmental delay, intellectual disability, and dysmorphic features. *Neurogenetics*. 2016 Jul;17(3):159-64. doi: 10.1007/s10048-016-0479-z. Epub 2016 Mar 22. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/27003583>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4907844/>)

Genomic Location

The *HIVEP2* gene is found on chromosome 6 (<https://medlineplus.gov/genetics/chromosome/6/>).

Last updated January 1, 2017