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Patau Syndrome

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Abstract

Patau syndrome is a chromosomal abnormility in which patient has an additionalchromosome 13 due to non disjunction of chromosome during meiosis. Some are caused by translocation while others are caused by mosaic patau syndrome.

Key words: Patau syndrome; Trisomy 13; Trisomy D.

Introduction

Patau syndrome is a chromosomal abnormility in which patient has an additional chromosome 13 due to non disjunction of chromosome during meiosis. A small percentage of cases occur when only some of the body's cells have an extra copy of chromosome 13, resulting in a mixed population of cells with differing numbers of chromosomes. This is called Mosaic Patau.

Case summary

A 22 years old rural habitat $G_2P_1L_1A_0$ female was admitted to our labor room with complaints of pain in the lower abdomen. The pain was continuous, dull aching, progressive, radiating to lower back and medial aspect of thigh. She had history of fever 3 days back, for which she took treatment details of which are not available. She was referred herein by a private practitioner.

According to her last menstrual period, her gestational age was 29 weeks. She was a registered case at PHC with 3 antenatal visits. No antenatal USG was done at PHC. She had taken iron and folic acid tablets with 2 doses of TT injection. She had family history of third degree consanguineous marriage. Her first child is a three year girl who was a full-term normal vaginal delivery without any congenital anomaly.

On examination, she was an averagely built lady; with pulse 80/min & blood pressure-120/70 mmHg. Mild pallor was present. Head circumference was 23 cm (<3rd percentile). Per abdominal examination showed uterus of 26 weeks and a fetus with cephalic presentation. Per vaginal examination showed 1 finger loose dilated cervix without any PV bleeding. USG showed live fetus of 26 weeks and 2 days gestational age with congenital anomaly with holoprosencephaly variant with hypotelorism and moderate polyhydroamnios.

Mother's investigation revealed Hb 10 gm%, TLC-11000, Platelet Count-2 lakhs/Cmm. Liver function tests and renal function tests

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Table 1

Clinical Features	Patau Syndrome	Edward Syndrome	Our Patient
Close set eyes	+	-	+
Microcephaly	+	+	+
Holoprocencephaly	+	-	+
Low set ears	+	+	+
Small lower jaw	+	-	+
Hypotelorism	+	-	+
Polydactyly	-	+	-
Inguinal hernia	-	+	-
Omphalocoele	+	+	-
Congenital heart defects	+	+	Can't be Evaluated
Underdeveloped Brain	+	+	+
Cleft lip/palate	+	+	-
Rocker Bottom Feet	-	+	-
Clenched hand with overlapping of fingers	+	+	+
Single umbilical artery	+	+	-

were within normal limits. VDRL, HbsAg and Tridot were negative. Urine showed traces of albumin. She delivered IUD fetus by preterm vaginal delivery.

The baby was an IUD fetus, with multiple congenital anomalies, listed in table no.1.

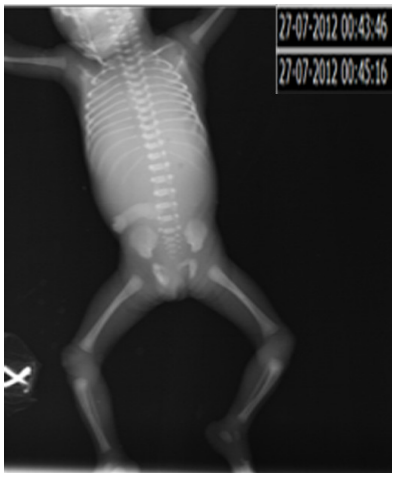
From above table we can rule out Edward syndrome. Ophthalmology opinion was taken which states presence of 2 orbits.

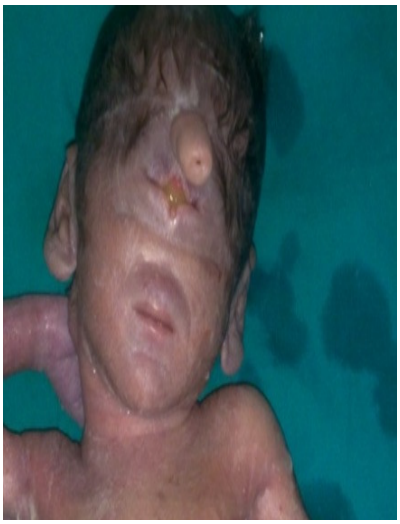
Following investigations were done.

