

CARDIOVASCULAR ASPECTS OF NOONAN SYNDROME

Noonan Syndrome (NS) is a syndrome that is characterised by short stature, facial anomalies and heart defects

Features of Noonan Syndrome

Heart defects	90%
Undescended testicles	77%
Feeding problems	76%
Easy bruising	56%
Short stature	50%
Development delay	10%

Congenital Heart Defects

Congenital Heart Defects (CHDs) are defects that occur during the development of a baby's heart and are present at birth. Congenital heart defects may occur as a familial condition, occur sporadically or can be associated with syndromes such as Noonan Syndrome.

Congenital Heart Defects in Noonan Syndrome

Noonan Syndrome (NS) is the most common syndrome associated with congenital heart disease after Down syndrome.

Pulmonary Stenosis

Pulmonary Stenosis refers to a narrowing of the pulmonary valve in the pulmonary artery, which extends from the right ventricle to the lungs. As a result, the right side of the heart must work harder to pump all of the blood through the narrowed valve. This may result in a thickening of the right side of the heart. This is the commonest feature of Noonan syndrome and is treated by opening the pulmonary valve to let more blood pass through the valve (see below)

Hypertrophic Cardiomyopathy

The main feature of hypertrophic cardiomyopathy is an excessive thickening of the heart muscle. In Noonan Syndrome there may be heart failure with cardiomyopathy but it tends to improve with age in some children.

Atrial Septal Defects

An Atrial Septal Defect is a hole in the dividing wall between the two upper chambers of the heart. As a result, blood flows through the hole from the left atrium to the right atrium. This increases the volume of blood to the right atrium, with the result that more blood flows through the lungs than would do so normally. In some cases, the hole closes without treatment. In others, the hole may need to be closed surgically.

Ventricular Septal Defect

A Ventricular Septal Defect is a hole in the dividing wall between the two lower chambers of the heart. If the hole is large, too much blood will be pumped to the lungs, leading to congestive heart failure. In most cases, the hole is small and produces no symptoms; the hole closes spontaneously as the muscular wall continues to develop after birth. If the hole is large, the hole may need to be closed surgically.

Mitral Valve Prolapse

The mitral valve is a valve of the heart, whose function is to keep the blood flowing back again when the heart contracts. In mitral valve prolapse the valve does not close as firmly as it should. It may close with a faint click, or may permit a tiny amount of blood to leak through, producing a heart murmur. Mitral valve prolapse is generally the most benign of the various types of heart murmurs and is probably genetic in origin.

Heart symptoms which may be caused by CHD

Arrhythmia

Disorders of the regular rhythmic beating of the heart is one of the most common disorders seen in the practice of cardiology. Irregular rhythms can be minor annoyances or may indicate a serious problem.

Ventricular Ectopic Beats

Every now and again there will be an extra beat from the heart. These occur in almost everyone at times and although they are rarely a problem they should be investigated if frequent.

Heart Failure

The heart must continue to pump with adequate force to pump the blood that the body needs. If it is unable to do so, the heart muscle weakens and the heart enlarges, leading to heart failure. Severe

heart failure symptoms are relatively uncommon. Where heart failure occurs, it requires specific medical treatment.

Heart block

In heart block, the heart beats more slowly than normal. In some cases, treatment is not necessary because the condition is usually temporary. In persistent cases heart block requires specific treatment.

How is Congenital Heart Disease Diagnosed?

Doctors usually diagnose congenital heart disease by scans during pregnancy or when they hear a heart murmur at birth. Occasionally children with less severe defects are not diagnosed until they are older. It will be followed by referral to a paediatric cardiologist who will take a family and medical history and do a physical examination and arrange a cardiac echo and other tests.

Symptoms of Congenital Heart Defect

The most common signs of congenital heart disease are:

Bluish tint to the skin, lips and fingernails ('blue baby')

Heart murmur

Rapid breathing

Shortness of breath

Poor feeding

Poor weight gain in infants

Delayed growth

Paleness

Tiring easily during exercise or activity (older children)

Infants and children with congenital heart disease may show differing degrees of these features depending on the severity of the defect.

Surgical & Mechanical Treatments

Diuretics treat the build-up of fluid in the heart and body.

ACE inhibitors (Angiotensin-converting Enzyme Inhibitors).

High blood pressure medicines that decrease the work the heart has to do and may help to enable the heart and blood vessels to work more efficiently.

Beta-blockers slow the heart rate and lower blood pressure to decrease the workload on the heart. These may be used to reduce thickening of the heart.

Anti-arrhythmic drugs treat the disturbances of heart rhythm.

Balloon Valvotomy

A standard treatment for pulmonary stenosis and the commonest form of treatment. A balloon on the tip of a catheter is positioned over the narrowed valve and inflated to stretch open the valve. It is usually very successful and carried out without complications. Occasionally if the valve is very thickened a direct surgical approach is needed.

Surgical Myectomy

Removal of part of the heart muscle to relieve the obstruction to blood flow in hypertrophic cardiomyopathy.

Heart Transplantation

Used for very severe heart failure. The child's heart is replaced with a healthy heart from a donor.

Medical Care

It is important for your child to have regular, ongoing medical care from the child's heart specialist or paediatrician and to administer prescribed medications according to their advice.

Glossary of terms

Atria (singular atrium)

The two upper chambers of the heart

Cardiomyopathy

Disease of the heart muscle

Cardiovascular

Relating to the heart and its blood vessels

Congenital

Present at birth

Familial

The term used to describe a disease or syndrome, which affects several members of the same family.

Fibrillation

Disturbance of the heart's normal electrical signals, causing abnormal heart rhythm

Hypertrophic

Overdeveloped or thickened

Prolapse

To slip down or fall out of place

Pulmonary Valve

The valve situated at the opening of the pulmonary artery into the right ventricle, through which blood is transported to the lungs

Septal/septum

Relating to a partition or division

Sporadic

Scattered; occurring in isolating cases

Stenosis

Narrowing

Syndrome

The term applied to a group of symptoms or features, occurring together regularly enough to constitute a disorder to which a particular name is given

Tachycardia

Abnormally rapid heartbeat

Ventricle

The two lower chambers of the heart