**The Libyan International Medical University**

**Faculty of Basic Medical Science**

# Congenital heart defect of Noonan Syndrome

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## Abstract :

Noonan syndrome is a common genetic disorder characterized clear by facial anomalies, congenital heart defects, chest deformities, undescended testes, short stature, and broad neck. The phenotypic of Noonan syndrome will be extremely variable, with a few affected subjects showing just minor features of typically the syndrome. Cardiac malformations will be also heterogeneous. Pulmonary stenosis, with or without dysplastic pulmonary valve and hypertrophic cardiomyopathy, most popular congenital heart defects discovered in Noonan syndrome.

Nevertheless, atrial septal defect, atrioventricular septal defect, left-sided obstructive lesions, tetralogy of Fallot and patent ductus arteriosus have also been described. Autosomal dominant inheritance features have been documented in some family members, although many cases seem to be sporadic.

The diagnosis of Noonan syndrome clinical or a DNA test for mutation analysis be carried out in blood.

## Introduction：

Noonan syndrome is autosomal dominant, approximately 50% of cases the disease is caused by mutations in the PTPN11 ( protein-phosphate non-receptor type 11) gene on chromosome12, which one is the most common hereditary diseases associated with the congenital heart defect, being second for the frequency of Down syndrome. The incidence of NS is estimated to be between 1:1000 and 1:2500 live births.

The syndrome was first recognized as a clinical entity in the sixties by Noonan and Ehmke. when they described several patients with pulmonary stenosis associated with characteristic facial anomalies, short stature, webbed neck, chest deformity, and undescended testes.

Pulmonary stenosis with or without dysplastic pulmonary valve and hypertrophic cardiomyopathy are the cardiac defects reported in Noonan syndrome, atrial septal defect, atrioventricular septal defect, left-sided obstructive lesions, tetralogy of Fallot and patent ductus arteriosus have also been described. NS may be is a clinical

diagnosis that difficult, particularly in adulthood. There is great variability in and the phenotype becomes less clear with increased age. Several scoring systems have been devised to help the diagnostic process. The most recent scoring system was developed in 1994.

Noonan syndrome affects equally to both males and females.

investigation of congenital heart disease of noonan syndrome, understanding places of injury in the heart.

## Methods :

Scientists examined 190 patients with Noonan syndrome. cardiac defect disease was seen in 84.2% (n=160), was revealed for pulmonary valve stenosis, atrioventricular septal defect, coarctation of the aorta, hypertrophic cardiomyopathy, atrial septal defect, left atrioventricular valve anomalies, ventricular septal defect, tetralogy of Fallot, patent ductus arteriosus, pulmonary atresia/intact ventricular septum, partial anomalous pulmonary venous return, and dilated ascending aorta.

## Results :

The proportion of incidence associated with several types of cardiac problems is shown in the table. Pulmonary stenosis and hypertrophic cardiomyopathy are usually generally the most popular congenital heart defects discovered in Noonan syndrome.

| Cardiac defect | Affected individuals | % |
|---|---|---|
| Pulmonary valve stenosis | 64/160 | 40% |
| Atrioventricular septal defect | 22/160 | 13.8% |
| Coarctation of the aorta | 20/160 | 12.5% |
| Hypertrophic cardiomyopathy | 14/160 | 8.8% |
| Atrial septal defect | 13/160 | 8.1% |
| Left atrioventricular valve anomalies | 7/160 | 4.4% |
| Ventricular septal defect | 7/160 | 4.4% |
| Tetralogy of Fallot | 6/160 | 3.8% |
| Patent ductus arteriosus | 2/160 | 1.3% |
| Pulmonary atresia/intact ventricular septum | 2/160 | 1.3% |
| Partial anomalous pulmonary venous return | 2/160 | 1.3% |
| Dilated ascending aorta | 1/160 | 0.6% |

## Discussion :

Pulmonary stenosis is frequently related to a thickened and even the dysplastic valve. most cases hard to gain a sufficient outcome using transcatheter the balloon dilatation of dysplastic valves, so surgery intervention will be likely to be capable to be needed.

Hypertrophic cardiomyopathy includes mostly the ventricular septum as asymmetric septal hypertrophy, but could also affect the ventricular free walls. Left ventricular outflow tract clogging may at times be produced.

Atrial septal defect, patent ductus arteriosus and tetralogy of Fallot are also identified in Noonan syndrome. Through the experience, left sided obstructive lesions and atrioventricular septal defects are also regularly diagnosed.

Left-sided anatomic blockage can occur at the particular valvular or supra valvular level, inside the sub-aortic placement as a result  left atrioventricular valve malformation, or as coarctation of the aortic.

The particular subgroup of patients along with Noonan syndrome and aortic coarctation demonstrates male variety, and physical manifestations overlapping with through Turner syndrome, so that will the involvement of putative lymphogenic genes located upon sex chromosomes continues to be recommended in these patients.

The atrioventricular septal malformations of subjects with Noonan disorder are most often as possible related (25% of the cases ) with sub-aortic stenosis.

The structural malformation in sub-aortic stenosis includes accessory fibrous tissue or abnormal insertion of the left atrioventricular valve and abnormal papillary muscle of the left ventricle.

Inquisitively, the abnormalities of the left atrioventricular valve leaflets and the subvalvular apparatus in patients with the atrioventricular septal defect are like those announced in patients with hypertrophic cardiomyopathy.

The particular pathogenesis of cardiac problems in Noonan syndrome has become attributed to a problem of cardiac jelly and extracellular matrix, in a few cases, as well as the same mechanism is probably active in the pathogenesis of the atrioventricular septal defect.

In addition, the development of leaflets and papillary muscles of the left atrioventricular valve associated with morphogenesis from the left ventricular outflow tract and atrioventricular and inter-ventricular septations.

Thereafter, in a few patients with Noonan syndrome, typically the developmental mechanism of the left ventricular myocardium and left The atrioventricular valve may be changing.

Uncommon cardiac defects occurring within Noonan syndrome are pulmonary atresia with an undamaged ventricular septum, which need to be considered within the spectrum of pulmonary valve stenosis with dysplasia from the leaflets, and dilated ascending aorta.

## Conclusion :

Pulmonary stenosis and hypertrophic cardiomyopathy are generally the most common congenital heart defects in Noonan syndrome. Patients with NS have a distinct spectrum of cardiac phenotypes that may have a natural history and response to therapy atypical to that normally seen in non-syndromic heart disease.

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