

**Noonan's syndrome: Cardiac pathology, outcomes and practice in a single institution**

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Introduction: Noonan's syndrome is the commonest genetic syndrome associated with CHD after Trisomy 21, often requiring cardiac intervention. We reviewed our practice in the last 13 years with regards to the type and rate of interventions as well as arrangements for follow-up and transition to adult services.

Methods: This was a retrospective review of patients' electronic records referred with Noonan's phenotype.

Results: Out of 128 patients, 105 patients were suitable for analysis (complete records). Only 17 (16%) patients had no cardiac abnormalities, of which 13 were discharged, some as early as at 3 years of age. There were 139 abnormalities in 87 patients, with valvar pulmonary stenosis the commonest type (64% of all patients). Other abnormalities included supralvalvar pulmonary stenosis (9.5%), branch pulmonary stenosis (4%) and subpulmonary stenosis (2%). There were 15 cases of ventricular hypertrophy (14%) of which 4 were of the obstructive type (4%). ASDs were identified in 17%, aortic valve abnormalities in 8.5%, 2.8% had mitral valve abnormalities, PDA in 1% and VSD in 4.7%. There was one case with a partial AVSD, one with ccTGA and one with aortic coarctation.

A total of 57 interventions were undertaken in 47 patients (overall intervention 54%). 56% of patients with valvar PS required intervention (50% had a transcatheter procedure and 6% surgery). One quarter of those patients needed re-intervention. There were 9 more interventions for LVOTO relief, ASD closure, aortic coarctation and AVSD repair.

There were 6 deaths (5.7%). The cardiac findings in these patients were ventricular hypertrophy, dysplastic aortic valve and multi-level pulmonary stenosis.

Most of our patients with a known cardiac abnormality remain under follow-up by a paediatric cardiologist. At 16-18 years their care is transferred to the cardiology service in peripheral hospitals (17% patients) or the Grown Up Congenital Heart disease programme (12%).

Conclusions: In patients with Noonan's syndrome there is a high rate of cardiac abnormalities and interventions, therefore suitable follow-up arrangements during childhood and adulthood should be in place.