Short Communication

TETRALOGY OF FALLOT IN DOWN SYNDROME (TRISOMY 21) – AN UNCOMMON ASSOCIATION

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ABSTRACT

Down Syndrome (trisomy 21) is the common disorder among chromosomal anomalies. This is frequently associated with congenital acyanotic heart disease. Tetralogy of fallot is an uncommon event in the trisomy 21. Tetralogy of fallot presents with cyanosis usually in the later part of infancy, but cyanosis is present since birth if Tetralogy of Fallot is accompanied with Down Syndrome.

KEY WORDS: Tetralogy of Fallot, Down Syndrome.

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INTRODUCTION

In Northern Ireland, Down Syndrome (trisomy 21) is the commonest cause of mental handicap.[1] It's association with congenital heart disease is well Known, the reported incidence being 40%-60%.[2-4] This congenital heart disease contributes significantly to the morbidity and mortality of children with Down Syndrome, who may develop congestive heart failure, pulmonary vascular disease, failure to thrive or pneumonia. Majority of the congenital heart disease in Down Syndrome is acyanotic heart diseases. Tetralogy of Fallot was not observed in any child with Down Syndorme.[5] We encountered one patient who suffered from Down Syndrome with Tetralogy of Fallot which is a rare association and wish to share this experience.

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CASE REPORT

A boy of eleven months was admitted in a medical college hospital with the complaints of bluish discoloration of lip and finger since birth and low grade fever, cough for seven days. Bluish discoloration aggravates during crying. He was born to an elderly mother and was completely immunized. There was no such illness in the family. On examination the child was cyanosed, Heart rate 130/m, weight 7.5kg. He had got mongoloid face with flat occiput, depressed nasal bridge, upward slanting of eyes, medial epicanthic fold. There was gap between the first and second toes with clinodactyly. On examination of the precordium there was left parasternal heave, pansystolic murmur was present in the left

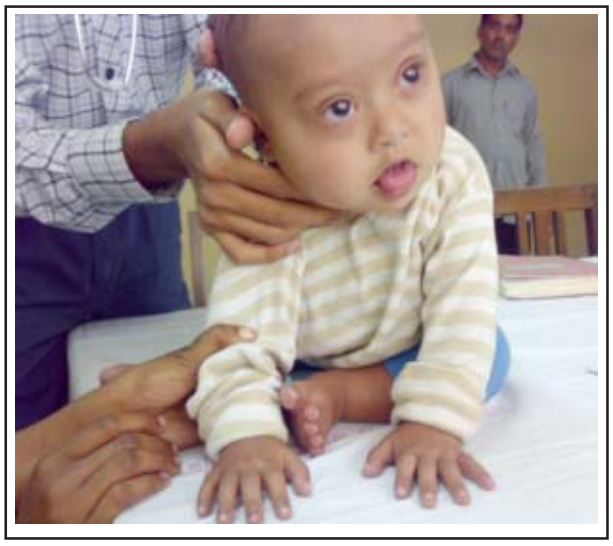

Figure-1: Patient with Down Syndrome.

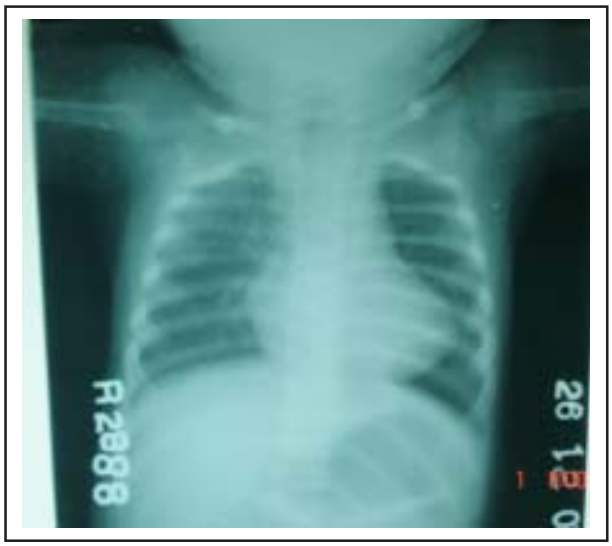

Figure-2: X-ray chest showing: boot shaped heart.

lower sternal border. There was also motor developmental delay.

The boy was clinically diagnosed with Congenital Cyanotic Heart Disease with Down Syndrome. On investigation his Hb was 78% TLC- 14,700/cum, Neutrophil- 82%, X-ray chest had the feature of boot shaped cardiac shadow. ECG showed right ventricular hypertrophy. Karyotyping showed trisomy 21. Tetralogy of Fallot was detected by Echocardiogram. Finally the child was diagnosed as Down Syndrome with Tetralogy of Fallot.

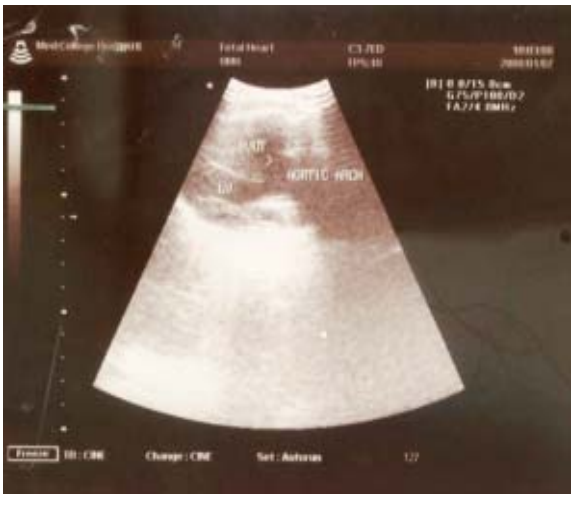

Figure-3: Echo cardiogram showing Tetralogy of Fallot

DISCUSSION

The most common abnormalities of chromosome number (aneuploidy) are trisomies. The most frequent trisomy in human is trisomy 21 or Down Syndrom.[6] The incidence of Down Syndrome is approximately 1 in 700 of all live birth in all population. The incidence varies with the age of the mothers. The incidence for mothers aged 25 years is 1 in 1400 and increases to reach an incidence of 1 in 46 for mother aged 45 years.[7] Congenital heart disease occurs in between 40%-60% of infants with Down Syndrome. Atrioventricular canal and ventricular septal defects are the commonest type of cardiac lesion seen.[7] In a study done by Tubman TRJ et al on 69 babies suffering from Down Syndrome, 34 had congenital heart disease. Among these 34 babies a atrioventricular septal defect, secundum atrial septal defects, a solitary patent ductus arteriosus, isolated ventricular septal defects and complications of heart defects was observed in the order of frequency.[5] None had the observation of Tetralogy of Fallot in the babies with Down Syndrome. In Tetralogy of Fallot often cyanosis is not present at birth, but increasing hypertrophy of the right ventricular infundibulum and patient's growth, cyanosis occur later in the first year of life. But in our case cyanosis was present since birth. This might be due to

increased hypertrophy of the right ventricular infundibulum in patient of Tetralogy of Fallot in child with Down Syndrome just at birth.

Although Down Syndrome is mostly associated with congenital acyanotic heart disease but Tetralogy of Fallot might be encountered. Patient of Tetralogy of Fallot with Down Syndrome might have early presentation of cyanosis although isolated Tetralogy of Fallot often have cyanosis later in the first year of life.

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