

### Cardiac involvement in Noonan syndrome: our experience

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#### Introduction:

□ Noonan syndrome (NS) is an autosomal dominant disorder with a high incidence of cardiovascular disease. PTPN11, RAF1 and SOS1 are some of the mutations that correlate most with cardiac pathology. Pulmonary valve stenosis (PVS) and hypertrophic cardiomyopathy (HCM) are the most frequently associated. HCM can represent a risk factor for arrhythmias or sudden death. Our objective is to study the prevalence of heart disease in our patients with NS.

#### Methods:

We reviewed the patients diagnosed with NS at our center in the last 30 years and analyzed the cardiac involvement data collected in the electronic medical record.

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#### Results: □

We obtained a sample of 41 subjects, of which the majority were male (56,1%). Only 9,7% of the subjects did not have heart disease. Among the congenital heart defects detected, 56,1% were PVS, followed by HCM 24,4%, 50% with biventricular hypertrophy). Other pathologies detected were: atrial septal defect 'ostium secundum' type (17%) and atrioventricular septal defect (4,9%), aortic coarctation (4,9%), dysplastic mitral valve (4,9%), idiopathic dilatation of main pulmonary artery and branches (7,3%). Less frequent (2,4% each) were: pulmonary atresia, tetralogy of Fallot, pulmonary arterial hypertension (PAH) and ductus. Regarding the need for some type of interventionism: 14 subjects (34,1%) required cardiac surgery and 8 (19,5%) underwent catheterization for the ones with PVS. Four patients were under treatment, mainly beta-blockers. Regarding electrocardiographic findings: 26,8% had a normal ECG, with the remaining presenting changes in the axis, with a greater percentage of left deviation (19,5%) and left anterior fascicular block in 14,6%. Two subjects had arrhythmias which consisted of frequent premature atrial beats. PTPN11 was the most frequent mutation (19,5% of all records) with PVS as associated heart disease, followed by RAF1 that was associated more with HCM. There were two deaths secondary to respiratory infection.

#### Conclusions:

Our findings are quite similar to what is described in the literature. PVS remains the most frequent finding followed by HCM. These patients also have a very typical electrocardiographic pattern and a high rate of interventionism. None ventricular arrhythmias nor sudden death were documented.