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. Typically, a single tumor develops, although some people have multiple tumors. The tumors can occur anywhere in the body. Tumors that form in the 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 not considered cancerous (malignant) 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. These tumors often recur, even after apparently complete removal.

The most common symptom of desmoid tumors is pain. Other signs and symptoms, which are often caused by growth of the tumor into surrounding tissue, 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 and cause limping or difficulty moving the arms or legs.

Desmoid tumors occur frequently in 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 an inherited condition are described as sporadic.

Frequency

Desmoid tumors are rare, affecting an estimated 1 to 2 per 500,000 people worldwide. In the United States, 900 to 1,500 new cases are diagnosed per year. Sporadic desmoid tumors are more common than those associated with familial adenomatous polyposis.

Causes

Mutations in the *CTNNB1* gene or the *APC* gene cause desmoid tumors. *CTNNB1* gene mutations account for around 85 percent of sporadic desmoid tumors. *APC* gene mutations cause desmoid tumors associated with familial adenomatous polyposis as well as 10 to 15 percent of sporadic desmoid tumors. 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. As part of the cell-signaling pathway, beta-catenin interacts with other proteins to control the activity (expression) of particular genes, which helps promote cell proliferation and differentiation. *CTNNB1* gene mutations 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 in an uncontrolled way.

The protein produced from the *APC* gene helps regulate levels of beta-catenin in the cell. When beta-catenin is no longer needed, the APC protein attaches (binds) to it, which signals for it to be broken down. Mutations in the *APC* gene that cause desmoid tumors lead to a short APC protein that is unable to interact with beta-catenin. As a result, beta-catenin is not broken down and, instead, accumulates in cells. Excess beta-catenin, whether caused by *CTNNB1* or *APC* gene mutations, promotes uncontrolled growth and division of cells, allowing the formation of desmoid tumors.

Learn more about the genes associated with Desmoid tumor

- APC
- CTNNB1

Inheritance

Most desmoid tumors are sporadic and are not inherited. Sporadic tumors result from gene mutations that occur during a person's lifetime, called somatic mutations. A somatic mutation in one copy of the gene is sufficient to cause the disorder. Somatic mutations in either the *CTNNB1* or the *APC* gene can cause sporadic desmoid tumors.

An inherited mutation in one copy of the *APC* gene causes familial adenomatous polyposis and predisposes affected individuals to develop desmoid tumors. The desmoid tumors occur when a somatic mutation occurs in the second copy of the *APC* gene. In these cases, the condition is sometimes called hereditary desmoid disease.

Other Names for This Condition

- Aggressive fibromatosis
- Deep fibromatosis
- Desmoid 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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Last updated March 1, 2013