Lymphedema-distichiasis syndrome

Description

Lymphedema-distichiasis syndrome is a condition that affects the normal function of the lymphatic system, which is a part of the circulatory and immune systems. The lymphatic system produces and transports fluids and immune cells throughout the body. People with lymphedema-distichiasis syndrome develop puffiness or swelling (lymphedema) of the limbs, typically the legs and feet. Another characteristic of this syndrome is the growth of extra eyelashes (distichiasis), ranging from a few extra eyelashes to a full extra set on both the upper and lower lids. These eyelashes do not grow along the edge of the eyelid, but out of its inner lining. When the abnormal eyelashes touch the eyeball, they can cause damage to the clear covering of the eye (cornea). Related eye problems can include an irregular curvature of the cornea causing blurred vision (astigmatism) or scarring of the cornea. Other health problems associated with this disorder include swollen and knotted (varicose) veins, droopy eyelids (ptosis), heart abnormalities, and an opening in the roof of the mouth (a cleft palate).

All people with lymphedema-distichiasis syndrome have extra eyelashes present at birth. The age of onset of lymphedema varies, but it most often begins during puberty. Males usually develop lymphedema earlier than females, but all affected individuals will develop lymphedema by the time they are in their forties.

Frequency

The prevalence of lymphedema-distichiasis syndrome is unknown. Because the extra eyelashes can be overlooked during a medical examination, researchers believe that some people with this condition may be misdiagnosed as having lymphedema only.

Causes

Lymphedema-distichiasis syndrome is caused by mutations in the *FOXC2* gene. The *FOXC2* gene provides instructions for making a protein that plays a critical role in the formation of many organs and tissues before birth. The FOXC2 protein is a transcription factor, which means that it attaches (binds) to specific regions of DNA and helps control the activity of many other genes. Researchers believe that the FOXC2 protein has a role in a variety of developmental processes, such as the formation of veins and the development of the lungs, eyes, kidneys and urinary tract, cardiovascular system, and the transport system for immune cells (lymphatic vessels).

Learn more about the gene associated with Lymphedema-distichiasis syndrome

- FOXC2

Inheritance

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

Other Names for This Condition

- Distichiasis-lymphedema syndrome
- Lymphedema with distichiasis

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Distichiasis-lymphedema syndrome (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0265345/)

Genetic and Rare Diseases Information Center

- Lymphedema-distichiasis syndrome (https://rarediseases.info.nih.gov/diseases/333/index)

Patient Support and Advocacy Resources

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

Catalog of Genes and Diseases from OMIM

- LYMPHEDEMA-DISTICHIASIS SYNDROME; LPHDST (https://omim.org/entry/153400)

Scientific Articles on PubMed

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

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