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Case Report

Antenatal diagnosis of Patau syndrome with previous anomalous baby

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ABSTRACT

Patau syndrome is the least common and most severe of the viable autosomal trisomies with median survival of fewer than 3 days was first identified as a cytogenetic syndrome in 1960. Patau syndrome is caused by an extra copy of chromosome 13. In this case report, we present antenatal imaging findings & gross foetal specimen correlation of foetus with Patau syndrome confirmed by karyotyping in third gravida who had significant previous obstetric history of gastrochisis in monochorionic and monoamniotic twins who died at 14 weeks of gestation.

Keywords: Patau syndrome, Trisomy 13, Ultrasound, Polydactyly

INTRODUCTION

Patau syndrome or trisomy 13 is the most severe of the three autosomal trisomies characterized by multiple congenital anomalies.

Incidence of trisomy 13 is about 1 in 15000 births.

The classical features of Trisomy 13 include defects of auricles, eyes (microphthalmia, strabismus, iris coloboma) and mouth (cleft lip, palate), holoprosencephaly, polydactyly, heart and scalp defects. Neonates with trisomy 13 die usually within the first few hours or days of life. The rare survivors have profound mental retardation and seizures.

CASE REPORT

A twenty eight year old third gravida presented for routine antenatal sonography. Patient had significant past history of anomalous child in previous pregnancy with polyhydramnios and gastrochisis in both monoamniotic and monochorionic twins who died at 14 weeks of gestation. During sonography post axial polydactyly was noted with evidence of bilateral cleft lip and cleft palate. Umbilical cord revealed single umbilical artery. Four-chamber view of heart revealed truncus arteriosus. Previous history of anomalous child, polyhydramnios and intrauterine foetal demise indicated possibility of chromosomal abnormality. Patient opted for termination of pregnancy.

Gross specimen findings of the foetus after medical termination of pregnancy were consistent with antenatal ultrasound findings. Karyotype study confirmed Patau syndrome.

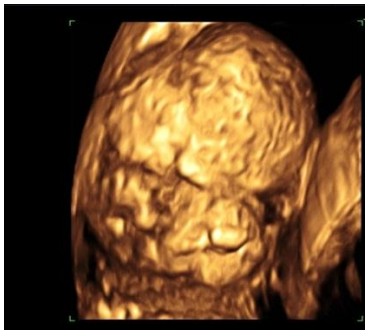

Figure 1: 3D imaging findings reveal bilateral cleft lip and cleft palate.

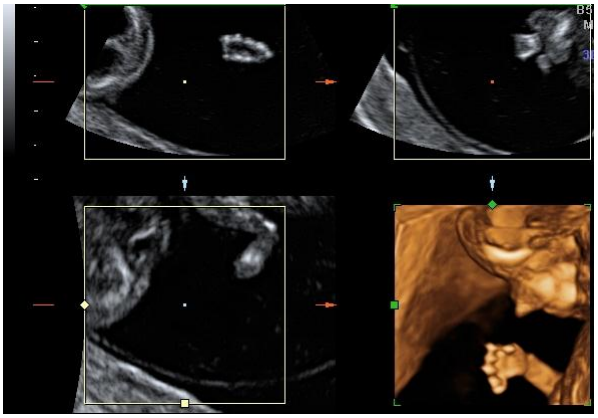

Figure 2: 3D imaging findings reveal post axial polydactyly along the ulnar side.

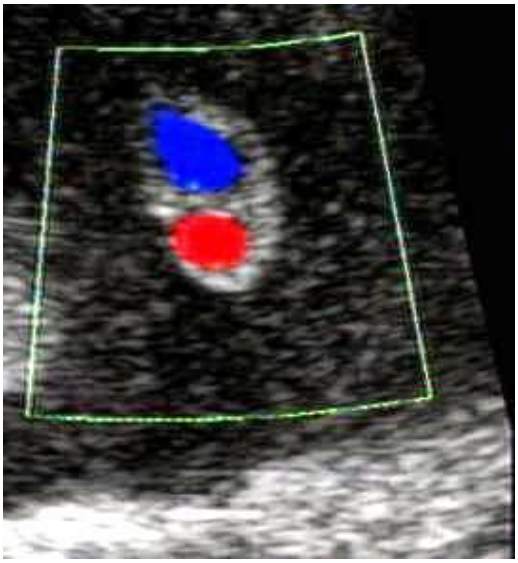

Figure 3: Colour Doppler findings reveal single umbilical artery.

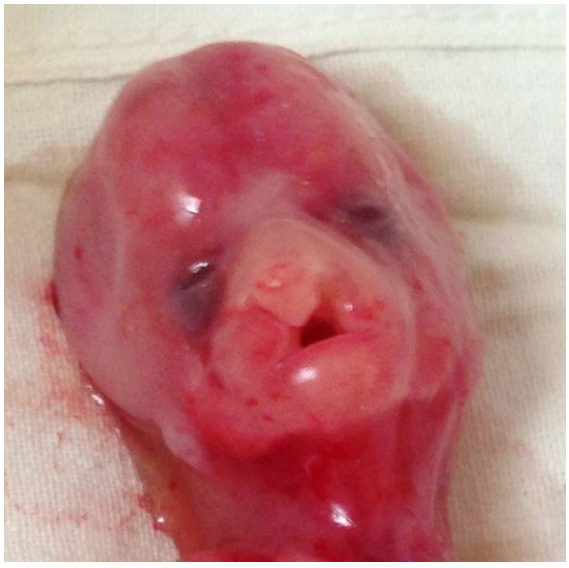

Figure 4(a): Bilateral cleft lip and cleft palate.

