Marfan Syndrome

Marfan syndrome is caused by mutations in the FBN1 gene. These lead to abnormalities in the protein fibrillin, which is an essential component of connective tissues. The severity of the symptoms differs widely between different individuals with the defective gene. The syndrome often leads to eye defects, such as lens displacement and myopia. There is an increased risk of retinal detachment, glaucoma, and cataracts. Cardiovascular defects, such as an enlarged aorta and heart valve problems, are common, and can be life threatening. Medication, typically beta blockers, or surgery may be needed to reduce the risk of serious heart failure. Musculoskeletal disorders often occur: these include loose joints, protrusion or indentation of the sternum, and curvature of the spine. Those with the syndrome tend to be unusually tall and thin.

Among the general population the risk of having Marfan's syndrome is about 1 in 5,000, although a parent with the syndrome has a 50:50 risk of their child inheriting the faulty copy of the gene, due to its nature as an autosomal dominant disorder. The disease seems to be spread evenly among all ethnic groups.

Sources

NIH, Genetics Home Reference: Marfan Syndrome. See

http://ghr.nlm.nih.gov/condition/marfan-syndrome

National Center for Biotechnology: Marfan Syndrome. See

http://www.ncbi.nlm.nih.gov/books/NBK1335/