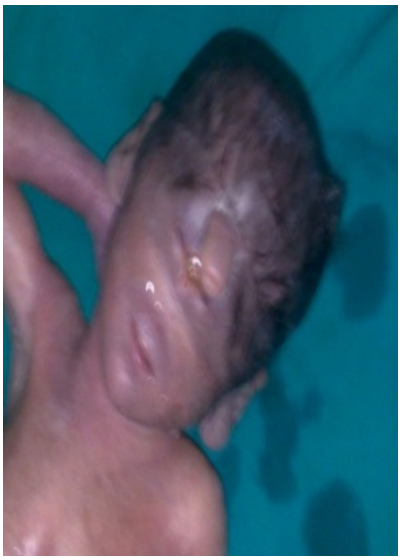
USG Abdomen: Malrotated organs; B/L polycystic kidney.

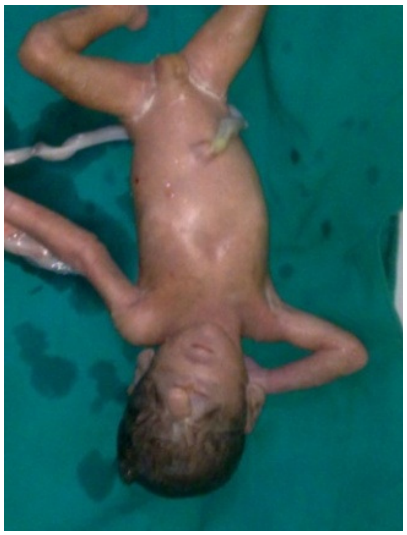
USG Cranium: Underdeveloped Brain

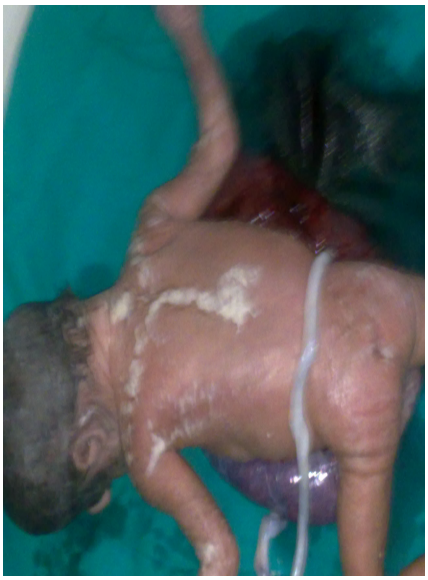
CT Scan Brain (plain): Microcephaly with absent septum pellucidum & a single large ventricle in brain surrounded by thin rim of cortex showing pachygyria. The thalami were partially separated by a rudimentary third ventricle. The small partly formed interhemispheric fissure and falx were seen only in high parietal region & were absent anteriorly Findings were suggestive of holoprosencephaly.

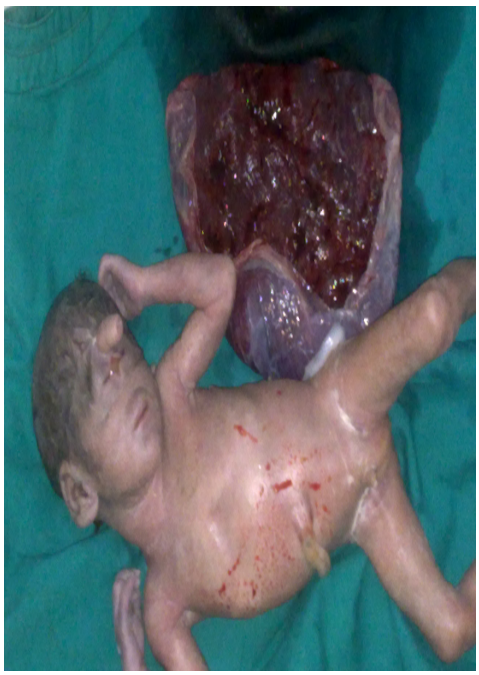












Discussion

Patau syndrome, or "Trisomy 13", as it was first called, was first observed by Thomas Bartholin in 1657. However, the actual genetic and chromosomal-related parts of it were discovered by Dr. Klaus Patau in 1960, hence the name "Patau syndrome". It affects about 1 in 12,000 live births. More than 80% of infants with Patau syndrome die within their first year of life.

