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Heart Failure as the Presenting Manifestation of Noonan Syndrome: A Case Report

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ABSTRACT

Noonan syndrome is a common non-chromosomal syndrome associated with congenital heart disease. Noonan syndrome is associated with short stature, pectus excavatum or pectus carinatum, webbing of neck, low set ears with moderate mental retardation. Here we are reporting a case of 16 yrs. old boy who presented to us with features of Noonan syndrome.

Key Words: *Noonan syndrome, Pectus excavatum, High arched palate, Cubitus valgus, Karyotyping*

INTRODUCTION

Noonan syndrome was first described by Noonan and Ehmke in 1963 as a multisystem disorder1. Noonan syndrome occurs in 1 in 2000 live births and affects both males and females2. This disorder is usually sporadic but affected siblings have been reported. We report a boy with Noonan syndrome.

CASE REPORT

A 16 year old boy with history of non-consanguineous marriage presented with breathlessness for 20 days & cough for 5 days. There was no history of fever, sore throat, chest pain & joints pain.

On examination, patient was conscious, oriented. His pulse was 100/min, regular, normovolumic with no radio–femoral delay. All the peripheral pulses were palpable. Respiration rate was 20/min. Blood pressure was 100/60 mm Hg. He had malocclusion of teeth, low posterior hair line, wide set nipples (inter nipple distance is more than 25% of total chest circumference), high arched palate (normally high arched palate is seen

as keep the patient's mouth wide open & look for palate keeping the eyes at the level of patient's upper incisor teeth, if roof of palate is not visible, it is known as high arched palate), low set ears (Normally 1/3rd of the length of pinna is seen above an imaginary horizontal line, extending from outer canthus of eye to ipsilateral pinna. If less than 1/3rd of length of pinna is seen above that line, low set ears are said to be present.), pectus excavatum and cubitus valgus. Examination of external genitalia was normal.

On general examination there was mild pallor rest general examination was normal. On abdomen examination there was no hepatosplenomegaly on cardiovascular examination he had ejection systolic murmur which was best heard in second left intercostal space, P2 was soft. Respiratory and CNS examination were unremarkable.

On investigations, Hb was 9.4 mg/dl, TLC and DLC were normal, LFT and KFT also were normal. prothrombin time, bleeding time and clotting time were normal. On ultrasound examination there was no organomegaly, both kidneys were normal in shape and size according to their age. On scrotal ultrasound both testis were normal in shape and size, electrocardiogram showed right ventricular hypertrophy. Echocardiography revealed severe valvular pulmonary stenosis.

During hospitalization patient was managed conservatively with diuretic and digoxin. Patient improved and was discharged after 10 days.

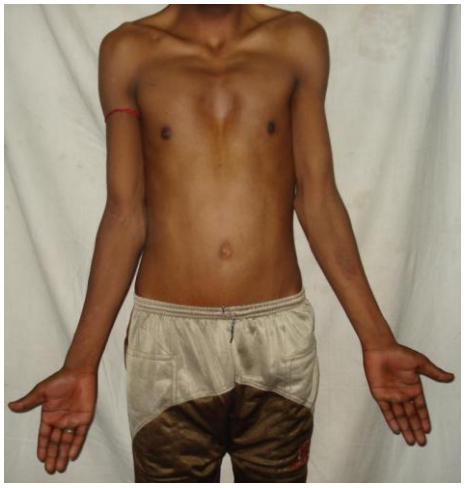

Figure 1: Pectus Excavatum with Cubitus Valgus

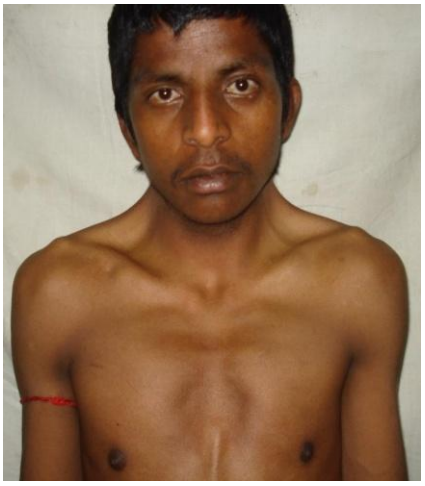

Figure 2: Short webbed neck, widely spaced nipples and Pectus Excavatum

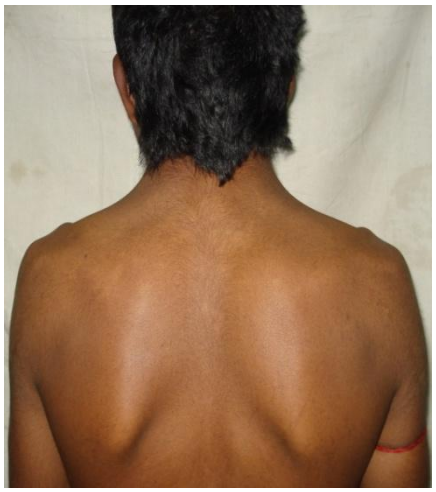

Figure 3: Low posterior hair line

DISCUSSION

Noonan syndrome is the most common non-chromosomal syndrome associated with congenital heart disease3. Noonan syndrome resembles Turner syndrome phenotypically but patient has apparently normal sex chromatin. These boys & girls have normal karyotype. Reports of male to male transmission suggest an autosomal dominance gene with variable expressivity. These patients have delayed pubertal development but there is no gonadal failure4. Common Abnormalities in Noonan syndrome are short stature, pectus carinatum or pectus excavatum, webbing of neck, cubitus valgus, hypertelorism, low set ears, downward slanted palpebral fissures and micrognathia2,4,6,8. Hernias, wide set nipples, malocclusion of teeth, clinodactyly, moderate mental retardation and sensorineural deafness can be seen2,6. Right sided congenital heart disease is commonly present. Pulmonary stenosis as a result of valve dysplasia is the common cardiac abnormality of Noonan Syndrome, Other cardiac defects may be ventricular septal defect, Patent ductus arteriosis, aortic stenosis, tetralogy of fallot, atrial septal defect, coarctation of aorta & hypertrophic obstructive cardiomyopathy (HOCM)3. About half of patients of Noonan Syndrome have mutation in the PTPN11 gene that encodes protein tyrosine phosphatase SHP2 (locus 12q24.1). A small proportion of affected individuals have a mutation in the KRAS gene9. Renal anomalies are seen in 10% patients but not clinically significant10.More than 50% of male patients have undescended testes. Hepatosplenomegaly not related to cardiac disease is seen in about 25% of cases10. Joint laxity, cervical spine fusion, bleeding diathesis, cryptorchidism, lymphoedema, follicular keratosis of face may also be seen10. Seizure disorder is seen in about 13% of the patients10.

The Phenotype of Noonan Syndrome differs from turner syndrome in the following respect: (1) Mental retardation is often present. (2) The cardiac defect is most often pulmonary valvular stenosis or ASD rather than an aortic defect. (3) Gonadal defects may be present, but normal sexual maturation usually occurs.

No diagnostic testing for Noonan syndrome is available. This syndrome is diagnosed clinically. Karyotyping may be necessary if full phenotype is not present.

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