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Case Report

A VERY INFREQUENT ASSOCIATION OF WILLIAM-BEURAN SYNDROME AND TETRALOGY OF FALLOT

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ABSTRACT

WB-S Autosomal Dominant Disorder is the most common genetic disorder. We report a case of 20 year old with infrequent association of WBS and TOF. Clinical examination and ECHO confirmed TOF, WB-S was suspected based on the clinical signs used in the scoring system of WB-S which were described by AAP(2001)[1], FISH study was performed in this patient because of having more than 3 clinical signs of WB-S and FISH study showed 7q11.23 deletion and remains the gold standard laboratory investigation for WB-S.

KEYWORDS: Tetralogy of Fallot, William Beuren Syndrome, Clinical Diagnosis, Fluroscence In Situ Hybridisation.

INTRODUCTION

William Beuran Syndrome is a complex neurodevelopmental disorder comprising cardiovascular disorder, mental retardation with peculiar cognitive profile, dysmorphic facies and body features with an estimated incidence 1 in 7500 to 1 in 20000 live births [2]. Around 75% - 80% of all patients with WB-S have some congenital heart defects. Facial features include prominent forehead, deep set eyes, low set ears, short nose, flattened nasal septum, full cheeks and wide mouth. Cognitive profile in WB-S ranges from mild to severe intellectual disability [3], language is often delayed. WB-S patients starts walking late at around 2 to 3 years of age [4]. Behavioural issue in WB-S include anxiety, attention problems, and friendly behaviour.

CASE REPORT

A 20 year old presented with complaints of breathlessness on exertion and cough with expectoration. He was born to a non-consanguineous parents with uneventful pregnancy and delivery. A delay in his developmental milestones, started walking at around 4 to 5 years of age with delayed speech and language disability with mild to moderate mental retardation. Past history revealed that patient had repeated respiratory infection since childhood for which they consulted doctor and diagnosed to have congenital heart defect at the age of 6 years. No significant family history. On physical examination he was found to have Elfin facies and similar characters which are found in patients with WB-S [figure 1]. He also has widely spaced nipples, supernumerary nipples [figure 2] Grade 3 clubbing [figure3], Pansystolic murmur of grade IV/VI on auscultation. Routine blood investigation, renal function test, liver function test, electrolytes, serum calcium all were within normal limits. ECG showed Right ventricular hypertrophy. Chest x-ray showed boot shaped heart, right aortic arch, double density right border with slightly pulmonary oligemia [figure4]. 2D Echocardiography showed Cyanotic Congenital Heart Disease, TOF consisting of VSD, 50% overriding of aorta, Right ventricular hypertrophy, Hypoplastic PA's [figure5]. Despite of speech therapy there is no improvement. FISH study confirms the diagnosis.

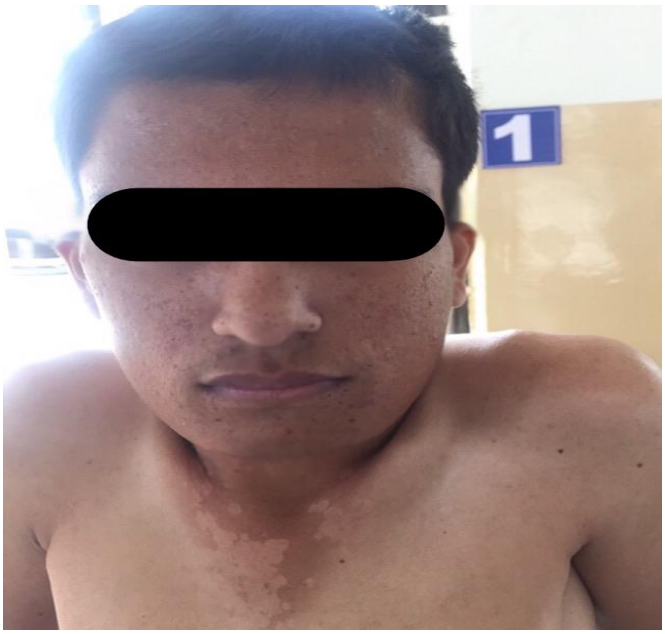

Fig 1: Typical facial appearance: low set ears, wide mouth, flat nasal bridge, wide forehead.

