Sick sinus syndrome

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Sick sinus syndrome (also known as sinus node dysfunction) is a group of related heart conditions that can affect how the heart beats. "Sick sinus" refers to the sinoatrial (SA) node, a natural pacemaker. The SA node generates electrical impulses that start each heartbeat. These signals travel from the SA node to the rest of the heart, signaling the heart (cardiac) muscle to contract and pump blood. In people with sick sinus syndrome, the SA node does not function normally. In some cases, it does not produce the right signals to trigger a regular heartbeat. In others, abnormalities disrupt the electrical impulses and prevent them from reaching the rest of the heart. Sick sinus syndrome tends to cause bradycardia, occasionally tachycardia or tachycardia-bradycardia syndrome. Symptoms include dizziness, light-headedness, syncope, palpitations, and confusion or memory problems. During exercise, many affected individuals experience chest pain, difficulty breathing, or fatigue. Once symptoms of sick sinus syndrome appear, they usually worsen with time. However, some people with the condition never experience any related health problems.

Sick sinus syndrome occurs most commonly in older adults, although it can be diagnosed in people of any age. The condition increases the risk of several life-threatening problems involving the heart and blood vessels. These include a heart rhythm abnormality called atrial fibrillation, heart failure, cardiac arrest, and stroke. Sick sinus syndrome accounts for 1 in 600 patients with heart disease who are over age 65. The incidence of this condition increases with age.

Sick sinus syndrome can result from genetic or environmental factors. In many cases, the cause of the condition is unknown.

Genetic changes are an uncommon cause of sick sinus syndrome. Mutations in two genes, SCN5Aand HCN4, have been found to cause the condition in a small number of families. These genes provide instructions for making proteins called ion channels that transport positively charged atoms (ions) into cardiac cells, including cells that make up the SA node. The flow of these ions is essential for creating the electrical impulses that start each heartbeat and coordinate contraction of the cardiac muscle. Mutations in these genes reduce the flow of ions, which alters the SA node's ability to create and spread electrical signals. These changes lead to abnormal heartbeats and the other symptoms of sick sinus syndrome.

A particular variation in another gene, MYH6, appears to increase the risk of developing sick sinus syndrome. The protein produced from the MYH6 gene forms part of a larger protein called myosin, which generates the mechanical force needed for cardiac muscle to contract. Researchers believe that the MYH6 gene variation changes the structure of myosin, which can affect cardiac muscle contraction and increase the likelihood of developing an abnormal heartbeat.

More commonly, SSS is caused by other factors that alter the structure or function of the SA node. These include a variety of heart conditions, other disorders such as muscular dystrophy, abnormal inflammation, or a shortage of oxygen (hypoxia). Certain medications, such as drugs given to treat abnormal heart rhythms or high blood pressure, can also disrupt SA node function. One of the most common causes of SSS in children is trauma to the SA node, such as damage that occurs during heart surgery.SIn older adults, SSS is often associated with age-related changes in the heart. Over time, the SA node may harden and develop scar-like damage (fibrosis) that prevents it from working properly. Most cases of SSS are not inherited. They are described as sporadic, which means they occur in people with no history of the disorder in their family.

When SSS results from mutations in the HCN4 gene, it has an autosomal dominant pattern of inheritance. Autosomal dominant inheritance means that one copy of the altered gene in each cell is sufficient to cause the disorder. In most cases, an affected person has one parent with the condition.

When SSS is caused by mutations in the SCN5A gene, it is inherited in an autosomal recessive pattern.

Genetic Testing Registry:

I.Sick Sinus Syndrome 1: Location 3p22.2, MIM 608567, Inheritance AR, Gene SCN5A

II.Sick Sinus Syndrome 2;INHERITANCEAutosomal dominantCARDIOVASCULAR

Heart

-Sinus bradycardia

- Atrial fibrillation (in some patients)
- Ventricular fibrillation (in some patients)
- Cardiac arrest (rare)
- Left ventricular noncompaction (in some patients)
- Biventricular hypertrabeculation (in some patients)
- Left ventricular hypertrophy (in some patients)
- Mitral valve prolapse (in some patients)
- Mitral valve myxoid degeneration (in some patients)
- Aortic valve regurgitation (in some patients)

VascularDilation of ascending aorta (in some patients)

III.Sick Sinus Syndrome 3Gene MYH6 Locus 14q11.2