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My46 Trait Profile

Stickler syndrome

Other Names: Hereditary progressive arthro-ophthalmopathy

Stickler syndrome is a genetic condition affecting main body systems including the eyes, ears, and joints. It is caused by mutations in the genes COL2A1, COL11A1, COL11A2, COL9A1 and COL9A2, each of which makes a protein involved in the development of connective tissues.

Characteristics of Stickler syndrome

Stickler syndrome is an inherited disorder of connective tissues. Features of the condition include complications in the eyes, ears, joints, and heart. Findings in the eye include: nearsightedness (myopia), cataracts, problem with the gel that fills the eyeball (vitreous anomaly), and dislocation of the lens of the eye (retinal detachments). Ear problems include hearing loss either due to nerve deafness or deafness due to changes in the inner ear. People with Stickler syndrome also are known to be loose jointed, and can have early onset arthritis and mild changes in the formation of the bones of the spine. Individuals with Stickler syndrome may have a small chin, flattened midface, and cleft palate. Some people with Stickler syndrome also have mitral valve prolapse (a weakness in one of the heart valves).

Although there is an increased risk of delayed language development, learning disabilities, or other mild cognitive problems in children with cleft lip and/or palate from any cause, the average IQ of individuals with SS is not significantly different from that of the general population.

There are five types of Stickler syndrome: Stickler syndrome type I, Stickler syndrome type II, Stickler syndrome type III, Stickler syndrome type IV, and Stickler syndrome type V. Individuals with Stickler syndrome type I tend to have milder hearing loss, nearsightedness, and type 1 membranous vitreous anomaly. Individuals with Stickler syndrome type II tend to have more severe hearing loss, nearsightedness, and type 2 beaded vitreous anomaly. Individuals with Stickler syndrome type III tend to have hearing loss and joint problems, however do not have eye problems. Since Stickler syndrome type IV and Stickler syndrome type V are quite rare, little is known about the characteristics of these two types.

Diagnosis/Testing

Individuals with Stickler syndrome have a change or mutation in one of several genes involved in the development of connective tissues. At present, there are 5 different collagen genes that have been associated with Stickler syndrome. Most people with Stickler syndrome have mutations in the COL2A1 gene, but mutations have also been found in the COL11A1, COL11A2, COL9A1, and COL9A2 genes. The effects of mutations that result in Stickler syndrome are very variable from person to person even in the same family.

Management/Surveillance

Management of Stickler syndrome involves evaluations of the different organ systems that can be involved. Affected individuals should be evaluated to see if they have a cleft palate. Regular eye and hearing evaluations are recommended through adulthood. Evaluation by a cardiology for mitral valve prolapse is also recommended. Due to the risk of lens dislocation, individuals with Stickler syndrome should avoid contact sports.

Mode of inheritance

Stickler syndrome may be inherited in one of two patterns of inheritance: autosomal dominant and autosomal recessive. They are explained below.

Autosomal dominant inheritance:

Stickler syndrome is most often inherited in an autosomal dominant pattern. This means inherited one COL2A1, COL11A1 or COL11A2 mutation is enough for an individual to be affected and show signs of Stickler syndrome. The mutation can be inherited from an affected parent or it can occur brand new (de novo) in an affected child.

Autosomal recessive inheritance:

Autosomal recessive inheritance accounts for a small proportion of Stickler syndrome. This inheritance pattern means that an individual has to inherit two mutations (i.e., one from each parent) to be affected. If both parents are carriers of a mutation, they have a 1 in 4 (25%) chance with each pregnancy of having a child with the condition.

Risk to family members

The risk to family members depends on the pattern of inheritance.

Autosomal dominant inheritance:

The risk to family members depends on whether or not the individual with Stickler syndrome has a parent affected with the condition. If a parent also has Stickler syndrome, the risk of having a child with Stickler syndrome is 50% with each pregnancy. If a parent does not have Stickler syndrome, the risk of other siblings being affected is very low.

Autosomal recessive inheritance:

Parents of a child with Stickler syndrome are carriers of Stickler syndrome. If a sibling of a child with Stickler syndrome is unaffected, he/she has a 2 in 3 (66%) chance of being a carrier of Stickler syndrome.

Special considerations

None

Resources

Cleft Palate Foundation
http://www.cleftline.org/
Stickler Involved People
http://www.sticklers.org/sip2/
Children's Craniofacial Association
http://www.ccakids.com/
Genetics Home Reference: Stickler syndrome
http://ghr.nlm.nih.gov/condition/stickler-syndrome

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