NOONAN SYNDROME

(Case Report)

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Introduction

Noonan syndrome is an autosomal dominant cardiofacial syndrome occurring in both sexes. It has somatic features similar to Turner syndrome but the karyotype is normal. Its incidence has been estimated at 1 in 1000 to 2500 live births. The congenital heart diseases commonly associated with it are pulmonary valve dysplasia, atrial septal defect and hypertrophic cardiomyopathy [1]. Since the first report of Noonan syndrome by Jacqueline Noonan in 1963 [2] many cases have been reported in western literature, but only a limited number have been reported from India [3-5]. We describe here an adult male of Noonan syndrome who presented with features of pulmonary stenosis.

Case Report

An eighteen years old male presented to our hospital with complaints of breathlessness on accustomed exertion of two months duration. He denied having chest pain, palpitation, paroxysmal nocturnal dyspnoea, syncope, cough or swelling of feet. There was no past history of migratory polyarthritides. He was born from a non consanguineous marriage and there was no history of exposure to teratogens during antenatal period. The developmental milestones had been normal except for poor performance at school. The family history was not contributory.

Physical examination revealed short stature (149 Cm), low set ears, ptosis, antimongoloid palpebral slant, high arched palate and pectus excavatum. Pulse, blood pressure and jugular venous pressure were normal. There was no pallor, cyanosis, clubbing or pedal edema. Apex beat was palpable in the left fourth intercostal space in the midclavicular line, normal in the character.

Grade-I/III left parasternal heave was present. A systolic thrill was palpable in the left upper sternal border. The first heart sound was normal but the second heart sound was not audible. A loud crescendo decrescendo murmur (Grade-V/VI) was present in the pulmonary area. It radiated towards left clavicle and increased on inspiration. There was no ejection click or fourth heart sound.

Examination of other systems was normal. Hemogram and urinalysis were normal. The electrocardiogram (Fig-2) showed normal sinus rhythm, extreme right axis deviation, 'P' pulmonale in II and VI, R=S in VI, ST segment depression and T wave inversion in VI V2 III avF and QS complexes in V4-V6. Chest

radiograph was normal. Echocardiography (Fig-3) and color flow imaging (Fig-4) showed a thickened pulmonary valve, dilated and hypertrophied right ventricle and dilated right atrium, interventricular septum was intact and pushed to left side. There was turbulent flow in the pulmonary artery. Other chambers and valves were normal.

Discussion

Cardinal features of Noonan syndrome are short stature, webbing of neck, sternal deformity, cubitus valgus, ptosis, low set ears, antimongoloid palpebral slant, mental retardation, cryptorchidism, lymphedema, hypogonadism and congenital heart disease. The diagnostic criteria [6] include four or more cardinal features with a normal karyotype. Our patient had

Fig. 1 : Note the antimongoloid slant, ptosis and low set ears

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Fig. 2 : Electrocardiogram showing right axis deviation, 'P' pulmonale and right ventricular strain

Fig. 3 : 2D Echo, parasternal short axis view showing thickened pulmonary valve

short stature, facial dysmorphism, sternal deformity, mental retardation and pulmonary stenosis. The Karyotype could not be done. He appeared to be a sporadic case as other members of family were normal. Although the responsible gene has not been identified, recently a gene causing Noonan syndrome has been mapped on chromosome 12. In a large series of 151 cases [7], the mean age of presentation was 12.6 years. Dysplastic pulmonary valve with valvular pulmonary stenosis was present in 62%, hypertrophic cardiomyopathy was observed in 20%, webbed neck was present in 23% of cases, sternal deformity in 95% and mental retardation was present in 35% of cases.

The intensity and splitting of second heart sound are important points in the auscultatory evaluation of pulmonary stenosis [8]. The intensity of the pulmonary component varies from normal to inaudible as severity changes from mild to severe. In our patient it was not audible indicating severe pulmonary stenosis. However, the timing of pulmonary component is more important than intensity. The greater the obstruction,

Fig. 4 : Colour doppler shows turbulent flow in the pulmonary artery

the longer the duration of right ventricular ejection, the later is pulmonary valve closure and wider the split. the electrocardiographic changes reported vary from normal in mild cases to features of right atrial enlargement, right ventricular hypertrophy with strain and presence of deep waves in precordial leads [1,8].

The disorder has to be differentiated from other cardiofacial syndromes i.e. Turner syndrome (45X0), Leopard syndrome (pigmentation), Williams syndrome (supravalvular AS), Fetal alcohol syndrome and Aarskog syndrome. Management includes prophylaxis against infective endocarditis and correction of underlying cardiac defect if possible. Our patient has been advised balloon valvuloplasty.

REFERENCES

1. Pyeritz RE. Genetics and cardiovascular disease. In: Braunwald E, ed. Heart disease; A textbook of cardiovascular medicine 4th ed. Philadelphia. W.B. Saunders Co. 1992; 1634-5.
2. Noonan JA, Ehmke DA. Associated non cardiac malformation in children with congenital heart disease. J Pediatr 1963;63:468-70.
3. Unnithan RR, Bahuleyan CG, Mathewroy VC. Noonan syndrome (case report). J Assoc Physicians India 1985;33:17-178.
4. Shah KO, Ayyer KH, Nair N. Noonan Syndrome case report. Ind Heart J 1969;31: 182-6 5.
5. Yadav BS, Indurkar M, Bisraya BN. Normal pregnancy in a case of Noonan syndrome with hypertrophic cardiomyopathy (case report). J Assoc Physicians India 1996;44:495.
6. Grumach MM, Conte FA. Disorders of Sex differentiation. In Williams RH Ed;Text book of endocrinology 6th Ed. Philadelphia. WB Saunders 1981, 471-2.
7. Sharland M, Burch M, McKenna MM, paton MA. A clinical study of Noonan syndrome. Arch Dis in Childhood 1992;67:178-83.
8. Perloff JK. The clinical recognition of congenital heart disease. 4th ed. Philadelphia WB. Saunders Co. 1994; 215-9.