Case Report

Rare Presentation of Edward Syndrome with Mesocardia

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Abstract
Edward syndrome is a chromosomal numerical disorder caused by the presence of all or part of an extra 18th chromosome. It is characterized by cardiac, renal, gastrointestinal tract and skeletal malformations. Here we report a case of a neonate admitted with increased work of breathing since birth. He had multiple congenital anomalies. On echocardiogram, mesocardia with atrial septal defect found, which is a rare presentation.

Keywords: Edward syndrome, mesocardia, rocker bottom feet.

Introduction
Edward syndrome is a genetic disorder with an incidence of 1:65001. It is named after John Hilton Edwards, who first described the syndrome in 19602. It is the second most common autosomal trisomy, after Down syndrome. Some of the clinical features are low birth weight, microstomia, micrognathia, low set malformed ears, prominent occiput, rocker bottom feet, flexion deformities of fingers, congenital heart disease, increased gyri and heterotopias of neurons1. There has been a 3:1 preponderance of females to males; more than 130 different abnormalities have been noted.2 We report a case of newborn with clinical features consistent with Edwards syndrome with mesocardia and atrial septal defect.
**Case report:**

Edward syndrome is a genetic disorder with an incidence of 1:6500\(^1\). It is named after John Hilton Edwards, who first described the syndrome in 1960\(^2\). It is the second most common autosomal trisomy, after Down syndrome. Some of the clinical features are low birth weight, microstomia, micrognathia, low set malformed ears, prominent occiput, rocker bottom feet, flexion deformities of fingers, congenital heart disease, increased gyri and heterotopias of neurons\(^1\). There has been a 3:1 preponderance of females to males; more than 130 different abnormalities have been noted.\(^2\) We report a case of newborn with clinical features consistent with Edwards syndrome with mesocardia and atrial septal defect.

**Case report:**

One hour old male neonate weighing 2.25 kg, product of 3\(^{rd}\) degree consanguineous marriage and 3\(^{rd}\) in birth order, referred from Government area hospital, Godavarikhani, Karimnagar district to neonatal intensive care unit, Chalmeda Anand Rao Institute Of Medical Sciences, Karimnagar with complaints of respiratory distress. Mother had regular antenatal checkups and she developed polyhydramnios during 3\(^{rd}\) trimester and targeted imaging for fetal anomalies scan revealed that the fetus was having right hydroureteronephrosis. At 38 weeks of gestational age mother felt decreased fetal movements and underwent emergency LSCS, delivered a full term, symmetrical intrauterine growth restricted baby who cried immediately after birth.

On examination, baby had microcephaly with head circumference of 31 cm, wide Anterior Fontanelle(3.5cm×3.5cm), low set ears, microphthalmia and micrognathia, hypertrichosis, characteristic clenched hand posture with overriding of 5th finger over ring finger and 2\(^{nd}\) finger over middle finger, Talipes equinovarus deformity of the right ankle, left leg showing rocker bottom foot and micropenis (fig.1). Head circumference was 31 cm and length was 41 cm. With this features, provisional diagnosis of Edward syndrome was made.

Baby was evaluated for early neonatal sepsis, the complete blood picture showing hemoglobin 18.2 gm/dl, total leucocyte count 16500/cumm, platelet count 2,95,000/cumm, C-Reactive Protein <0.6, Serum electrolytes sodium 140meq/dl, potassium 4.5 meq/dl, Chloride 102 meq/l, Blood urea and serum Creatinine was within normal limits. Chest X ray showed bilateral patchy infiltrates may be due to retained fluid suggestive of Transient Tachypnea of Newborn (TTNB) with mesocardia. [Fig: 2] 2D Echo revealed that baby was having Mesocardia with ASD with aneurysmal dilatation of Interventricular Septum. Neurosonogram report given as dorsal cyst or prominent cistern magna. Ultrasound Abdomen showed gross right hydroureteronephrosis. Chromosomal analysis revealed an extra 18\(^{th}\) chromosome (Fig 3).

Baby was treated for TTNB with oxygen, warmth care and supportive treatment. Baby was discharged on 14\(^{th}\) day of life. At the time of discharge baby was active,
accepting feeds and had no respiratory distress. On follow up at two month of age baby was found to be not gaining weight and baby developed right inguinal hernia. On 70th day of life baby was readmitted with severe respiratory distress, diagnosed as bronchopneumonia, baby was ventilated. On 5th day of post admission (75th day of life) baby expired.

**Discussion:**
Edward syndrome is considered to be a fatal congenital disorder with mean survival of 1-3 months\(^3\). About 95% die in utero, among live born infants, only 50% live up to 2 months and only 5-10% would be able to complete their first birthday.\(^4,5\) The longest survival time reported till date is 50 years\(^6\). 80% of the cases were reported to be female.

Edward syndrome occurs because of non-dysjunction of the 18th chromosome during meiosis of the reproductive cells of the mother and the father. Advanced maternal age, environmental factors and low socioeconomic level play a role in the incidence of Edward Syndrome\(^8\). Advanced maternal age was not present in this case but socio economic status was falling under lower class of modified Kuppuswamy scale.

History of polyhydramnios, single umbilical artery, intrauterine growth retardation and decreased fetal movements during the prenatal period indicate suspicious Edward’s syndrome\(^9\). These have been documented in the present case.

There are hallmark features present in a majority of trisomy 18 patients. These features include mental and developmental delay, growth deficiency, abnormal craniofacial profile, clenched hands with overlapping digits, internal organ malformations including inguinal or umbilical hernias, and multiple congenital heart defects\(^3, 10\). Many structural defects associated with Trisomy 18 have been reported. In 97% of cases with trisomy 18, structural disorders are found in at least three organs. In Edward’s syndrome, VSD has been reported with a rate of 67%, underdevelopment of reproductive organs in 50%, horseshoe kidney in 32%, omphalocele in 14%, diaphragm hernia in 11% and oesophageal atresia in 11%\(^11\). Only two cases accompanied by meningocele have been reported \(^{12, 13}\). Oesophageal atresia, meningocele and thinning of corpus callosum were reported in earlier cases. A newborn with Edward syndrome associated with mesocardia with ASD has not been reported till date although one case in utero was reported to have mesocardia but the fetus was terminated. In our case, since this rare feature of mesocardia with ASD was found, the baby was of profound interest.

According to a study, fetuses with trisomy 18 have an abnormal immunological development. They found that the mean T cell and NK cell counts were significantly lower and the mean B cell count was not significantly different\(^14\), these findings could explain the normal laboratory parameters in our case inspite of clinical signs and symptoms which were in favor of sepsis.
The major causes of death in Edward syndrome are sudden death due to central apnea, cardiac failure due to cardiac malformations and respiratory infections due to hypoventilation, aspiration, upper airway obstruction or combination of these factors\textsuperscript{15}. In the present case respiratory infections leading to respiratory failure was the cause of death

**Conclusion:**

Majority of the clinical features are typical of Edward syndrome in this baby except mesocardia which was seen in our case is not reported till now. So mesocardia may be considered as one of the rare feature of Edward syndrome
References

2. Edwards Jh, Harnden Dg, Cameron Ah, Crosse Vm, Wolff Oh. A new trisomic syndrome. Lancet 1960; 1:787-90
15. Anna Cereda, John Carey. The trisomy 18 syndrome. Orphanet journal of Rare disease 2012; 7:81
Figures

Figure 1: Microthalmia

Figure 2: Xray Mesocardia
Figure 3: Karyotyping report