MPPH Syndrome

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CCND2 gene

CCND2 gene

cyclin D2

Normal Function

The CCND2 gene provides instructions for making a protein called cyclin D2. Cyclins are a family of proteins that control how cells proceed through the multi-step cycle of cell division. Cyclin D2 helps to regulate a step in the cycle called the G1-S transition, in which the cell moves from the G1 phase, when cell growth occurs, to the S phase, when the cell's DNA is copied (replicated) in preparation for cell division. Cyclin D2's role in the cell division cycle makes it a key controller of the rate of cell growth and division (proliferation) in the body.The cyclin D2 protein is regulated by a chemical signaling pathway called the PI3K-AKT-mTOR pathway. This signaling influences many critical cell functions, including the creation (synthesis) of new proteins, cell proliferation, and the survival of cells. The PI3K-AKT-mTOR pathway is essential for the normal development of many parts of the body, including the brain.

Health Conditions Related to Genetic Changes

Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome

At least seven mutations in the CCND2 gene have been found to cause megalencephaly-polymicrogyria-polydactyly-hydrocephalus (MPPH) syndrome. This rare condition affects the development of the brain, causing an unusually large brain and head size (megalencephaly) and a brain abnormality called bilateral perisylvian polymicrogyria (BPP). Some affected individuals also have an extra finger or toe on one or more of their hands or feet (polydactyly).Each of the known mutations changes a single protein building block (amino acid) in the cyclin D2 protein. These changes prevent the protein from being broken down (degraded) when it is no longer needed. The resulting buildup of cyclin D2 in cells triggers them to continue dividing when they otherwise would not have, leading to abnormal cell proliferation. In the brain, the increased number of cells leads to rapid and abnormal brain growth starting before birth. It is less clear how a buildup of cyclin D2 contributes to polydactyly, although the extra digits are probably related to abnormal cell proliferation in the developing hands and feet.

More About This Health Condition

Other Names for This Gene

G1/S-specific cyclin-D2 KIAK0002 MPPH3

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

Tests of CCND2

Scientific Articles on PubMed

PubMed

Catalog of Genes and Diseases from OMIM

CYCLIN D2; CCND2

Gene and Variant Databases

NCBI Gene

ClinVar

References

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Citation on PubMed

Tsunekawa Y, Kikkawa T, Osumi N. Asymmetric inheritance of Cyclin D2 maintains proliferative neural stem/progenitor cells: a critical event in brain development and evolution. Dev Growth Differ. 2014 Jun;56(5):349-57. doi: 10.1111/dgd.12135. Epub 2014 May 17. Citation on PubMed

Genomic LocationThe CCND2 gene is found on chromosome 12.

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