Desmoid tumor

Description

A desmoid tumor is an abnormal growth that arises from connective tissue, which is the tissue that provides strength and flexibility to structures such as bones, ligaments, and muscles. Affected individuals typically develop a single tumor, although some have multiple tumors. Desmoid tumors most often develop when people are in their 30s or 40s, although they can occur anytime between adolescence and late adulthood.

Tumors that form in the abdomen or abdominal wall are called abdominal desmoid tumors, those that arise from the tissue that connects the abdominal organs are called intra-abdominal desmoid tumors, and tumors found in other regions of the body are called extra-abdominal desmoid tumors. Extra-abdominal tumors occur most often in the shoulders, upper arms, and upper legs.

Desmoid tumors are fibrous, much like scar tissue. They are generally noncancerous (benign) because they do not spread to other parts of the body (metastasize); however, they can aggressively invade the surrounding tissue and can be very difficult to remove surgically. Desmoid tumors can recur, even after they are removed. In about 20 percent of cases, the tumors shrink or disappear with minimal or no treatment (spontaneously regress).

Desmoid tumors may not cause any signs or symptoms. When they do cause symptoms, the most common one is pain. The pain is often due to the tumor pressing against nearby organs, tissues, or blood vessels. Other signs and symptoms are often caused by growth of the tumor into the surrounding tissue, and they can vary based on the size and location of the tumor. Intra-abdominal desmoid tumors can block the bowel, causing constipation. Extra-abdominal desmoid tumors can restrict the movement of affected joints, making it difficult to move the arms or legs.

Desmoid tumors can also occur in combination with other conditions. Desmoid tumors are found in 10 to 30 percent of people with an inherited form of colon cancer called familial adenomatous polyposis (FAP). These individuals typically develop intra-abdominal desmoid tumors in addition to abnormal growths (called polyps) and cancerous tumors in the colon. Desmoid tumors that are not part of FAP are described as sporadic.

Frequency

Desmoid tumors are rare, affecting an estimated 2 to 6 per 1 million people worldwide. In the United States, about 1,000 new cases are diagnosed per year. About 10 percent of all desmoid tumors occur in people with FAP. For reasons that are unclear, women develop desmoid tumors more often than men with women accounting for 70 percent of cases.

Causes

Variants (also called mutations) in the *CTNNB1* gene account for around 90 percent of sporadic desmoid tumors. Variants in the *APC* gene cause the desmoid tumors that are associated with FAP. Both genes are involved in an important cell signaling pathway that controls the growth and division (proliferation) of cells and the process by which cells mature to carry out specific functions (differentiation).

The *CTNNB1* gene provides instructions for making a protein called beta-catenin. This protein interacts with other proteins to control the activity (expression) of particular genes, which helps promote cell proliferation and differentiation. *CTNNB1* gene variants lead to an abnormally stable beta-catenin protein that is not broken down when it is no longer needed. The protein accumulates in cells, where it continues to function and allows uncontrolled cell proliferation and the formation of desmoid tumors.

The protein produced from the *APC* gene helps regulate the levels of beta-catenin in the cell by attaching (binding) to other proteins to form a complex. When this complex binds to beta-catenin, it helps to break down the protein. Variants in the *APC* gene cause cells to produce an abnormally short version of the APC protein that is unable to form the complex. As a result, beta-catenin is not broken down and, instead, accumulates in cells. Excess beta-catenin promotes uncontrolled growth and division of cells, causing the formation of polyps and, occasionally, desmoid tumors in people with FAP.

Learn more about the genes associated with Desmoid tumor

- APC
- CTNNB1

Inheritance

Most desmoid tumors are not inherited. These tumors are caused by variants in genes that occur during a person's lifetime, called somatic variants. Somatic variants in the *CTNNB1* gene can cause sporadic desmoid tumors. A somatic variant in one copy of the gene is sufficient to cause the disorder.

An inherited variant in one copy of the *APC* gene causes FAP and predisposes affected individuals to develop desmoid tumors. FAP is inherited in an autosomal dominant pattern, which means one copy of the altered *APC* gene in each cell is sufficient to cause the disorder. In most cases, an affected person has one parent with the condition.

Other Names for This Condition

- Aggressive fibromatosis
- Desmoid fibromatosis
- Desmoid-type fibromatosis
- Familial infiltrative fibromatosis
- Hereditary desmoid disease
- Musculoaponeurotic fibromatosis

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Desmoid disease, hereditary (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1851124/)

Genetic and Rare Diseases Information Center

- Desmoid tumor (https://rarediseases.info.nih.gov/diseases/1820/index)

Patient Support and Advocacy Resources

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

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Desmoid tumor%22)

Catalog of Genes and Diseases from OMIM

- DESMOID DISEASE, HEREDITARY; DESMD (https://omim.org/entry/135290)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28Fibromatosis,+Aggressive%5BMAJR%5D%29+AND+%28desmoid+tumor%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D)

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