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Perspcetive

Genetic Insights in Marfan Syndrome

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DESCRIPTION

The pleiotropic connective tissue disorder known as Marfan syndrome (MFS) is inherited as an autosomal dominant trait and is brought on by mutations in the FBN1 gene, which codes for fibrillin, a crucial protein of the extracellular matrix that helps determine the final form of a micro fibril. There aren't many cases of autosomal recessive transmission reported worldwide. 66 exons make up the FBN1 gene, which is found on chromosome 15q21.1. The clinical signs and symptoms that aid in the diagnosis of MFS before concentrating on cardiovascular symptoms, medical and surgical treatments for thoracic aortic aneurysms and/or dissections (TAAD), the mechanisms underlying the development of aneurysms and acute dissections, and biomarkers associated with the emergence of TAADs. A Dutch study examined the effectiveness of losartan. A blocker of the angiotensin II receptor-1; no further therapy clinical trial). They discovered that losartan slows the rate of aortic dilatation in Marfan syndrome patients. Later, they also discovered that losartan has no significant effect on patients with dominant negative (qualitative) mutations, but has a favorable effect on patients with Marfan syndrome who have an FBN1 mutation that results in haploinsufficiency (quantitative mutation). In addition, a French team conducted a 3-year experiment in which individuals with Marfan syndrome receiving beta-receptor blockade were given losartan or a placebo. Losartan lowers blood pressure, but it has no impact on the increase in aortic diameter, the researchers found. Therefore, beta-receptor blockers continue to be the standard of care for Marfan syndrome patients and serum transforming growth factor beta, total homo cysteine, and lysyl oxidase as three potential biochemical indicators. Additionally, plasma indicators of oxidative stress have been linked to the clinical characteristics of one possibility for potential biomarkers of clinical severity is Marfan syndrome.

Microfibril

Mutations in fibrillin-1, a crucial part of the elastic microfibril, are linked to the classic Marfan syndrome. Fibrillin-1 is a 350 kD glycoprotein that is processed and released into the extracellular matrix from a 375 kD precursor (ECM). Latent transforming growth factor-binding proteins (LTBPs) are stabilized in the ECM by polymerizing to create microfibrils. Transforming growth factor (TGF) is kept dormant by LTBPs. TGF signaling may become excessive if fibrillin-1 and LTBPs are unable to interact properly. The majority of fibrillin-1 mutations are missense, the microfibrillar assembly may be negatively affected. As with ectopia lentis, which is frequently linked to missense mutations that result in cysteine substitutions within the epidermal growth factor-like domains of the protein, other aspects of the Marfan phenotype may be caused by abnormal fibrillin or decreased amounts of fibrillin (haploinsufficiency), despite the fact that cysteine residues are essential to the function of the suspensory ligament of the eye. It has been demonstrated that the severity of the disorder varies greatly; different mutations in the same codon can cause either the severe neonatal form of Marfan syndrome or the more prevalent adult variant. Similarly, mutations in the gene's core region (exons 24-32), sometimes known as the "neonatal region," may be linked to phenotypes ranging from severe neonatal Marfan syndrome to isolated Ectopia lentis.

Genetic insight of the disease

The MFS disease-related gene is a mutation in the fibrillin-1 gene (FBN1) on 15q21.1) is the primary cause of MFS (MFS1), and the mark is seen in MFS. The 230-kb gene FBN1 produces the structural protein fibrillin-1, a matrix that is found in both elastic and non-elastic tissues, which has been linked to the second kind of this condition called MFS type 2 (MFS2) with often minor or non-existent ocular involvement. When combined with normal fibrillin proteins in micro fibrils, aberrant fibrillin proteins produced as a result of FBN1 mutations lead to connective tissues that are structurally less sound. The majority of the alterations, or 2/3 of them, involve replacing cysteine.

About 13% of all reported mutations are small insertions, deletions, or duplications. The remaining 13% of the identified mutations are different types of splicing genes. The two main types of mutations found in the FBN1 gene are premature termination codons (PTCs) and in-frame mutations.

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