POGZ gene

pogo transposable element derived with ZNF domain

Normal Function

The *POGZ* gene provides instructions for making a protein that is found in the cell nucleus. The POGZ protein is part of a group known as zinc finger proteins, which 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). The folded configuration of the zinc finger domain stabilizes the protein and allows it to attach (bind) to other molecules.

In the cell nucleus, the POGZ protein attaches (binds) to chromatin, which is the network of DNA and proteins that packages DNA into chromosomes. Binding of the POGZ protein is part of the process that changes the structure of chromatin (chromatin remodeling) to alter how tightly regions of DNA are packaged. Chromatin remodeling is one way gene activity (expression) is regulated; when DNA is tightly packed gene expression is lower than when DNA is loosely packed. Regulation of gene expression by the POGZ protein is thought to be important to brain development, but the specific function of POGZ in the brain is not well understood.

Health Conditions Related to Genetic Changes

White-Sutton syndrome

At least 17 *POGZ* gene mutations have been found to cause White-Sutton syndrome. This disorder is characterized by intellectual disability, specific facial features, and other signs and symptoms affecting various parts of the body, particularly vision problems and gastrointestinal problems. Most affected individuals have features of autism spectrum disorder (ASD), a varied condition characterized by impaired social skills, communication problems, and repetitive behaviors. *POGZ* gene mutations are thought to impair the ability of the POGZ protein to bind to chromatin, leading to abnormal gene expression that affects development of the brain and other body systems. However, little is known about the specific changes in gene expression and how they lead to the development of intellectual disability and other signs and symptoms of White-Sutton syndrome.

Autism spectrum disorder

POGZ gene mutations have also been identified in people with ASD without other typical features of White-Sutton syndrome (described above). While the exact disease mechanism is unknown, studies suggest that the *POGZ* gene mutations associated with ASD result in a POGZ protein with impaired ability to bind to chromatin. Lack of chromatin binding likely results in abnormal chromatin remodeling, altering the normal expression of genes involved in brain development and leading to ASD. It is unclear why *POGZ* gene mutations lead to White-Sutton syndrome in some individuals and ASD without additional signs and symptoms in others.

Other Names for This Gene

- KIAA0461
- MRD37
- putative protein product of Nbla00003
- WHSUS
- zinc finger protein 280E
- zinc finger protein 635
- ZNF280E
- ZNF635
- ZNF635m

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of POGZ (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=23126[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28POGZ%5BTIAB%5D%29+OR+%28pogo+transposable+element+derived+with+ZNF+domain%5BTIAB%5D%29%29+OR+%28%28KIAA0461%5BTIAB%5D%29+OR+%28zinc+finger+protein+280E%5BTIAB%5D%29+OR+%28zinc+finger+protein+635%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)

Catalog of Genes and Diseases from OMIM

- POGO TRANSPOSABLE ELEMENT-DERIVED PROTEIN WITH ZNF DOMAIN; POGZ (https://omim.org/entry/614787)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/23126)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=POGZ[gene])

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Genomic Location

The *POGZ* gene is found on chromosome 1 (https://medlineplus.gov/genetics/chromosome/1/).

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