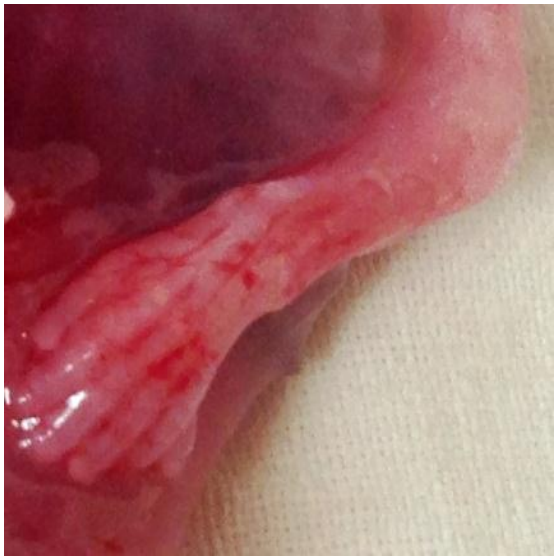

Figure 4(b): Post axial polydactyly.

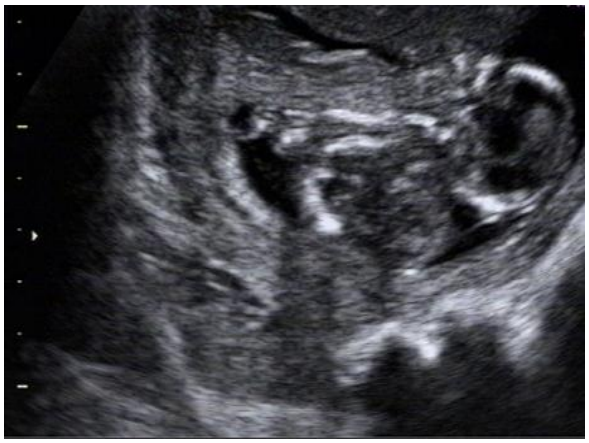

Figure 5: Image from previous pregnancy: gray scale ultrasound findings of the foetus showing gastrochisis in patient's previous pregnancy.

DISCUSSION

Trisomy 13 or Patau syndrome was first identified as a cytogenetic syndrome in 1960. Trisomy 13 is the third most common autosomal trisomy at birth, with Trisomy 21, followed by trisomy 18, occurring more frequently.[1,2]

Trisomy 13 affects 1 in 5000 births or 1 in 20000 liveborns.[3,4]

There are a few genetic variants of this disorder. The most frequent one is free trisomy 13 (1/12000) and is characterised by the presence of an extra copy of chromosome 13, a medium-length acrocentric chromosome in each cell of the body.[5]

A small percentage of cases have a mixed population of cells, some presenting normal karyotype and the others having an extra copy of the chromosome 13. The third

variant is represented by a Robertsonian translocation (1/56000-1/80000).[6]

Trisomy 13 usually involves a maternal meiotic error of nondisjunction; however, paternal errors do occur.[3]

Patau syndrome is a clinically severe condition associated with low survival rates because of malformations of the Central Nervous System (CNS), cardiac, circulatory and urogenital systems. This syndrome occurs in all ethnicities and equally affects both males and females.

Congenital anomalies include abnormalities of face, brain, extremities and heart. Midline facial cleft, hypotelorism, cyclopia, microphthalmia and absence of nose. Intracranial anomalies that can be seen with trisomy 13 include holoprosencephaly, microcephaly, abnormal posterior fossa, agenesis of corpus callosum and ventriculomegaly.

Trisomy 13 is present about 50% of the time when holoprosencephaly is detected on fetal ultrasound.[13]

More than 90% of fetuses with this trisomy have cardiac defects and include ASD, VSD and PDA. Approximately 40% of foetuses with trisomy 13 have echogenic intracardiac foci.[9] 30% of the affected foetuses have enlarged echogenic kidneys similar to polycystic kidneys.[10] Abnormalities of extremities include polydactyly and radial dysplasia. polydactyly is found in approximately 80% fetuses with Trisomy 13.[7] Patau syndrome is expressed prenatally and is fully evident at birth.

Extremity malformations are commonly found in a wide range of Chromosome defects. The detection of abnormal hands and feet should stimulate a search for other markers of chromosome abnormalities.[7] Clubfoot has been associated with multiple chromosomal abnormalities including trisomy 18 and 13.[11]

Other features or trisomy 13 may include clenched hands, close-set eyes, hernias, coloboma, low-set ears, mental retardation, seizures, single palmar crease, microcephaly, microphthalmia, micrognathia, cryptorchidism cutis aplasia, but the lesion has a strong association with trisomy 13.[8,14]

A significant number of cases that are trisomic for chromosome 13 end in spontaneous abortion, foetal demise, or stillbirth.[8] The main differential diagnosis of Trisomy 13 is Meckel gruber syndrome because of the similarity of the findings polydactyly, neural tube defects (posterior encephalocele) and enlarged echogenic kidneys.[7]

Alterations in chromosomes are seen in 6% to 7% of stillborns, making clinical and parental counseling regarding autopsy and karyotyping an essential component of obstetrical care.[12]

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