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Case Report

Patau Syndrome Mimicking Edwards Syndrome – A Case Report

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Abstract:

Patau syndrome (Trisomy 13) is a rare chromosomal anomaly incidence being 1 in 12000-20,000 live births, caused by the presence of an extra copy of chromosome 13, with advanced maternal age being a common attributable risk factor. It is characterized by multiple congenital anomalies involving the central nervous system, cardiovascular, musculoskeletal, and genitourinary systems, and is associated with high neonatal morbidity and mortality. We report the case of a 1 month 15 days old female infant who presented with progressive respiratory distress, making weaning from oxygen support difficult. On physical examination, the infant exhibited multiple dysmorphic features, including microcephaly, microphthalmia, low-set ears, postaxial polydactyly, hypertonia, rocker-bottom foot, overlapping fingers, clenched hand and umbilical hernia.

Further evaluation revealed significant cardiac anomalies on two-dimensional echocardiography, including a 6mm atrial septal defect (ASD),8mm ventricular septal defect (VSD), and 4mm patent ductus arteriosus (PDA), complicated by severe pulmonary hypertension. Chest radiograph showed a normal cardiothoracic ratio, while ultrasonography of abdomen and pelvis revealed a left renal cortical cyst, indicating associated genitourinary involvement.

Owing to the presence of overlapping fingers and Rocker bottom foot, an initial clinical suspicion of Edwards's syndrome was made. Karyotyping was subsequently performed. Cytogenetic analysis confirmed the diagnosis of Patau syndrome (Trisomy 13). The child then shifted to the intensive care unit, received ventilatory support, pulmonary vasodilators, and decongestive therapy (intravenous furosemide), along with comprehensive supportive care. Despite intensive supportive care the infant's condition deteriorated and she eventually succumbed. This case highlights sometimes Patau Syndrome can mimic Edwards Syndrome. It also emphasizes the importance of early recognition of trisomy syndromes, the role of antenatal screening and diagnosis by chorionic villous sampling biopsy and amniocentesis by karyotyping, and the need for genetic counseling to guide management and parental decision-making.

Keywords: Patau syndrome (Trisomy 13), Edwards syndrome, Chromosomal anomaly, Rocker-bottom foot, Hypertonia.

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Introduction

Patau syndrome is a rare chromosomal anomaly characterized by the presence of an additional copy of chromosome 13 and is classically associated with craniofacial dysmorphism, post-axial polydactyly, and severe multisystem congenital anomalies. The incidence of the condition is estimated to be 1 in 12,000–20,000 live births worldwide, with a female predominance. More than 90% of affected infants die within the first month of life, and only a small proportion survive beyond infancy. Trisomy 13 results from meiotic nondisjunction, mosaicism, or chromosomal translocation. Infants with Patau

syndrome commonly exhibit congenital heart defects, central nervous system malformations, ocular anomalies, renal abnormalities, hypertonia, and severe developmental impairment. Due to the poor prognosis, management is primarily supportive, focusing on symptom relief and family-centred care. We report a case of Patau syndrome in a female infant presenting beyond the neonatal period with complex congenital heart disease and renal involvement.

Case Report: A 1-month-15-day-old female infant, second order birth was admitted with complaints of hurried breathing since day 20 of life. The infant was delivered by lower segment caesarean section and had a history of birth asphyxia, requiring resuscitation at birth. She was born out of non-consanguinity. Mother is 23 years old. Elder sibling is 4 years old with no significant illness

On admission, the infant was tachypneic with signs of respiratory distress including poor feeding and hypoxemia in room air.

Clinical examination revealed a sick-looking infant with generalised hypertonia. Dysmorphic features included microphthalmia with small palpebral fissures, flattened nasal bridge, low-set ears, and full cheeks. Limb examination showed clenched hands with overlapping fingers, Rocker bottom foot and post-axial polydactyly of the limbs. An umbilical hernia was also noted. Ultrasonography of the abdomen and pelvis revealed a left renal cortical cyst, while other abdominal organs were normal.

Cardiovascular examination revealed wide fixed splitting of S2. 2D echocardiography demonstrated 6mm atrial septal defect, 8mm large ventricular septal defect,4mm large patent ductus arteriosus, and severe pulmonary arterial hypertension.

Considering morphological features typical of Edwards like Rocker bottom foot, overlapping fingers initially Edwards Syndrome was suspected. Karyotyping was sent and it came out to be Patau Syndrome (Trisomy 13).

The infant was started on anti-failure medications. Initially managed with moist oxygen, the child subsequently required endotracheal intubation and mechanical ventilation due to worsening respiratory distress. The parents received genetic counselling and were informed about the poor prognosis and need for supportive care. Gradually clinical condition deteriorated and subsequently succumbed.

