Edward syndrome (Trisomy 18): A case report

Ezhil Arasi Nagamuthu and Neelala Neelaveni

Department of Pathology, Osmania Medical College, Hyderabad, Andhra Pradesh, India

ABSTRACT

The trisomy 18 syndrome (Edwards syndrome) is an autosomal chromosomal disorder due to the presence of an extra chromosome 18. The main clinical features include prenatal growth retardation, characteristic craniofacial features, distinctive hand posture, nail hypoplasia, short hallux, short sternum and major malformations of heart. One such case at 22-24 weeks of gestation was terminated and its case report was illustrated with the phenotypic features and fetal autopsy. The demonstration of an extra chromosome 18 on the karyotype added to the clinical diagnosis.

Keywords: Trisomy 18; Edwards syndrome; Chromosomal disorder; Rockerbottom feet; Karyotype.

INTRODUCTION

The Trisomy 18 syndrome (Edwards syndrome) is a common autosomal chromosomal disorder due to the presence of an extra chromosome 18. The first reported infants were described in 1960 by Edwards et al. and Smith et al. Trisomy 18 represents the second most common autosomal trisomy syndrome after trisomy 21 (Down syndrome).\(^1,2\) Karunakaran and Pai reported the first case in the Indian literature in 1967.\(^3\)

The syndrome pattern comprises a recognizable pattern of major and minor anomalies, an increased risk of neonatal and infant mortality, and significant psychomotor and cognitive disability. The main clinical features represent the clues for the diagnosis in the perinatal period and include prenatal growth retardation, characteristic craniofacial features, distinctive hand posture (overriding fingers), nail hypoplasia, short hallux, short sternum. Internal anomalies, particularly involving heart are common. The demonstration of an extra chromosome 18, or less commonly a partial trisomy of the long arm of chromosome 18, on the standard G-banded karyotype allows for confirmation of the clinical diagnosis.\([1, 2]\)

A 23 years old Primi gravida with gestational age of 22-24 weeks reported at Modern Government Maternity Hospital/ Osmania Medical College, Hyderabad, Andhra Pradesh, India. The ultrasound scanning showed ventriculomegaly, absent corpus callosum, rockerbottom feet. In view of multiple congenital anomalies, the couple decided to undergo termination of pregnancy. Later, the foetus was sent for autopsy.

On external examination, the foetus showed prominent occiput, lumbar meningocele (Fig. 1), hypertelorism, low set fawn like ears (Fig. 2), rockerbottom feet (Fig. 3), clenched hands with overlapping fingers (Fig. 4).
MATERIALS AND METHODS

After autopsy, the organs were fixed in 10% formalin for processing. After gross analysis representative sections were given for tissue processing. Sections were processed routinely with paraffin embedding and stained with haematoxylin and eosin.

Figure 1: Prominent Occiput & Lumbar meningocele

Figure 2: Hypertelorism & Low set fawn like ears
Figure 3: Rockerbottom feet

Figure 4: Clenched hands with overlapping fingers
Figure 5: Agenesis of right kidney

Fig. 6: Sections of left kidney
The histopathological examination of the sections of the left kidney show normal development of glomeruli and tubules.

![Fig. 7: Cytogenetic map](image)

The cytogenetic work up was done to identify the chromosomal aberration. It showed trisomy 18.

The diagnosis of Trisomy 18 (Edward syndrome) was based on the findings of hypertelorism, prominent occiput, low set fawn like pointed ears, clenched hands with overlapping fingers, rocker bottom foot and ultrasound findings of ventriculomegaly, absent corpus callosum. The cytogenetic karyotype of trisomy 18 confirmed the diagnosis.

### RESULTS AND DISCUSSION

Trisomy 18 is a chromosomal disorder resulting in a syndrome characterized by specific dysmorphic features and organ malformations. Trisomy 18 is also called as Edward Syndrome or Trisomy E.

The live birth prevalence of trisomy 18 ranges from 1/3600 to 1/10,000 with the best overall estimate in livebirths as 1 in 6,000.\textsuperscript{1,2,5} The prevalence at birth is higher in females compared to males (F:M %, 60.4).\textsuperscript{2,5}

It is well known that trisomy 18 pregnancies have a high risk of fetal loss and stillbirth. Currently most diagnoses are made in the prenatal period based on screening by maternal age or maternal serum marker screening and amniocentesis, or detection of sonographic abnormalities during the second and third trimester followed by pregnancy termination in a significant percentage of cases.\textsuperscript{1,2,5}

Genetic counseling: When prenatal or neonatal diagnosis of trisomy 18 is made, the counseling of the family should be realistic, but not desolate. The parents can find it difficult to accept the lack of certainty of the newborn situation, but they have to be prepared for both the probability of death and the possibility of living. The recurrence risk, for a family with a child with complete trisomy 18 is usually stated as 1%.\textsuperscript{2}

### CONCLUSION

The available literature and our experience support the fact that careful pathological examination of foetus can confirm clinical diagnosis and to explain the cause of intrauterine fetal demise.

It helps to identify unexpected anomalies that may provide further clues to a diagnostic syndrome and also assist in family planning, genetic counselling for future pregnancies.

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REFERENCES