

# Lymphedema-Distichiasis Syndrome

Subjects: Genetics & Heredity

Contributor: Camila Xu

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.

Keywords: genetic conditions

---

## 1. Introduction

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.

## 2. 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.

## 3. 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).

### 3.1. The gene associated with Lymphedema-distichiasis syndrome

- *FOXC2*

## 4. 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.

## 5. Other Names for This Condition

- distichiasis-lymphedema syndrome

- lymphedema with distichiasis

---

## References

1. Brice G, Mansour S, Bell R, Collin JR, Child AH, Brady AF, Sarfarazi M, Burnand KG, Jeffery S, Mortimer P, Murday VA. Analysis of the phenotypic abnormalities in lymphoedema-distichiasis syndrome in 74 patients with FOXC2 mutations or linkage to 16q24. *J Med Genet.* 2002 Jul;39(7):478-83.
2. Erickson RP, Dagenais SL, Caulder MS, Downs CA, Herman G, Jones MC, Kerstjens-Frederikse WS, Lidral AC, McDonald M, Nelson CC, Witte M, Glover TW. Clinical heterogeneity in lymphoedema-distichiasis with FOXC2 truncating mutations. *J Med Genet.* 2001 Nov;38(11):761-6.
3. Fang J, Dagenais SL, Erickson RP, Arlt MF, Glynn MW, Gorski JL, Seaver LH, Glover TW. Mutations in FOXC2 (MFH-1), a forkhead family transcription factor, are responsible for the hereditary lymphedema-distichiasis syndrome. *Am J Hum Genet.* 2000 Dec;67(6):1382-8.
4. Mellor RH, Brice G, Stanton AW, French J, Smith A, Jeffery S, Levick JR, Burnand KG, Mortimer PS; Lymphoedema Research Consortium. Mutations in FOXC2 are strongly associated with primary valve failure in veins of the lower limb. *Circulation.* 2007 Apr 10;115(14):1912-20.
5. Sutkowska E, Bator A, Trompeta K, Szuba A. Different lymphoscintigraphic patterns in patients with lymphedema distichiasis. *Lymphology.* 2010 Jun;43(2):73-7.
6. Vreeburg M, Heitink MV, Damstra RJ, Moog U, van Geel M, van Steensel MA. Lymphedema-distichiasis syndrome: a distinct type of primary lymphedema caused by mutations in the FOXC2 gene. *Int J Dermatol.* 2008 Nov;47 Suppl 1:52-5. doi:10.1111/j.1365-4632.2008.03962.x.

---

Retrieved from <https://encyclopedia.pub/entry/history/show/11695>