

ZFP57 gene

ZFP57 zinc finger protein

Normal Function

The protein produced from the *ZFP57* gene is a member of a family called zinc finger proteins, which are involved in many cellular functions. Zinc finger proteins each contain one or more short regions called zinc finger domains. These regions include a specific pattern of protein building blocks (amino acids) and one or more charged atoms of zinc (zinc ions).

Zinc finger proteins attach (bind) primarily to DNA. In most cases, these proteins attach to regions near certain genes and turn the genes on and off as needed. Proteins that bind to DNA and regulate the activity of particular genes are known as transcription factors. Some zinc finger proteins can also bind to other molecules, including RNA (a chemical cousin of DNA) and proteins.

The ZFP57 protein is involved in the regulation of other genes by the addition of methyl groups, consisting of one carbon atom and three hydrogen atoms (methylation). Methylation is important in many cellular functions. These include determining whether the instructions in a particular segment of DNA are carried out or suppressed (gene silencing), regulating reactions involving proteins and lipids, and controlling the processing of chemicals that relay signals in the nervous system (neurotransmitters).

Health Conditions Related to Genetic Changes

6q24-related transient neonatal diabetes mellitus

Mutations in the *ZFP57* gene cause about 10 percent of cases of 6q24-related transient neonatal diabetes mellitus, a type of diabetes that occurs in infants. This disorder results from the overactivity (overexpression) of certain genes in a region of the long (q) arm of chromosome 6 called 6q24.

People inherit two copies of their genes, one from their mother and one from their father. Usually both copies of each gene are active, or "turned on," in cells. In some cases, however, only one of the two copies is normally turned on. Which copy is active depends on the parent of origin: some genes are normally active only when they are inherited from a person's father; others are active only when inherited from a person's mother. This phenomenon is known as genomic imprinting.

The 6q24 region includes paternally expressed imprinted genes, which means that normally only the copy of each gene that comes from the father is active. The copy of each gene that comes from the mother is inactivated (silenced) by methylation.

ZFP57 gene mutations cause a generalized impairment of gene silencing called hypomethylation of imprinted loci (HIL), which affects many imprinted regions including the imprinted region of 6q24. The resulting overactivity of genes in the 6q24 region that are involved with the regulation of insulin secretion and the self-destruction of cells (apoptosis) may reduce the number of insulin-secreting beta cells or impair their function in affected individuals. Insulin controls how much glucose (a type of sugar) is passed from the blood into cells for conversion to energy. A shortage of insulin results in the impaired blood glucose control associated with diabetes mellitus.

Because HIL resulting from *ZFP57* gene mutations can affect expression of many imprinted genes, this mechanism may account for the additional health problems that occur in some people with 6q24-related transient neonatal diabetes mellitus, such as malformations of the brain, heart, or kidneys.

Other Names for This Gene

- bA145L22
- bA145L22.2
- C6orf40
- TNDM1
- zfp-57
- ZFP57_HUMAN
- zinc finger protein 57 homolog
- zinc finger protein 57 homolog (mouse)
- zinc finger protein 698
- ZNF698

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ZFP57 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=346171\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=346171[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28ZFP57%5BTIAB%5D%29+OR+%28%28TNDM1%5BTIAB%5D%29+OR+%28zfp-57%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+AND+%22last+3600+days%22%5Bdp%5D%29>)

Catalog of Genes and Diseases from OMIM

- ZFP57 ZINC FINGER PROTEIN; ZFP57 (<https://omim.org/entry/612192>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/346171>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=ZFP57\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=ZFP57[gene]))

References

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Genomic Location

The *ZFP57* gene is found on chromosome 6 (<https://medlineplus.gov/genetics/chromosome/6/>).

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