Neck Webbing in a Male Patient A Case Report

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Case Description

In the month of November, 2013, there came a male patient with the complaints of frequent vomiting and body ache. His age was 35 years. When the patient was exposed for clinical examination, there were prominent skin folds on either side of the neck. It was our first experience to see a male person with webbing of neck. On examination, his height was 5 feet and 4 inches and was of normal intelligence. The pulse rate was 76 per minute, and blood pressure was 184/82 mm Hg. On examination of chest, no significant abnormality or deformity was observed. However, there was prominent grade III systolic murmur in the 2nd aortic area. Respiratory system was normal and on abdominal examination, no organomegally was found. The patient was asked as usual to undress for genital examination, the size of testes were smaller and firm, not compatible to his age. Right testis was higher as compared to left and hernial orifices were normal. Despite of one of his presenting complaints, no musculoskeletal deformity was observed and also no abnormal pigmentation of skin was seen.

The clinical findings prompted us to diagnose the patient as a case of Noonan syndrome which is a counter part of Turner's syndrome in females. We asked the patient to get admitted so that the detailed clinical examination and necessary investigation including karyotyping be carried out. However, the patient left against medical advice and did not report to OPD for further discussion and councelling.

61

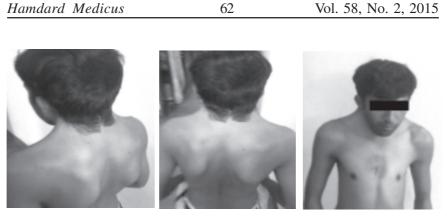


Fig. 1

Fig. 2

Fig. 3

Discussion

Noonan syndrome is a relatively uncommon genetic anomaly affecting both sexes. It has an autosomal dominant trait. Its vertical transmission is 50% to his offspring^{1, 2}. The severity of the clinical features are variable from patient to patient and among family members. However, the common clinical signs and symptoms are congenital heart diseases present in almost 2/3 of patients with 50% suffering from pulmonary valvular stenosis and have septal defects 10%^{3, 4}. In the early stages cyanosis is not present until reversal of shunt takes place. We are reporting this case, because it is relatively an uncommon chromosomal anomaly secondly clinical examination is necessary because such type of malformations if not observed minutely might go unnoticed. Once syndrome is identified and confirmed by karyotyping genetic counceling should be provided its transmission to the biological offspring in future can be prevented by genetic engineering.

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63

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