

Association of Congenital Diaphragmatic Hernia in a Baby with Patau Syndrome

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Introduction

Patau syndrome or trisomy 13 is one of the most severe forms of trisomy and the affected children are usually viable for short duration. Median survival is reported as 7-10 days. Gestational age is the main predictor of mortality.¹ Frequent clinical manifestations include neural tube defects, brain anomalies; specially holoprocencephaly, microphthalmia, cleft lip and palate, congenital heart disease, overlapping and flexion of fingers, polydactyly and Rocker bottom feet.¹

There are only very few documented associations of Patau syndrome with congenital diaphragmatic hernia.² Here we report a case of a child with Patau syndrome associated with diaphragmatic hernia.

Case report

A baby boy weighing 2500g was delivered by an emergency LSCS at 34 weeks due to major degree placenta previa with bleeding. Parents were non consanguineous and healthy. Baby was resuscitated at birth and 1min APGAR score was 6 and 5min APGAR score was 9. Baby was detected to have several congenital anomalies at birth including low set ears, wide anterior fontanelle, bilateral cleft lip and palate, bilateral overlapping flexed fingers, left side talipes equinovarus deformity and bilateral Rocker bottom feet. His abdomen was scaphoid and heart sounds were best heard in the right side of the precordium with prominent systolic murmur. Baby was admitted to Neonatal Intensive Care Unit. Due to persistent respiratory distress with metabolic acidosis, he was intubated and ventilated. Chest xray revealed a left sided congenital diaphragmatic hernia. An echocardiogram was done and it revealed moderate size Ostium secundum Atrial Septum Defect.

Considering the presence of congenital anomalies and the poor prognosis of the condition, conservative management was planned after discussing with the paediatric surgical team and the parents. Baby succumbed to death on day 16 of life.

Karyotyping report obtained in few weeks after the death confirmed the diagnosis of Patau syndrome.

Discussion

Patau syndrome results mainly due to non dysjunction and incidence increases with advanced maternal age.² Babies with Patau syndrome usually presents with a low APGAR score at birth. Clinical manifestations detected in this child were microphthalmia, cleft lip and palate, congenital heart disease, overlapping and flexion of fingers and Rocker bottom feet. Unlike many babies with Patau syndrome that are small for their gestational age, this baby was grown adequately for his age.

Diaphragmatic hernia (DH) is a communication between the thoracic cavity and abdominal cavity with possibility of abdominal contents in the thorax. The prognosis and severity depends on the type of hernia and associated anomalies. Associated pulmonary hypoplasia is the major limiting factor for survival. The commonest type is Bochdalek type (90% of Congenital DH) in which defect is in the postero-lateral aspect of the diaphragm.³

The incidence of DH is 1:2000 to 1:5000 in live births. Female to Male ratio is 2:1 and most defects occur in the left side (85%).³ Associated anomalies include brain anomalies, Oesophageal atresia, Omphalocele, Cardiovascular anomalies. DH is a recognized anomaly in many chromosomal syndromes. 10% of diaphragmatic Hernia is associated with chromosomal anomalies.³ The most common are trisomy 18 and isochromosome 12p (Pallister Killian syndrome).

Trisomy 21, Turner syndrome, Cornelia de Lange syndrome, Denys Drash syndrome, Donnai Barrow syndrome, Fryns syndrome, spondylocoastal dysostosis, Meachan syndrome are other associated syndromes.⁴ Even though there is a recognized association between Trisomy 13 and congenital diaphragmatic

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hernia, it is a rare occurrence. There are only few case reports available in literature.

In a similar case report published in 2015, the baby had undergone corrective surgery for diaphragmatic hernia but had succumbed to death later.² We opted for conservative management in this child considering the known poor prognosis of the condition with associated multiple anomalies.

Reference

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