ORIGINAL RESEARCH PAPER
Volume - 13 | Issue - 10 | October - 2024 | PRINT ISSN No. 2277 - 8179 | DOI : 10.36106/ijsr
INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

EDWARD SYNDROME- A PAIR OF CASES

Neonatology

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ABSTRACT

Edward's syndrome or Trisomy 18 is the second most common autosomal trisomy after Down's syndrome (trisomy 21). Prevalence 0f trisomy 18 in live births is around 1 in 6000-8000, however overall prevalence is more 1:2500 to 1:2600 due to high frequency of fetal losses and termination of pregnancy after antenatal diagnosis. The syndrome is characterised by Dysmorphic facies (prominent occiput. Low set ears, Micrognathia), overriding of fingers, rocker bottom foot and multisystemic multiple congenital anomalies. Edward syndrome can be diagnosed antenatally by screening tests like ultrasonography and Non-invasive prenatal testing. Postnatally karyotyping can be sent in suspected Trisomy 18. Following cases were diagnosed postnatally with the help of clinical features and karyotyping. Both the cases discussed below were born with cardiovascular defects and talipes equinovarus and other associated anomalies and both of them succumbed to respiratory distress within 1st week of life. Babies born with Edward have very poor prognosis, 50% of babies born with Edward syndrome die within First week of life and 90-95% do not survive beyond the 1st year of life. These case reports reflect the pressing need for the through active foetal surveillance antenatally for recognition of defects in early stage as no treatment option is yet available for this complex syndrome.

KEYWORDS

Edwards Syndrome, Trisomy, overriding of fingers, omphalocele.

INTRODUCTION:

Trisomy 18 also known as Edward's syndrome, is an autosomal hereditary syndrome. [1] Prevalence 0f trisomy 18 in live births is around 1 in 6000-8000, however overall prevalence is more 1:2500 to 1:2600 due to high frequency of fetal losses and termination of pregnancy after antenatal diagnosis. [2] There is 3:1 female predominance as most males die during pregnancy.[3] This syndrome is characterised by multiple congenital anomalies affecting multiple systems. [4] Natural history of Edward's syndrome includes a limited survival rate of 10% and those who survive are mentally retarded. [5] [6] [7] Even though there is no definitive treatment option for Edwards syndrome, there are some conservative management options tailored to each patient based on parental needs and choices. Recently the American academy of paediatrics changed its guidelines and does not recommend withholding neonatal resuscitation to patients with Edwards syndrome. Tube feeding done for neonatal patients and gastrostomy for older patients. diuretics and digoxin recommended for heart failure and palliative or corrective cardiac surgeries recommended for cardiovascular defects. infections managed with standard approach and psychiatric intervention given wherever psychosocial support is needed. [8] We report such 2 cases who were born with multiple congenital anomalies and succumbed in the first week of life.

Case Details:

Case 1: A male child born out of non-consanguineous marriage with birth order of 3 out of three children was referred to us on the first day of life. Parents had no significant medical history with a maternal age of 24 years and no significant family history was noted on either side of the family. Baby was full-term, 40 weeks gestation, delivered by LSCS (Low segment caesarean section) in view of previous two LSCS and antenatal scans suggestive of uteroplacental insufficiency and omphalocele. He had perinatal asphyxia with a birth weight of 1.37 kg. On examination, the baby was sick, lethargic, and respiratory distress was present with an APGAR score of 4. Anthropometry showed that the baby was small for gestational age. Baby had dysmorphic facies in the form of low set ears, dolichocephaly. He also had multisystem congenital malformations like complex congenital heart disease (double outlet right ventricle (DORV), large inlet ventricular septal defect (VSD), myxomatous thickened valves with significant prolapse and small patent ductus arteriosus (PDA)) which were diagnosed with 2D ECHO, Omphalocele, Bilateral congenital talipes equinovarus (CTEV) with partial syndactyly of toes, overriding of fingers with interphalangeal joint contractures, bilateral cryptorchidism. (Figure 1, 2, 3, 4, 5, 6) Clinical diagnosis of Trisomy 18 was made. Ultrasonography (USG) Skull was normal. Parents were counselled regarding the diagnosis; its genetic basis and poor survival rates and written consent was taken for genetic karyotyping for diagnosis. Other blood parameters were within normal limits. Baby started on tube feeds along with intravenous fluids, shifted to paediatrics surgery for surgical correction of omphalocele but succumbed to death post surgery. Karyotyping later confirmed the diagnosis of Edward syndrome.

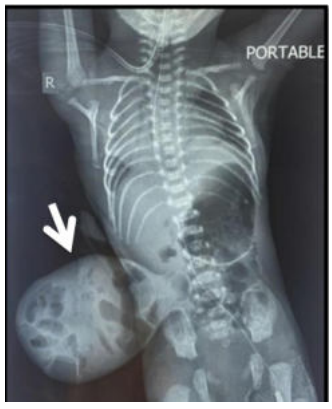

Fig 1: Babygram showing giant omphalocele.

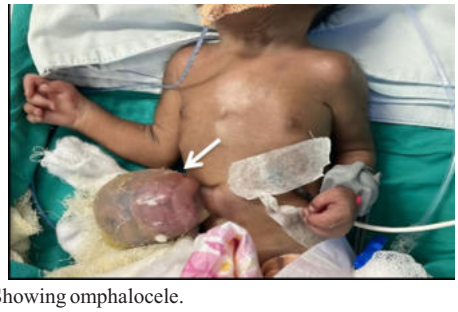

Fig 2: Showing omphalocele.

