# Noonan's Syndrome

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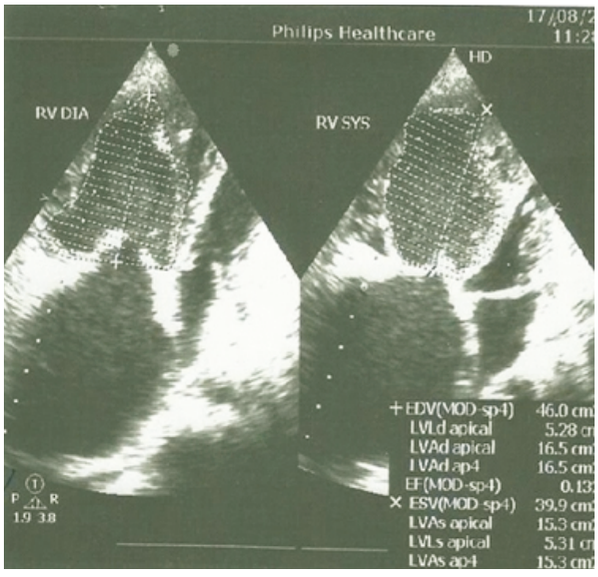


**Figure 1. 2 D-Echocardiography**

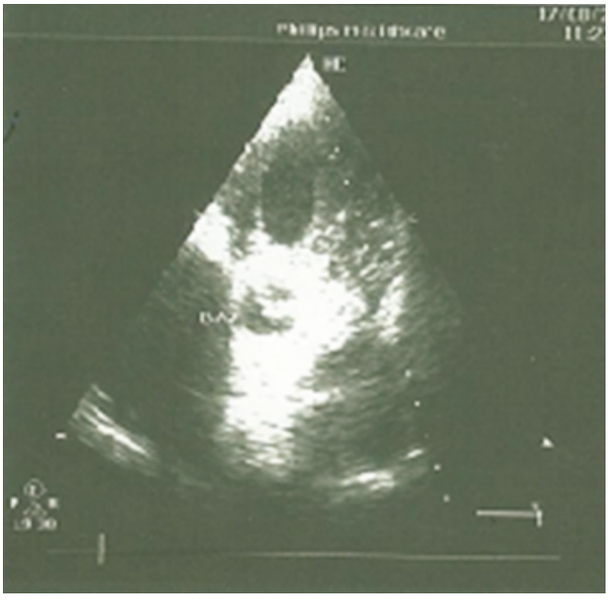

**Figure 2. 2D-Echocardiography**

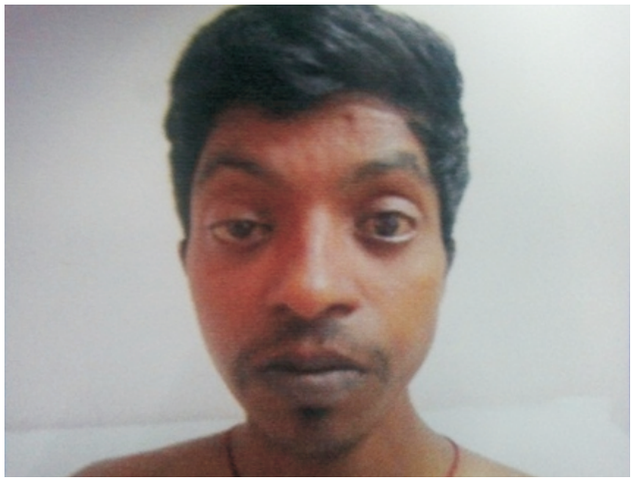

**Figure 3. Facial dysmorphism (hypertelorism, right partial ptosis,proptosis)**

A 32 year old man was evaluated for breathlessness, scrotal and limb swelling of 2 years duration. He was treated with diuretics and was on irregular medications. A working diagnosis of congestive cardiac failure was done and the cause evaluated. There was no history of diabetes mellitus/ hypertension/bronchial asthma/tuberculosis. There was no history of consanguineous marriage.

Dysmorphic facial features were hypertelorism, right partial ptosis, proptosis, high arched palate, epicanthic fold, low set ears, low hair line, scoliosis, undescended testis, webbed neck, pectus excavatum, short metacarpal bones, hallux valgus deformity [1,2].

Findings of cardiovascular system examination were raised jugular venous pressure, presence of epigastric pulsations and parasternal heave, ejection systolic murmur at pulmonary area and aortic area, pan systolic murmur at tricuspid area. Bilateral normal vesicular breath sounds heard. Chest X-ray showed cardiomegaly, prominent aortic knuckle and uplifting of apex with scoliosis. ECG interpretation was marked right axis deviation, right bundle branch block pattern with right ventricular strain. Echocardigraphy revealed congenital heart disease with moderate pulmonary

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artery stenosis with severe tricuspid regurgitation, dilated right atrium and right ventricle and bicuspid aortic valves with mild aortic stenosis, small left ventricular cavity with left ventricular ejection fraction of 50 % [3]. There was right ventricle dysfunction with its ejection fraction of 12%.

The patient was diagnosed as Noonan's syndrome in congestive cardiac failure requiring diuretic therapy to improve his symptoms and quality of life. Karyotyping report is awaited.

Mortality in case of Noonan's syndrome depends on the severity of congenital heart disease [4]. Adults at 32 years, surviving with Noonan's syndrome is rare. Additional feature of bicuspid valves with aortic stenosis in our case further adds to its rarity.

**References**

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