MEGF8 gene

multiple EGF like domains 8

Normal Function

The *MEGF8* gene provides instructions for making a protein whose function is unclear. Based on its structure, the Megf8 protein may be involved in cell processes such as attaching cells to one another (cell adhesion) and helping proteins interact with each other. Researchers also suspect that the Megf8 protein plays a role in the normal shaping (patterning) of many parts of the body during embryonic development.

Health Conditions Related to Genetic Changes

Carpenter syndrome

At least six mutations in the *MEGF8* gene have been found to cause Carpenter syndrome, a condition characterized by irregular skull formation, finger and toe abnormalities, and many other features. These mutations reduce or eliminate the function of the Megf8 protein. Researchers suspect that the amount of protein function that is retained may contribute to the variability in signs and symptoms. It is unclear how *MEGF8* gene mutations cause Carpenter syndrome. The mutations likely interfere with normal patterning of many parts of the body, which contributes to the features of this disorder.

Other Names for This Gene

- C19orf49
- EGF-like domain-containing protein 4
- EGF-like-domain, multiple 4
- EGFL4
- epidermal growth factor-like protein 4
- FLJ22365
- HBV pre-s2 binding protein 1
- MEGF8_HUMAN
- multiple EGF-like-domains 8
- multiple epidermal growth factor-like domains protein 8

- SBP1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of MEGF8 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1954[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28MEGF8%5BTIAB%5D%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

- MULTIPLE EPIDERMAL GROWTH FACTOR-LIKE DOMAINS 8; MEGF8 (https://omim.org/entry/604267)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/1954)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=MEGF8[gene])

References

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Genomic Location

The *MEGF8* gene is found on chromosome 19 (https://medlineplus.gov/genetics/chromosome/19/).

Last updated May 1, 2013