CARDDAC 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.

AIM: to study the prevalence of heart disease in our patients with NS

RESULTS

41 subjects
58% males

HEART DISEASE

41 subjects
58% males

NO 9.8%

YES 88.2%

Regarding the need for some type of interventionism:
• 14 subjects (34%) required cardiac surgery.
• 8 (19.5%) underwent catheterization for the ones with PVS.
• Four patients were under treatment, mainly beta-blockers.

ELECTROCARDIOGRAPHIC FINDINGS
• 21.5% normal ECG.
• Changes in the axis, with a greater percentage of left deviation (35.7%).
• Left anterior fascicular block in 25%.
• Two subjects had frequent premature atrial beats.

GENETIC FINDINGS
PTPN11 was the most frequent mutation (57.1% of all records) with PVS as associated heart disease, followed by RAF1 that was associated more with HCM.

CONCLUSIONS
• 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.