Original Research Paper

Volume - 13 | Issue - 06 | June - 2023 | PRINT ISSN No. 2249 - 555X | DOI : 10.36106/ijar

Paediatrics

EDWARDS' SYNDROME (TRISOMY 18) : A CASE REPORT

Dr. Nitish Kumar Post Graduate Student, Dept. Of Paediatrics, Patna medical college and hospital (PMCH), Patna.

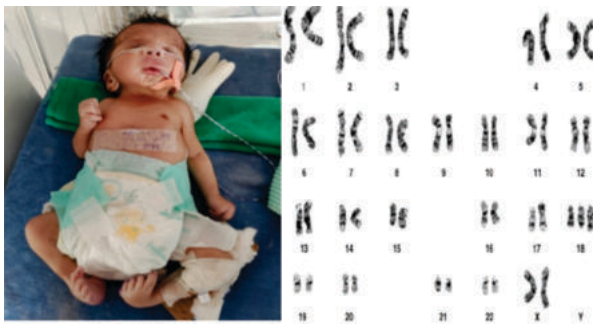
Dr. Gopal Sharan* Professor, Dept. Of Paediatrics, PMCH, Patna. *Corresponding Author

ABSTRACT Edward syndrome is the 2nd most common autosomal trisomy in newborns after down syndrome. These cases are result of maternal nondisjunction of chromosome 18. They have significant structural abnormalities mostly detected on ultrasound. Finding includes cardiac abnormalities , ventriculomegaly, lemon shaped skull, increased nuchal translucency, growth restriction, limb and facial defects. Edward syndrome is almost always fetal. Few infants that survive have severe neurocognitive and physical defects.

KEYWORDS : Ventriculomegaly , clenched hands , rocker bottom feet

INTRODUCTION

A 2.5 kg birth weight male baby with limb and facial deformity was born by normal vaginal delivery to a primi mother at term gestation and was brought on day 18 of life at our SNCU; referred from a hospital in jehanabad , Bihar. On presentation the child was lethargic and in respiratory distress, baby was put on oxygen inhalation by CPAP and maintained Spo2 - 95%. On clinical examination the child had microcephaly , micropthalmia , short palpebral fissures , low set ears . On oral cavity examination - micro retrognathia , microstomia and cleft palate was present. In skeletal exam-baby had short neck , clenched hands with overriding of fingers and thumb hypoplasia . Baby also had narrow pelvis, dislocated hip and rocker bottom feet. There was no history of ANC and antenatal USG during pregnancy. There was no history of any drug intake on any know toxin exposure and no previous IUDS or abortion.



DISCUSSION

Edwards syndrome is an autosomal chromosomal disorder due to extra copy of chromosome 18 . It is one of the autosomal trisomy 2nd in frequency only to trisomy 21. Edward syndrome was first reported by Edward et al in 1960,who reported a neonate with multiple congenital malformation and cognitive deficit . Smith et al , confirmed the extra copy of chromosome 18 as underlying cause for Edward syndrome. Complete trisomy 18 is the most common form (94%) . In this type , every cell contains three copies of chromosome 18. Mosaic trisomy is the second most common type(<5%).Partial trisomy 18 accounts for 2% . In this type only a partial segment of chromosome 18q is present in triplicate . Trisomy 18 has a positive correlation with advancing maternal age . Recurrence risk for complete trisomy 18 is 0.5%to 1% for subsequent pregnancy.

DIAGNOSIS

The evaluation and diagnosis of trisomy 18 begin in the antenatal period. Maternal serum screening can show low levels of alpha feto protein, human chorionic gonadotropin and unconjugated estriol. Serum and genetic markers are more useful when combined with classic ultrasound finding such as increased nuchal translucency . Noninvasive prenatal testing using cell free fetal DNA in maternal plasma has a role in diagnosing trisomy 18 but alone only has 60.7% PPV. Combined with ultrasound, NIPT has a PPV of 100% and NPV of upto 100% by the 2 nd trimester.

PROGNOSIS

- Almost 40% of fetuses die during labour and 1/3 if the surviving fetuses are delivered preterm .
- The median survival for the Edward syndrome ranges from 3 days to 14.5 days
- The survival percentage is 60 - 75% at the first week , 20-40% at one month and 10% at one year.
- 5% - 10% of Edward syndrome patient survive beyond the 1 st year of life .

SUMMARY

Edward syndrome is a genetic condition that causes physical growth delays during fetal development . Life expectancy for children is short due to several life threatening complication . If a parent had a child with Edward syndrome and becomes pregnant again its unlikely they will have another child with same condition . Diagnosis of Edward syndrome begins during pregnancy and conformation occur either before or after baby is born . There is no cure for this condition and so only way to prevent the disease is to diagnose it early prenatally so fetus can be aborted. So regular antenatal checkup along with USG scan are the cornerstone to suspect the disease at the early stage and conformation of diagnosis with either invasive or non-invasive prenatal testing.

REFERENCES

1. Nelson text book of pediatrics,21e;658-664
2. Rasmussen SA,Wong LY, Yang Q May KM ,Friedman JM.population based analysis of mortality in trisomy 13 and 18,pediatrics[pubmed]
3. Cereda A, carey JC(2012) The trisomy 18 syndrome.orphaned J Rare Dis 7:81.