ECTOPIA LENTIS SYNDROME

In some families, dislocation of the lens of the eye (ectopia lentis) is the predominant feature that passes from generation to generation. Sometimes, this occurs along with some of the skeletal (bone and joint) features of Marfan syndrome. This combination of features is called ectopia lentis syndrome.

Ectopia lentis syndrome is differentiated from Marfan syndrome by imaging to ensure that there is no enlargement of the aorta.

People with ectopia lentis syndrome do not have the heart and blood vessels problems associated with Marfan syndrome. Therefore, it can only be differentiated from Marfan syndrome or emerging Marfan syndrome through ongoing medical follow-up, including frequent echocardiograms to ensure that there is no enlargement of the aorta, the large blood vessel that takes blood away from the heart.

What other names do people use for ectopia lentis syndrome?

Ectopia lentis syndrome is also known as familial ectopia lentis.

How prevalent is ectopia lentis syndrome?

Ectopia lentis syndrome is much less common than Marfan syndrome. There aren't any good estimates regarding the frequency of this condition in the general population.

What are the characteristics of ectopia lentis syndrome?

Features of ectopia lentis syndrome include:

- Nearsightedness (myopia)
- Fluctuating or blurred vision
- Increased pressure in the eye (glaucoma) or retinal detachment
- Variable skin and skeletal manifestations that are also found in Marfan syndrome

What causes ectopia lentis syndrome?

The most common cause of ectopia lentis syndrome is a change in the FBN1 (fibrillin-1) gene, the same gene that causes Marfan syndrome. In some families, ectopia lentis is passed down as a dominant trait; that is, a child only needs to inherit a single abnormal copy of the gene from an affected parent to have the condition. Less commonly, ectopia lentis syndrome can be inherited as a recessive trait; that is, a child needs to inherit an abnormal copy of the defective gene from both parents. In this circumstance, the parents are unaffected because each has one normal copy of the gene in question. Recessive ectopia lentis is not associated with a risk for vascular disease. Two genes have been associated with recessive familial ectopia lentis syndrome: ADAMTSL4 and LTBP2. An ophthalmologist can often distinguish between dominant and recessive ectopia lentis based upon family history and physical findings.

How is ectopia lentis syndrome diagnosed?

The diagnosis of ectopia lentis syndrome is made by an ophthalmologist using a slit lamp exam. Anyone suspected of having dominant ectopia lentis syndrome should have a comprehensive evaluation by a geneticist and cardiologist to exclude the diagnosis of Marfan syndrome.

Is there a genetic test for this gene?

Gene testing is available for both dominant and recessive ectopia lentis syndrome.

How is ectopia lentis syndrome managed?

Ectopia lentis syndrome may be treated in several ways:

- Blurred vision caused by lens dislocation can be corrected with glasses or contact lenses (special flat contacts may be required for proper correction).
- The use of aphakic (refracting around the lens) glasses, in combination with Atropine sulfate 1% ophthalmic drops to enlarge the pupil, serve as an effective alternative to surgery.
- In severe cases, the lens of the eye may need to be removed. Every effort should be made to avoid surgery until absolutely necessary to achieve adequate vision because of potential complications of surgery. Artificial lenses can be placed in the eye after lens removal once growth is near complete.

What is the life expectancy of someone with ectopia lentis syndrome?

Life expectancy is normal for people with ectopia lentis syndrome. Patients with dominant ectopia lentis syndrome should have intermittent echocardiograms to exclude aortic root enlargement, even if this has not been seen in other family members because it can impact life expectancy if it not treated.

Do you have questions? Would you like more information?

- Call our help center, 800-862-7326, ext. 126 to speak with a nurse who can answer your questions and send you additional information.
- Visit our website at marfan.org. You can print information that interests you and ask questions online.