

## **An Uncommon Congenital Cardiovascular Malformation with Turner Syndrome – A Case Report**

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**KEY WORDS** Congenital cardiovascular malformations; Chromosome abnormality; Transposition of great arteries; Turner syndrome

**ABSTRACT** The association of Turner syndrome (TS) with congenital cardio vascular malformations is well established. Bicuspid aortic valve and coarctation of aorta have been associated most commonly with Turner syndrome. There are also rare reports of atrial septal defect and aortic aneurysm with Turner syndrome. We report a uncommon case of transposition of great arteries (TGA) associated with turner syndrome (45, X, 16qh+).

### **INTRODUCTION**

Turner Syndrome, also known as Monosomy X (or Gonadal dysgenesis or 'Ullrich – Turner Syndrome), is a rare genetic disorder and occurs in 1 in 2500 female births. The karyotype can be either due to monosomy X (45,X), mosaicism (45,X/46,XX) or a structural abnormality of the X chromosome (Zinn et al. 1993; Wolff et al. 2000). Few studies suggest that some Turner patients have Y chromosomal material in addition to the one X Chromosome (Tsuchiya et al. 1995; Quilter et al. 1998). The syndrome is characterised by short stature, ovarian failure and presence of other clinical features such as webbed neck, congenital cardiovascular malformations (CCVM), renal abnormalities, cubitus valgus and neurocognitive deficits. The CCVM ranges 17 to 47% among Turner syndrome cases and have an increased risk of mortality. Here we first report a child with Turner syndrome who had transposition of great arteries as cardio vascular malformation.

### **MATERIALS AND METHODS**

Informed consent was obtained from parents in accordance with the ethics committee of the 'Institute of Cardio Vascular Diseases'. Transthoracic M-mode, two dimensional and pulsed Doppler echocardiography were performed with a Sonos 5500 system. On the day of echocardiographic examination, 2ml of venous

blood was collected in a sodium heparin precoated syringe. Phytohemagglutinin stimulated lymphocyte cultures were set up following the method of Hungerford (1965). GTG banding method of Seabright (1971) was employed. Photograph of metaphase plate was taken under an oil immersion lens (100x) using TMAX 100 film in Olympus microscope. Chromosomal anomalies were designated according to the standard nomenclature (ISCN 1995).

### **RESULTS**

The female patient YAR was the third child born to 37 year old mother and 41 year old father. The parents are healthy and had consanguineous (First degree) marriage. Other two sibs were normal and there was no family history of congenital anomalies. She was born at full term of an uncomplicated pregnancy. The birth weight was 2850g, had a length of 41 cm and the head circumference was 34 cm.

The health problem started soon after the birth (asphyxia). She was admitted in the neonatal ward for 15 days and was diagnosed to have a complex cardiac defect. General examination of the proband revealed the following features: dolichocephaly (Fig. 1a), flat face, drooping eyelids (ptosis), sunset eyes (Fig. 1b), low set ears, webbing of the neck (Fig. 1c), cutis laxa, swelling of the hands and feet, short fourth metacarpels, broad chest and cardiac murmur. Laboratory

