Isolated Pulmonary Valvular Stenosis – Noonan Syndrome – A Case Report

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Abstract
Noonan syndrome is an autosomal dominant dysmorphic characterized by hypertelorism, a downward eyeslant, and low-set posteriorly rotated ears. Other features include short stature, a short neck with webbing or redundancy of skin, cardiac anomalies, epicanthic fold, deafness, motor delay, and a bleeding diathesis. In this case report a 20 years male presented with severe pulmonary stenosis with classical skeletal abnormalities.

Key words: Autosomal dominant, pulmonary stenosis

Introduction
Noonan Syndrome (NS) is a relatively common congenital genetic condition which affects both males and females equally. It used to be referred to as the male version of Turner's syndrome; however, the genetic causes of Noonan syndrome and Turner syndrome are distinct. The principal features include congenital Heart Malformation, short stature, learning problems, indentation of the chest, impaired blood clotting, and a characteristic configuration of facial features. The syndrome is named after Dr Jacqueline Noonan. It is believed that between approximately 1 in 1,000 and 1 in 2,500 children worldwide are born with NS. It is one of the most common genetic syndromes associated with congenital heart disease, similar in frequency to Down syndrome. However, the range and severity of features can vary greatly in patients with NS. Therefore, the syndrome is not always identified at an early age. This case is diagnosed and managed in BSMMU.

Discussion
Noonan Syndrome (NS) is a relatively common congenital genetic condition which affects both males and females equally. It used to be referred to as the male version of Turner's syndrome; however, the genetic causes of Noonan syndrome and Turner syndrome are distinct. The principal features include congenital Heart Malformation, short stature, learning problems, indentation of the chest, impaired blood clotting, and a characteristic configuration of facial features. The syndrome is named after Dr Jacqueline Noonan. NS may be inherited in an autosomal dominant pattern with variable expression. A person with NS has up to a 50% chance of transmitting it to a child. The fact that an affected parent is not always identified for children with NS suggests several possibilities: a parent could carry the gene without being affected (incomplete penetrance); manifestations are variably expressed and could be so subtle as to go unrecognized (variable expressivity). A high proportion of cases represent new, sporadic mutations. Noonan syndrome is heterogeneous, comprising more than one similar condition of differing cause, some not inherited. In our case, the patient showed short stature, triangular shape in face, webbed neck, low hairline at the
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nape of the neck, widely set eyes (hypertelorism), proptosis (bulging eyes), depression of breast bone (pectus excavatum), Shiled chest and widely apart nipple, winging of the scapula, low sets ears, Deeply grooved philtrum (top lip line), Micrognathia (undersized lower jaw), High arched palate, Malalinghed teeth, articulation difficulties, Cubitus valgus, Flat feet, Clumsiness, poor coordination, motor delay, Mental retardation, learning disabilities.

His pulse 84/min, regular, symmetrically palpable all peripheral pulse, BP was 110/80 mm Hg, RR 16/min. JVP was not raised, Patient is mildly anaemic, not ecteric, cyanosis, clubbing and oedema not present. Apex beat located in Lt 5th ICS normal in character, RV heave present, P2 was absent, S1 is audible all the cardiac area & normal, pulmonar component of S2 is soft in pulmonary area. Thrill present on the pulmonary area. There is a ejection systolic murmur grade 4/6 on the pulmonar area radiates towards the Lt side of the neck. Other systemic examination reveals no abnormalities. His investigation showed serum creatine 1mg/dl, Hb-14.7gm/dl, ECG showed RAH & RVH with strain pattern. Echo 2D – M mode, colour & spectral doppler showed main pulmonary artery narrowed then poststenotic dilation of main pulmonary artery, Left pulmonary artery dilated, RA, RV dilated, mosaic flow from RV outflow to pulmonary artery during systole, Pressure gradient of PV is 146mm Hg. USG of KUB regions showed rt sided pelvis dilated and are cystic lesion (6.6 x 6.8) cm size. CXR P/A view showed RV type of hypertrophy. Cardiac catheterization RV graphy showed, RV pressure 130 mm Hg, PA – pressure 80 mm Hg and pressure gradient 50 mm Hg. Balloon valvuloplasty attempted but unsuccessful patients awaits for valvotomy at convenient time. If unsuccessful than valve replacement.

Conclusion

Noonan syndrome is a relatively common congenital genetic condition which affects both males and females equally. Despite identification of four causative genes, the diagnosis of noonan syndrome is still based on clinical features. In other words, it is made when physician feels that a patient has enough of the features to warrant the label indicating association. The patient can be tested for mutations in the PTPN11, SOS, or KARS gens, however absence of a mutation will not exclude the diagnosis as there are more as yet undiscovered genes that cause NS. The principal values of making such a diagnosis are that it guides additional medical and developmental evaluations, it excludes other possible explanations for the features, and it allows more accurate recurrence risk estimates.

Reference


