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CASE REPORT

PATAU SYNDROME- THE LEAST COMMON AND THE MOST SEVERE VIABLE AUTOSOMAL TRISOMY

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ABSTRACT:

Patau syndrome also known as trisomy 13 is due to a chromosomal abnormality associated with intellectual disability and multiple physical abnormalities like cardio vascular defects, brain or spinal cord abnormalities, cleft palate or lip, extra fingers or toes and decreased muscle tone. Infants with trisomy 13 die within the first few days or weeks of life. About 5-10% of infants with this condition live beyond their first year of life.

Key words: Patau Syndrome, Autosomal Trisomy, chromosomal abnormality

INTRODUCTION

'Patau Syndrome', synonymous with trisomy 13, is a rare chromosomal abnormality which affects approximately 1 in 15000 live births and is associated with multisystemic abnormalities.[1,2] Amongst the three trisomies compatible with extra-uterine life namely, Patau , Down & Edward Syndromes, Patau Syndrome is the most severe and has the least incidence and hence merits attention.[3] First identified as a cytogenetic syndrome in 1960, Patau syndrome is caused by an extra copy of chromosome 13, a medium length acrocentric chromosome in which a person has three copies of genetic material instead of the usual two copies. So, the extra DNA from chromosome 13 appears in some or all the cells of the body. Normal development is affected by this extra material. Most cases are not inherited. In these cases, trisomy 13 is caused by events in either the sperm or egg that form the fetus.[4] The clinical features are however severe. Mental deficiency is a consistent feature. The other frequent clinical feature includes polydactyly, flexed fingers, rocker bottom feet, facial clefting, neural tube defects and heart defects.[4]

Most often, Patau syndrome is recognized at birth with the presence of structural birth defects and poor neurological performance.[4]

Key words: Patau syndrome, Trisomy 13, Cytogenetic syndrome

CASE SUMMARY:

A 24 year old primigravida with 19 weeks' gestation, ultrasonographically diagnosed with an anomalous baby due to the presence of oligohydamnios, evidence of IUGR and omphalocele. The specific diagnosis responsible for the anomalies could not be made with conviction on

radiology. Patient underwent medical termination of pregnancy. A perinatal autopsy was conducted after parental consent through a linear midline incision from symphysis mentii to symphysis pubis circumventing the umbilicus. Thoracic and abdominal organs which were removed enmasse revealed multiple abnormalities.

Autopsy findings

1. Gross examination:

a) Weight : 400grams
b) Crown heel length : 24.5 cms
c) Crown rump length : 17 cms
d) Left foot : 2 cms
e) Head circumference : 14 cms
f) Chest circumference : 10.5 cms

2. External abnormalities:

a)Cleft lip/Cleft palate (Figure-1)
b)Short webbed neck with cystic hygroma (Figure-2)
c)Lowset ears
d)Coarctation of aorta (Figure-3)
e)Absent forearm bones ,Agenesis of fingers
f)Omphalocele with protrusion of liver and intestinal coils (Figure-4)
g)One umbilical artery and one umbilical vein

3.Microscopy:

1) Sections from the sponge-like swelling in the neck showed thin walled vascular channels filled with RBCs and lined by endothelium. Smooth muscle bundles were also seen. Features were suggestive of **Cystic hygroma**.(Figure-5)
2) Congestive changes were seen in bilateral kidneys, adrenals, liver, spleen and Intestine.

Autopsy diagnosis: Patau syndrome

DISCUSSION

Patau syndrome is well known for its high mortality rate and multiple congenital abnormalities resulting in severe physical and mental impairment. It is the third most common autosomal trisomy in newborn after trisomy 21 and 18. [5]

It recognition is important since a personal or close family history of giving birth to an affected child increases the risk. The risk also increases with increased maternal age but it is not as significant as in Down's syndrome (trisomy 21) or Edward's syndrome (trisomy 18).[6]

Many fetuses do not survive until term and are stillborn or get aborted spontaneously. [7]

Cleft lip and palate, ear malformations, omphalocele and abnormalities of the hand as seen in the present case have also been reported by other authors. [5,7]

Differential diagnosis from Edward syndrome on autopsy alone can be difficult since it can also have similar features. Cytogenetic studies and chromosomal analysis are necessary to confirm the diagnosis following which parents need a great deal of support and counseling.

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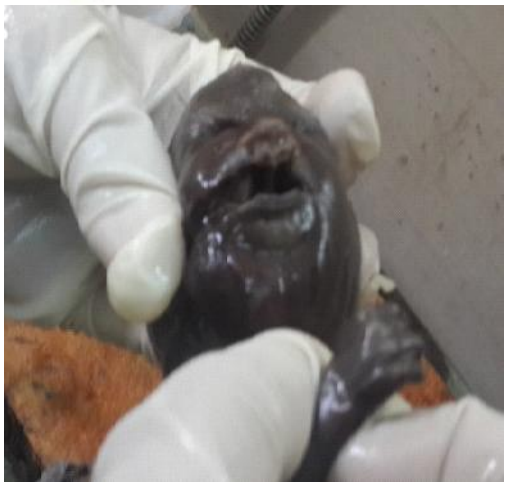