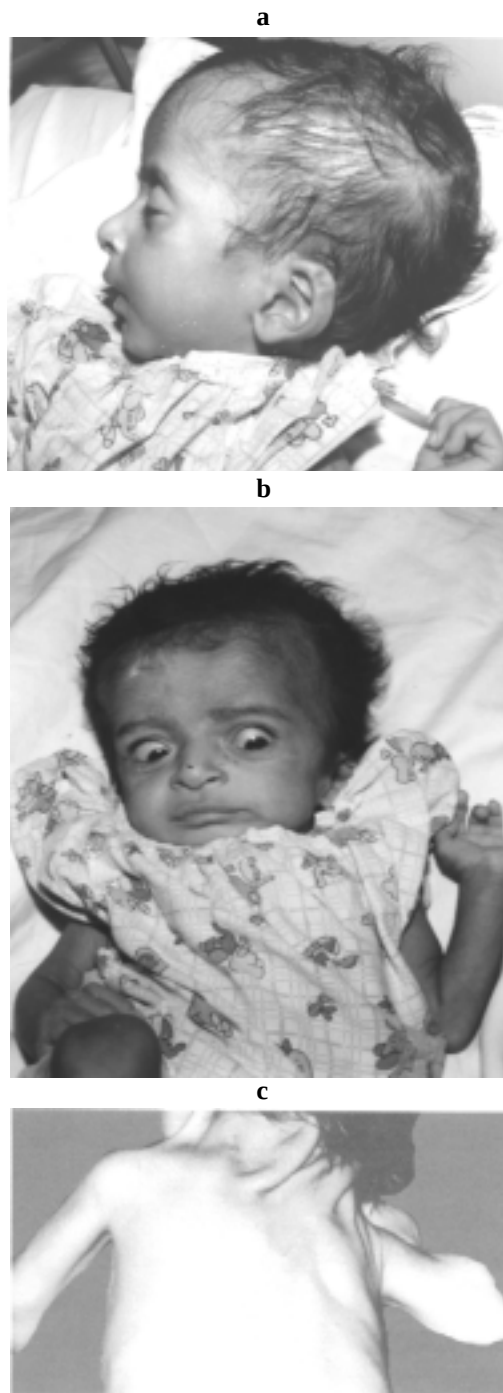


Fig. 1a. Dolichocephaly  
1b. Sunset eyes  
1c. Webbed Neck

investigation revealed hypothyroidism. The electro cardiographic report revealed right ventricular hypertrophy. Transthoracic echocardiography revealed transposition of great vessels with intact ventricular septum, small PDA and ASD.

Chromosomal analysis of the proband was performed on 25 well spread metaphase plates and monosomy of X chromosome was diagnosed that is, 45,X,16qh+ (Fig. 2). This patient also possessed a variant chromosome 16. The child was subjected to arterial switch repair. The findings on echocardiography were confirmed during surgery. The child remained hemodynamically stable for 48 hours after surgery, but had persistent low cardiac output. The parental karyotypes were normal.

### DISCUSSION

The spectrum of CCVM in patients with Turner syndrome is variable (Nora et al. 1991; Hyett et al. 1997; Prandsraller et al. 1999; Douchin

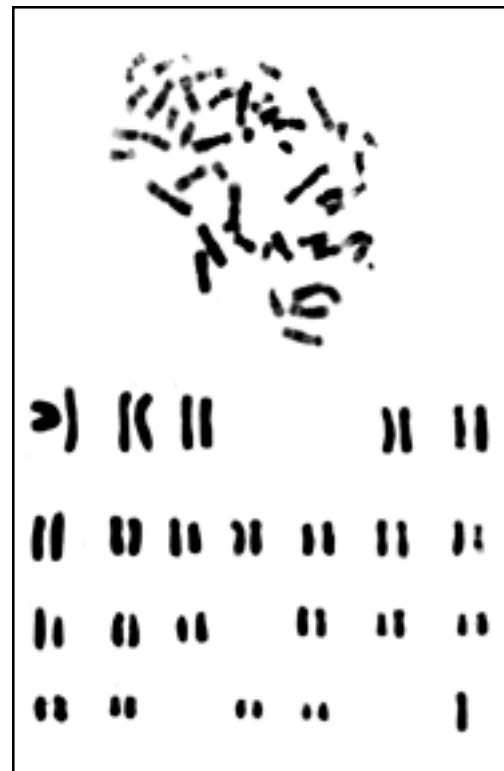


Fig. 2. Karyotype with metaphase plate

et al. 2000; Gopal et al. 2001). Mazzanti and Cacciari (1998) studied a large series of 594 Turner patients from Italy. The prevalence of cardiovascular malformations was 23%. Bicuspid aortic valve and coarctation of aorta were the most prevalent defects. Hirose et al. (1999) reported a rare case of atrial septal defect associated with Turner syndrome. Further, a case of ruptured dissecting aneurysm (Stanford type A) was experienced (Hirose et al. 2000) and emergent total arch replacement was performed successfully. Our patient is the first case of TGA associated with TS which exemplifies the varied spectrum and combination. There may be X-linked factors independently, synergistically or additively involved in the development of cardiovascular defects in these patients. The present case adds to the diversity of clinical abnormalities due to Turner syndrome. Genetic counselling, family screening and fetal echocardiography should be available to these families.

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