CEP57 gene

centrosomal protein 57

Normal Function

The *CEP57* gene provides instructions for making a protein whose function is not completely understood. Within cells, the CEP57 protein is located in structures called centrosomes. Centrosomes have a role in cell division and the assembly of microtubules. Microtubules are fibers that help cells maintain their shape, assist in the process of cell division, and are essential for the movement (transport) of materials within cells. CEP57 seems especially important for the organization and stability of specialized microtubules called spindle microtubules, which are important for cell division. Before cells divide, they copy all of their chromosomes. Spindle microtubules, which are produced by centrosomes, attach to the duplicated chromosomes and pull one copy of each to opposite ends of the cell so that each new cell contains one complete set of chromosomes.

The CEP57 protein is also involved in the transport of certain molecules along microtubules, particularly a protein called fibroblast growth factor 2 (FGF2). FGF2 is an important signaling molecule that helps regulate growth and development of cells and tissues, and its transport inside the cell is important for relaying signals that instruct the cell how to function.

Health Conditions Related to Genetic Changes

Mosaic variegated aneuploidy syndrome

At least three *CEP57* gene mutations have been found to cause mosaic variegated aneuploidy (MVA) syndrome type 2. This condition is characterized by cells with abnormal numbers of chromosomes, a situation known as aneuploidy. Affected individuals grow slowly before and after birth and may have mild intellectual disability and other health problems.

CEP57 gene mutations involved in MVA syndrome type 2 likely reduce the amount of functional CEP57 protein in cells. It is unclear how these changes lead to the features of the condition. Researchers speculate that impairment of the protein's role in stabilizing spindle microtubules may prevent the normal separation of chromosomes during cell division, leading to aneuploidy. Although they are unsure how *CEP57* gene mutations lead to the other features of MVA syndrome type 2, some suggest that the shortage of

functional CEP57 protein prevents proper transport of FGF2, which may impair cell signaling and lead to problems with growth and development.

Other Names for This Gene

- centrosomal protein 57kDa
- centrosomal protein of 57 kDa isoform a
- centrosomal protein of 57 kDa isoform b
- centrosomal protein of 57 kDa isoform c
- FGF2-interacting protein
- KIAA0092
- MVA2
- PIG8
- proliferation-inducing protein 8
- testis-specific protein 57
- translokin
- TSP57

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of CEP57 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=9702[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28CEP57%5BTIAB%5D%29+OR+%28centrosomal+protein+57%5BTIAB%5D%29%29+OR+%28%28FGF2-interacting+protein%5BTIAB%5D%29+OR+%28KIAA0092%5BTIAB%5D%29+OR+%28MVA2%5BTIAB%5D%29+OR+%28PIG8%5BTIAB%5D%29+OR+%28TSP57%5BTIAB%5D%29+OR+%28centrosomal+protein+57kDa%5BTIAB%5D%29+OR+%28centrosomal+protein+of+57+kDa+isoform+a%5BTIAB%5D%29+OR+%28centrosomal+protein+of+57+kDa+isoform+b%5BTIAB%5D%29+OR+%28centrosomal+protein+of+57+kDa+isoform+c%5BTIAB%5D%29+OR+%28proliferation-inducing+protein+8%5BTIAB%5D%29+OR+%28testis-specific+protein+57%5BTIAB%5D%29+OR+%28translokin%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

- CENTROSOMAL PROTEIN, 57-KD; CEP57 (https://omim.org/entry/607951)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/9702)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=CEP57[gene])

References

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Genomic Location

The *CEP57* gene is found on chromosome 11 (https://medlineplus.gov/genetics/chromosome/11/).

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