Figure 1: Microphthalmia

Figure 2: Abnormal posturing and umbilical hernia

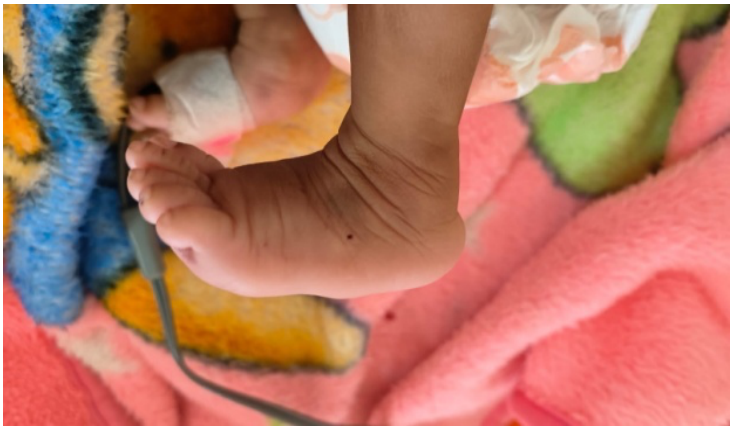

Figure 3: Rocker bottom foot

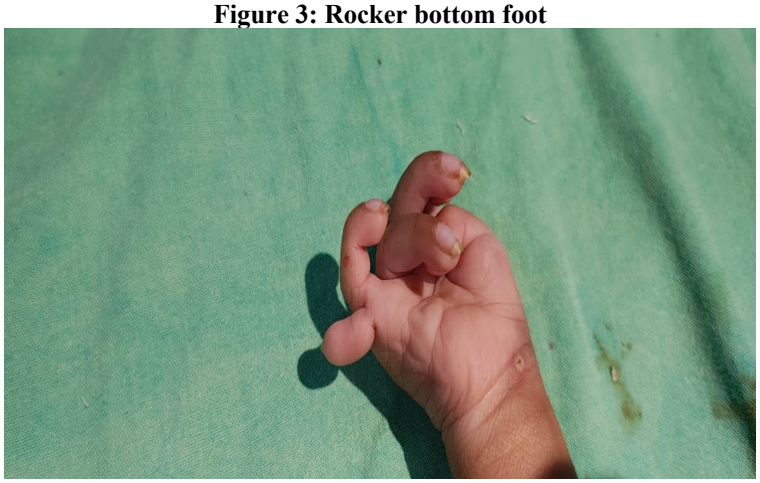

Figure 4: Post axial polydactyly of hand

Figure 5: Overlapping fingers

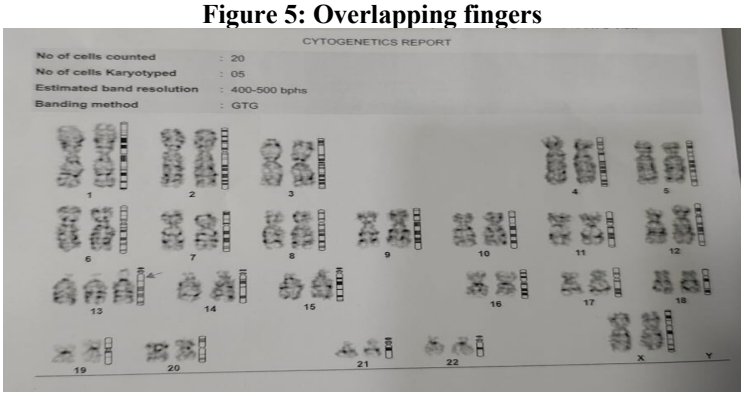

Figure 6: Karyotyping showing extra copy of chromosome 13

Figure 7: On Mechanical ventilation

Discussion

This illustrates the phenotypic overlap between Patau syndrome and Edwards syndrome. Features such as rocker-bottom feet, clenched hands, overlapping fingers, and hypertonia are classically described in Trisomy 18 but may also be present in Trisomy 13, leading to diagnostic confusion. In contrast, post-axial polydactyly, microphthalmia, and severe ocular abnormalities are more suggestive of Trisomy 13.

Congenital heart defects occur in over 80% of infants with Trisomy 13 and are a major contributor to early mortality. Renal anomalies further compound morbidity. Given the poor prognosis, management remains supportive, and ethical considerations regarding the extent of intervention should involve shared decision-making with the family.

Conclusion

Patau syndrome may be clinically suspected based on characteristic dysmorphic features and multisystem involvement, even when atypical features mimicking Edwards syndrome are present. Cytogenetic confirmation is essential for accurate diagnosis. Early recognition allows appropriate prognostication, avoidance of unnecessary aggressive interventions, and timely genetic counselling done.There is no definitive treatment, and management remains supportive. Early diagnosis is essential for prognostication, avoidance of unnecessary aggressive interventions, and appropriate parental counselling. Antenatal screening and diagnosis by chorionic villous sampling biopsy and amniocentesis with karyotyping if further pregnancy is planned though it is rare.

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