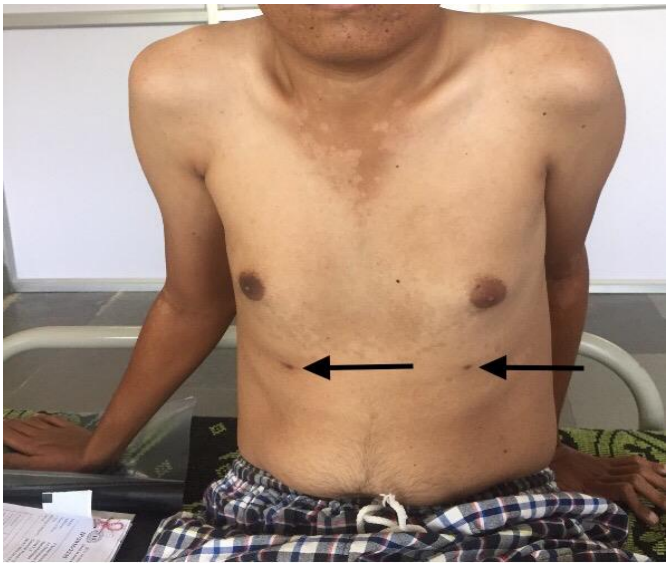

Fig 2: widely spaced nipples, supernumerary nipples

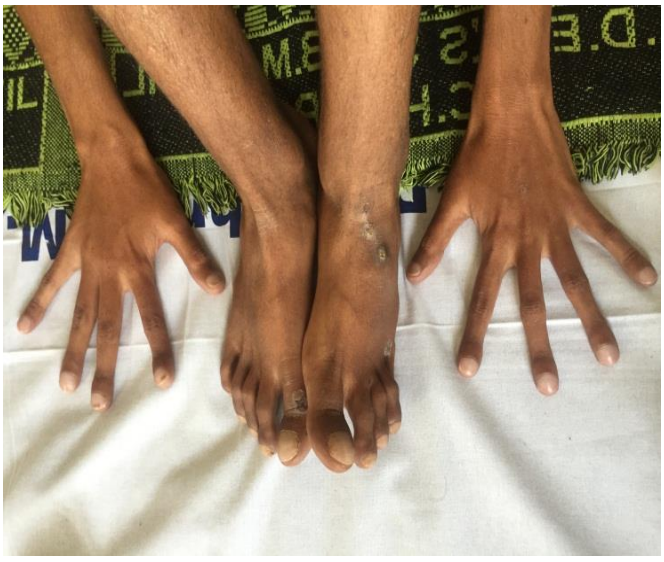

Fig 3: Grade 3 clubbing

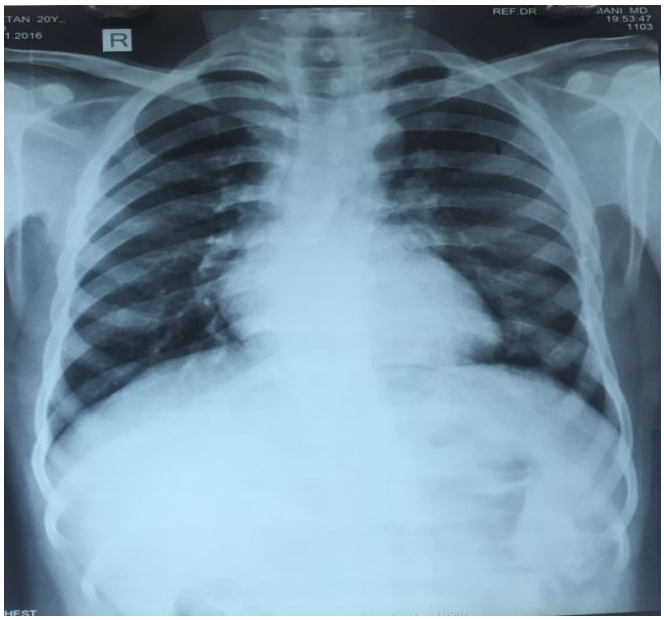

Fig 4: boot shaped heart, right aortic arch, double density right border with slightly pulmonary oligemia

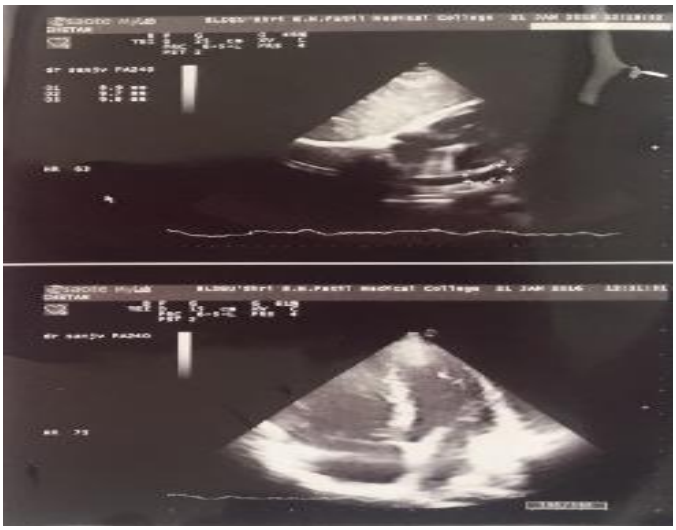

Fig. 5: VSD, 50% overriding of aorta, Right ventricular hypertrophy

DISCUSSION

Dr J C P Williams in 1961 and Beuran in 1964 was first described William Beuran syndrome. We here report a case with overall there are very few case of WB-S and TOF in literature [5,6], VSD are present in 4% to 9% of all patients with WBS. Other cardiovascular anamolies found in association with WB-S are SVAS which is most common, Patent ductus arteriosus, stenosis of outlying arteries (renal, cerebral, carotid, coronary, brachiocephalic, subclavian and mescenteric), Coarctation of aorta, mitral valve incompetence. The percentage of WBS cases were associated with Congenital Heart Defects are 80% [6]. The facial features are recognisable around 4-5 months of age and becomes typical during childhood and puberty [7]. We here first suspected as case of WBS based on clinical signs which were used in scoring system for diagnosis of WBS, score of < 3 were classified as “uncertain” and score of 4-10 were classified as

"classic" [4] and considered for FISH study showed micro-deletion of chromosome 7 (7q11.23).Despite of typical facial features and heart murmur diagnosis was made at age of 20 years, delay in diagnosis may be because of facial dysmorphisms which evolves over age and may be discrete in infancy and early childhood.

CONCLUSION

Mortality and Morbidity rates are high because of cardiovascular defects and their sequele and can prevent sudden cardiac deaths. So early diagnosis of the syndrome is important to initiate the treatment and prognosis can be improved by early recognition and management.

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