Multiple cutaneous and mucosal venous malformations

Description

Multiple cutaneous and mucosal venous malformations (also known as VMCM) are bluish patches (lesions) on the skin (cutaneous) and the mucous membranes, such as the lining of the mouth and nose. These lesions represent areas where the underlying veins and other blood vessels did not develop properly (venous malformations). The lesions can be painful, especially when they extend from the skin into the muscles and joints, or when a calcium deposit forms within the lesion causing inflammation and swelling.

Most people with VMCM are born with at least one venous malformation. As affected individuals age, the lesions present from birth usually become larger and new lesions often appear. The size, number, and location of venous malformations vary among affected individuals, even among members of the same family.

Frequency

VMCM appears to be a rare disorder, although its prevalence is unknown.

Causes

Mutations in the *TEK* gene (also called the *TIE2* gene) cause VMCM. The *TEK* gene provides instructions for making a protein called TEK receptor tyrosine kinase. This receptor protein triggers chemical signals needed for forming blood vessels (angiogenesis) and maintaining their structure. This signaling process facilitates communication between two types of cells within the walls of blood vessels, endothelial cells and smooth muscle cells. Communication between these two cell types is necessary to direct angiogenesis and ensure the structure and integrity of blood vessels.

TEK gene mutations that cause VMCM result in a TEK receptor that is always turned on (overactive). An overactive TEK receptor is thought to disrupt the communication between endothelial cells and smooth muscle cells. It is unclear how a lack of communication between these cells causes venous malformations. These abnormal blood vessels show a deficiency of smooth muscle cells while endothelial cells are maintained. Venous malformations cause lesions below the surface of the skin or mucous membranes, which are characteristic of VMCM.

Learn more about the gene associated with Multiple cutaneous and mucosal venous malformations

- TEK

Inheritance

VMCM is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to increase the risk of developing venous malformations.

Some gene mutations are acquired during a person's lifetime and are present only in certain cells. These changes, which are not inherited, are called somatic mutations. Researchers have discovered that some VMCM lesions have one inherited and one somatic *TEK* gene mutation. It is not known if the somatic mutation occurs before or after the venous malformation forms. As lesions are localized and not all veins are malformed, it is thought that the inherited mutation alone is not enough to cause venous malformations.

In most cases, an affected person has one parent with the condition.

Other Names for This Condition

- Mucocutaneous venous malformations
- VMCM
- VMCM1

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Multiple Cutaneous and Mucosal Venous Malformations (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1838437/)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Catalog of Genes and Diseases from OMIM

- VENOUS MALFORMATIONS, MULTIPLE CUTANEOUS AND MUCOSAL; VMCM (https://omim.org/entry/600195)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28venous+malformations%5BTIAB%5D%29+AND+%28TIE2%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

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