Figure 1

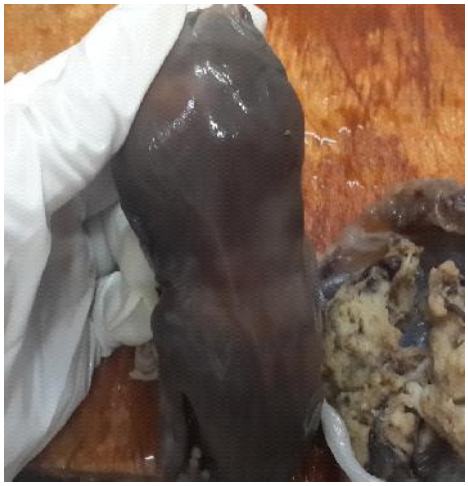

Figure 2

Figure 3

Figure 4

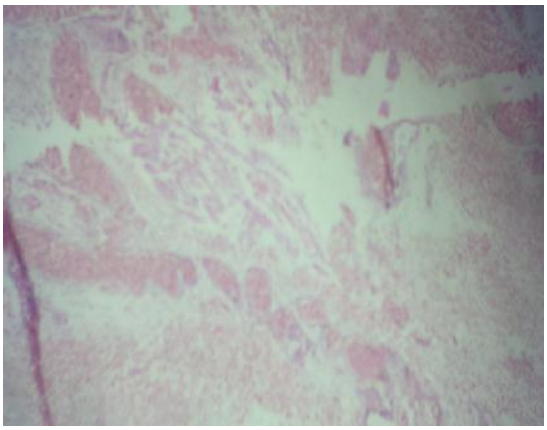

Figure 5

FIGURES

Figure 1- Cleft lip/Cleft palate; **Figure 2-** Short webbed neck **Figure 3-** Coarctation of aorta;**Figure 4-** Omphalocele with protrusion of liver and intestinal coils; **Figure 5-** Thin-walled vascular channels filled with RBCs

Both parents should undergo chromosomal analysis as the chance of a balanced translocation in one of the parents is high. This has significance in future pregnancies because of a higher risk of recurrence.[8]

Screening and or prenatal diagnosis are mandatory in future pregnancies. Women with a previous trisomy pregnancy, especially those under 35 years, have an increased risk in future pregnancies.[8]

Specific ultrasound findings like nuchal translucency, cardiac defects, neural tube defects, facial clefting, renal abnormalities and omphalocele may suggest trisomy 13 and subsequent cytogenetic study will help in confirmation.[9]

A study from king college hospital revealed that fetal examination for exomphalos and megacystis (enlarged urinary bladder) can identify >90% fetuses with trisomy 13. [10]

Multiple marker screening tests in the first trimester may also help identify a fetus with trisomy 13or18

Markers like maternal age, nuchal translucency measurement, pregnancy associated-plasma protein A (PAPPA) and human chorionic gonadotrophin also help in diagnosis.

Fetal anomaly scan may reveal the anomalies as in the present case.

Second trimester screening test can help if the pregnant mother presents later. [9]

In a study of 44 cases of trisomy 13, 64 % of cases were detected after chromosomal analysis following abnormalities observed on fetal anomaly scanning in second trimester, 3% of cases were detected on serum screening programme for Down syndrome and 11 % were detected postnatally in a UK based study.[6]

Prenatal amniocentesis or chorionic villus sampling also helps in definitive diagnosis.

CONCLUSION:

Perinatal autopsy is the gold standard for diagnosis of Patau syndrome since ultrasonography may not provide a conclusive opinion as in the present case. Genetic counseling is mandatory once the diagnosis is confirmed by cytogenetic studies.

REFERENCES:

1) Baty BJ, Jorde LB, Blackburn BL, Carey JC. Natural history of trisomy 18 and trisomy 13: II. Psychomotor development. Am J Med Genet. 1994 Jan 15. 49(2):189-94.

2) Morris JK, Savva GM. The risk of fetal loss following a prenatal diagnosis of trisomy 13 or trisomy 18. Am J Med Genet A. 2008 Apr 1. 146(7):827-32.

3) Stafford CO. A Case Study of Trisomy 13: Balancing Hope and Reality. Pubmed.gov.2015 Aug; 15(4):285-9.

4) Summar K, Lee B. Cytogenetics. In : Kliegman RM, Stanton BF, St. Geme JW, Shor NF, Behrman RE,eds. Nelson Textbook of Paediatrics. 19th ed. Philadelphia, PA: Sainders Elsevier, 2011: chap 76.

5) Tsukada K, Imataka G, Suzumura H,et al; Better prognosis in newborns with trisomy 13 who received intensive treatments: a retrospective study of 16 patients. Cell Biochem Biophys. 2012 Jul;63(3):191-8.

6) Parker MJ, Budd JL, Draper ES, et al; Trisomy 13 and trisomy 18 in a defined population: epidemiological, genetic and prenatal observations; prenat Diagn.2003 Oct;23(10):856-60.

7) Plaiasu V, Ochiana D, Motei G,et al; Clinical relevance of cytogenetics to pediatric practice. Postnatal findings of Patau syndrome-Review of 5 cases. Maedica (Buchar). 2010 Jul;5(3):178-85.

8) De Souza E, Halliday J, Chan A, et al; Recurrence risks for trisomies 13,18, and 21. Am J Med Genet A. 2009 Dec; 149A(12):2716-22.

9) Chitayat D, Langlois S, Wilson RD; Prenatal screening for fetal aneuploidy in singleton pregnancies. J Obstet Gynaecol Can. 2011 Jul;33(7):736-50.

10) 10)Papageorghiou AT, Avgidou K, Spencer K, et al; Sonographic screening for trisomy 13 at 11 to 13(+16) weeks of gestation. Am J Obstet Gynecol. 2006 Feb; 194(2):397-401.