Patau syndrome, also known as trisomy 13 and trisomy D, is a chromosomal abnormality, a syndrome in which a patient has an additional chromosome 13 due to a nondisjunction of chromosomes during meiosis. Some are caused by Robertsonian translocations, while others are caused by mosaic Patau syndrome. The extra chromosome 13 disrupts the normal course of development, causing heart and kidney defects. Like all nondisjunction conditions (such as Down syndrome and Edwards syndrome), the risk of this syndrome in the offspring increases with maternal age at pregnancy, with about 31 years being the average.

Dr. Klaus Patau

Genetics

Patau syndrome is most often the result of trisomy 13, meaning each cell in the body has three copies of chromosome 13 instead of the usual two. Patau syndrome can also occur when part of chromosome 13 becomes attached to another chromosome (translocated) before or at conception in a Robertsonian translocation. Affected people have two copies of chromosome 13, plus extra material from chromosome 13 attached to another chromosome. With a translocation, the person has a partial trisomy for chromosome 13 and often the physical signs of the syndrome differ from the typical Patau syndrome.

Most cases of Patau syndrome are not inherited, but occur as random events during the formation of reproductive cells (eggs and sperm). An error in cell division called non-disjunction can result in reproductive cells with an abnormal number of chromosomes. For example, an egg or sperm cell may gain an extra copy of the chromosome. If one of these atypical reproductive cells contributes to the genetic makeup of a child, the child will have an extra chromosome 13 in each of the body's cells. Mosaic Patau syndrome is also not inherited. It occurs as a random error during cell division early in fetal development.

Patau syndrome due to a translocation can be inherited. An unaffected person can carry a rearrangement of genetic material between chromosome 13 and another chromosome. This rearrangement is called a balanced translocation because there is no extra material from chromosome 13. Although they do not have signs of Patau syndrome, people who carry this type of balanced translocation are at an increased risk of having children with the condition.

Manifestation

Of those fetuses that do survive to gestation and subsequent birth, common abnormalities include:

Nervous system

Mental and motor challenged, Microcephaly, Holoprosencephaly (failure of the forebrain to divide properly).

Eye Defects

Microphthalmia, Peters anomaly (a type of eye abnormality), cataract, iris and/or fundus (coloboma), retinal dysplasia or retinal detachment, sensory nystagmus, cortical visual loss, and optic nerve hypoplasia.

Spinal defect

Meningomyelocele.

Musculoskeletal and cutaneous defects

Polydactyly (extra digits), Low-set ears, Prominent heel, Deformed feet known as rocker-bottom feet, Omphalocele (abdominal defect), Abnormal palm pattern, Overlapping of fingers over thumb, Cutis aplasia (missing portion of the skin/hair), Cleft palate.

Urogenital defects

Abnormal genitalia, Kidney defects (polycystic kidney).

Other defects

Heart defects (ventricular septal defect), Single umbilical artery.

Recurrence risk

Unless one of the parents is a carrier of a translocation the chances of a couple having another trisomy 13 affected child is less than 1% (less than that of Down syndrome).

Treatment

Medical management of children with Trisomy 13 is planned on a case-by-case basis and depends on the individual circumstances of the patient. Treatment of Patau syndrome focuses on the particular physical problems with which each child is born. Many infants have difficulty surviving the first few days or weeks due to severe neurological problems or complex heart defects. Surgery may be necessary to repair heart defects or cleft lip and cleft palate. Physical, occupational, and speech therapy will help individuals with Patau syndrome reach their full developmental potential.

Prognosis

More than 80% of children with Patau syndrome die within the first year of life.

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