Edward's Syndrome (Trisomy 18)– Autopsy Report of a Rare Case

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INTRODUCTION

Edward's syndrome is a genetic disorder characterized by Trisomy 18. Incidence of Edward syndrome is reported to be around 1 in 3500 to 7000. It is known to be second most common Trisomy after Down's syndrome (Trisomy 21). It is noted to be more common in girls than in boys in ratio 3:1. But here we report the autopsy report of male foetus. Trisomy 18 has multisystem involvement. Cardiovascular system is most commonly involved, followed by extremities, Urinary system, Head and Neck, Gastrointestinal tract and genitals. There are 3 types of Trisomy 18- Full, Partial and Mosaic type. The most common type is Full Trisomy 18, accounting for about 95% of cases. In Full Trisomy 18, every cell of the body has extra chromosome 18. Both Partial and Mosaic type of Trisomy 18 are very rare. In Partial Trisomy 18, each cell has two copies of chromosome 18 plus a segment of extra material from chromosome 18. In Mosaic Trisomy 18, the extra chromosome is present in some of the cells of the body.¹

In our case we have used Quantitative fluorescent polymerase chain reaction (QF-PCR) to detect Trisomy 18. It has entered the field of prenatal diagnosis to overcome the need to culture foetal cells, hence to allow rapid diagnosis of some selected chromosomal anomalies like aneuploidy in chromosomes 13, 18, 21, X and Y. QF-PCR is feasible on fewer cells, and since the analysis can easily be automated, many samples can be processed at the same time and the whole process taking around 30 min. QF-PCR is based on the amplification of chromosome-specific DNA sequences (STR, short tandem repeats) polymorphic in length between subjects. By means of fluorescent primers, the amplified segments can be visualized and quantified as peak areas on automated DNA scanners. Normal heterozygous subjects are expected to show two peak areas (peaks ratio 1:1) for each chromosome analysed, while trisomies are visualized either as an extra peak (triallelic subjects) or as a 2:1 ratio peak between the two areas.^{2,3}

PRESENTATION OF CASE

29 years old primigravida of non-consanguineous marriage, came for routine antenatal check-up. The antenatal ultrasonography report showed single, intrauterine pregnancy around 20-22 weeks of gestation with following anomalies- cleft lip, cleft palate, ventricular septal defect and absent pulmonary valve.

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In view of multiple congenital anomalies, the couple decided on termination of pregnancy. The foetus was sent for the autopsy. We received the foetus along with placenta weighing 750 grams. Detailed external examination was done for anomalies. Gross facial abnormalities were seen comprising of cleft lip and cleft palate involving bilateral complete lip and palate with collapse of the nose. Both upper limbs were fixed in semi flexion with clenched fists, right hand is showing index finger and fifth finger overlapping middle two fingers. Upper limbs and lower limbs were showing contractures of the joints and could not be extended for the purpose of examination. No other external anomaly noted. Autopsy was done using standard protocol. In the Thorax-right lung appeared anatomically normal with three lobes and the left lung is hypoplastic and displaced upwards. Heart was enlarged occupying the entire left hemithorax and displacing the left lung. In the abdomen, liver was mildly enlarged. Rest of the organs were showing normal anatomy and situs.

Foetal heart blood sample was collected and sent for Quantitative Fluorescent-PCR. The findings of antenatal ultrasonography and examination findings at autopsy were taken into account and possibility of Trisomy 18 was considered. Confirmation of the diagnosis was done by QF-PCR, which is reliable, rapid and economic technique. The couple was given genetic counselling regarding the risk of recurrence in future pregnancies.

PATHOLOGICAL DISCUSSION

Edward's syndrome is an autosomal chromosomal disorder named after John Hilton Edwards, who first described the syndrome in 1960. In 1967, Karunakaran and Pai reported first case from India.⁴ The presence of extra chromosome is because of nondisjunction. In parent-of-origin analyses the extra chromosome is most often of maternal origin, the result of an error during the segregation of chromosomes in meiosis or postzygotic mitosis.⁵ The non-disjunction errors increase in frequency with increase in maternal age. This is also observed in other Autosomal Trisomy syndromes.

Characteristic Findings

Low birth weight, closed fists with index finger overlapping the 3rd digit and the 5th digit overlapping the 4th, narrow hips with limited abduction, short sternum, rocker bottom foot, microcephaly, prominent occiput, micrognathia, cardiac and renal malformations, intellectual disability.⁶

Prenatal Diagnosis

Triple test (15-20 weeks).

- MSAFP- Decreased.
- uE3- Decreased.
- Beta-hCG- decreased.

Positive screening test should be followed by diagnostic test, amniocentesis (15-20 weeks).

Confirmation of Diagnosis

Gold standard test is karyotyping. QF-PCR is preferred over conventional karyotyping because it is reliable, rapid and economical.

Prognosis

~92% of children die in the first year, with full support ~25% may survive 1 year. Survivors have significant neurodevelopmental delay.

Risk of Recurrence:

In a family with a child with complete trisomy 18, the risk of recurrence is usually stated as 1%.⁷

CONCLUSIONS

Though the awareness of prenatal diagnosis is increasing in patients, few cases might reach autopsy without definitive diagnosis. In such cases autopsy plays a pivotal role in diagnosing unexpected anomalies. The findings at autopsy of foetus can confirm the clinical diagnosis and help in identifying unexpected anomalies. It also helps in genetic counseling for future pregnancies.

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