



Edward's syndrome: A clinical approach

Pooja Rani

Department of Pediatric Nursing, Shri Mata Vaishno Devi College of Nursing, SMVDU (Kakryal), Katra, Jammu & Kashmir, India

Abstract

Edward's syndrome, also known as Trisomy 18, is a rare but serious condition. It affects how long a baby may survive. Sadly, most babies with Edward's syndrome die before or shortly after being born. A small number (about 13 in 100) babies born alive with Edward's syndrome live past their 1st birthday and these children often have severe intellectual disability ^[1]. This article presents a case report of newborn baby with Edward's Syndrome.

Keywords: Trisomy 18, blepharophimosis, ocular hypertelorism, amniocentesis

Introduction

Edward's syndrome, termed medically as Trisomy 18 is a type of birth defect which is caused due to the presence of added genetic material i.e., a third copy of the chromosome 18 instead of the natural 2 copies. It is the second most common birth defect after Trisomy 21, i.e., Down's syndrome ^[2].

The live born prevalence is estimated as 1/6,000-1/8,000, but the overall prevalence is higher (1/2500-1/2600) due to the high frequency of fetal loss and pregnancy termination after prenatal diagnosis. The prevalence of Trisomy 18 rises with the increasing maternal age. The recurrence risk for a family with a child with full Trisomy 18 is about 1%.

Types of Trisomy 18

- i. **Full Trisomy 18:** The extra chromosome is in every cell in the baby's body. This is the most common type of Trisomy 18.
- ii. **Partial Trisomy 18:** The child has only part of an extra chromosome 18. That extra part may be attached to another chromosome in the egg or sperm (called a translocation). This type of Trisomy 18 is very rare.
- iii. **Mosaic Trisomy 18:** The extra chromosome 18 is only in some of the baby's cells. This form of Trisomy 18 is also rarely found.

Symptoms

Babies with Trisomy 18 have many serious health problems and physical defects, including: Cleft Lip/Cleft Palate, Blepharophimosis, Ocular Hypertelorism, Ptosis, Choroid Plexus Cyst, Clenched Fists with Overlapping Fingers, Small Jaw (Micrognathia), Unilateral Choanal Atresia, Web Neck, Absence of Distal Crease in Fifth Finger, Defects of Lungs, Kidneys, and Stomach/Intestines, Feeding Problems, Heart Defects, Low-Set Ears, Severe Developmental Delays, Slowed Growth, Small Head (Microcephaly) and Weak Cry.

Diagnosis

A routine prenatal screening or ultrasound is done throughout the 18th to 20th week of pregnancy for diagnosis of Trisomy 18. Amniocentesis and Chorionic villi sampling (CVS) tests can also be done for prenatal genetic diagnosis of the foetus ^[3].

Case Report

A male preterm baby (35 weeks) was delivered through normal vaginal delivery to a multigravida mother aged 37 years on 24th August 2020 at Sai Nursing Home, Delhi. He had mild respiratory distress at the time of birth and needed oxygen by hood. On the very next day he was referred to Guru Teg Bahadur Hospital, Delhi for further treatment. The baby had dysmorphic features like prominent occiput, low set posteriorly rotated pinna, micrognathia, clenched hands and overlapping digits, club foot, unilateral choanal atresia, web neck and absence of distal crease in fifth finger. Treatment was initiated focusing on survival of baby and the sustaining measures were taken when the baby exhibited signs of apnea on the third and fourth day of life which required bag and mask ventilation. Orogastric feeding was started and the baby was discharged on sixth day of life as per the request of the parents.

Discussion

Most of the features presented in the case fitted Edward's Syndrome yet Microcephaly, Cleft Lip/Cleft Palate, Blepharophimosis, Ocular Hypertelorism, Ptosis, Choroid Plexus Cyst and Cryptorchidism were absent in the case. In majority of the cases, death occurs due to apnea and heart abnormalities within first two months. It is impossible to predict the exact expectancy of life in neonate with Edward's Syndrome. Survival beyond a year of life is around 5-10% only. Having a child with Trisomy 18 can sometimes be emotionally overwhelming and it's important

for parents to get support during this difficult time. Organizations such as the Chromosome 18 Registry & Research Society and The Trisomy 18 Foundation are working for Trisomy 18 (Edward's Syndrome) ^[4].

References

1. NHS. Edward's syndrome. Available at: <https://www.nhs.uk/conditions/Edward's-syndrome/>
2. Netmeds. Edward's Syndrome: Causes Symptom and Treatment. Available at: <https://www.netmeds.com/health-library/post/Edward's-syndrome-causes-symptoms-and-treatment>
3. Medline plus. Trisomy 18. Available at: <https://medlineplus.gov/genetics/condition/Trisomy-18/#frequency>
4. Grow by web MD. What is Trisomy 18? Available at: <https://www.webmd.com/baby/what-is-Trisomy-18#2>
5. Medicine net. Trisomy 18 (Edward's syndrome). Available at: https://www.medicinenet.com/Trisomy_18_Edward's-syndrome/article.htm
6. Indian Journal of Continuing Nursing Education. Edward's Syndrome. A case study 2019;20:1.