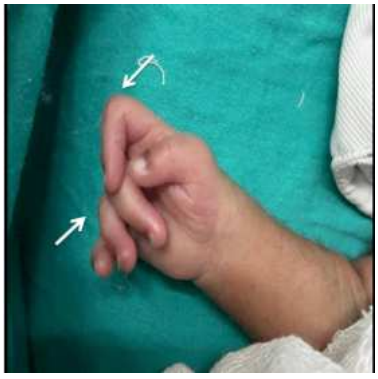

Fig 3: Showing overriding of fingers with interphalangeal joint contractures.

Fig 4: Showing Bilateral congenital talipes equinovarus (CTEV).

Fig 5, 6: Dysmorphic facies in the form of low set ears, dolichocephaly.

Case 2: A male child born out of non-consanguineous marriage with birth order of 2 out of two children, at 39 weeks of gestation with nil significant family history and nil significant medical history in parents with maternal age of 27 years. Antenatal scans were suggestive of polyhydramnios, skeletal dysplasia and giant cisterna magna. Baby had perinatal asphyxia. Baby had an APGAR score of 5.

Baby had facial dysmorphism in the form of microcephaly, sloping forehead, hypertelorism, flat nasal bridge, microtia, low set ears, retrognathia and micrognathia. Right upper limb was deformed with bowing of the forearm, bilateral short forearms with overriding of fingers was present. Bilateral CTEV along with bilateral cryptorchidism was seen. (Figure 7, 8, 9, 10) Cardiovascular system had complex congenital heart disease (large Perimembranous Ventricular septal defect, large Patent ductus arteriosus with biventricular systolic dysfunction with severe pulmonary arterial hypertension (PHA). Baby started on intravenous fluids, tube feeds and oxygen by hood.

The USG skull and abdomen were normal. Parents were counselled regarding the condition and consent taken for karyotyping. Baby gradually deteriorated and expired on the third day of life. Karyotyping revealed Trisomy 18. Comparison of clinical features of both babies is given in Table 1.

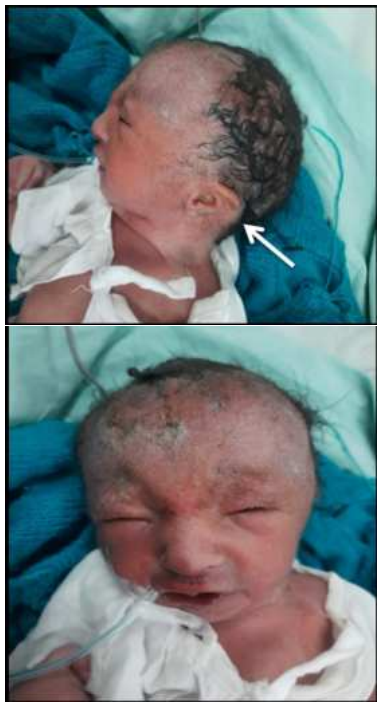

Fig 7, 8: Dysmorphism in the form of sloping forehead, hypertelorism, flat nasal bridge, microtia, low set ears, retrognathia and micrognathia.

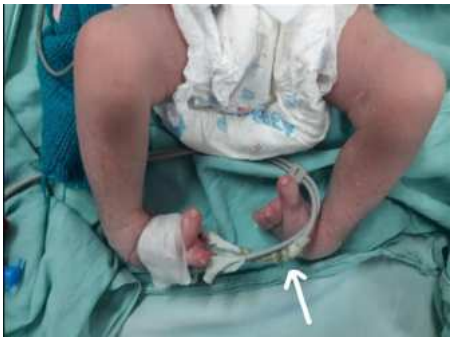

Fig 9: Showing Bililateral CTEV

Fig 10: Showing bilateral short forearms with overriding of fingers, deformed right upper limb with bowing of the forearm.

Discussion: John Hilton Edwards, et al., discovered Edwards syndrome (trisomy 18) in 1960. [1] The syndrome is female predominant as most of male die before birth, which is in contrast to our case reports as both babies were males. [3] Advanced maternal age is linked with this syndrome, however in the present cases maternal age was found to be less than 30 years in both cases. [5] [9] This syndrome is associated with small placenta, polyhydramnios and decreased foetal movements with low birth weight, weak cry or no cry at birth and difficulty in swallowing along with muscular hypertonia and

hypoplasia of the skin and subcutaneous tissues.[4] Children with trisomy 18 may have small head(microcephaly), short stature, mental retardation, cranio-facial abnormalities such as a small face, prominent occiput, micrognathia, cleft lip/cleft palate, ocular hypertelorism, small mouth; limb abnormalities including overlapping fingers, nail hypoplasia, hip abnormalities, short sternum, underdeveloped thumbs, absent radius, webbing of the second and third toes, clubfoot, rocker bottom feet. [10] [11] In 80 to 100% cases structural cardiovascular abnormalities are found with VSD being most common, underdevelopment of reproductive organs in 50%, horseshoe kidney in 32%, omphalocele in 14%, diaphragmatic hernia in 11% babies and oesophageal atresia has been reported with a rate of 11%. In the reported cases, facial dysmorphism, cardiovascular, gastrointestinal, central nervous system abnormalities were present. Finger and limb abnormalities were also noted.[12]

A case of Edward's syndrome which was diagnosed post mortem with genetic test was described by Gerakove et al in 2022. It was a female child which survived for 26 days after birth and succumbed to respiratory distress in the end. She had malformed ear lobes, micrognathia, high arched palate, equinovalgus on bilateral feet with contracture of fingers which were similar to our case reports. However, she also reported tumour formation in the submandibular region, duplicated collecting system in kidney with rectovaginal fistula which were not seen in our cases.[13] A case of Edwards syndrome which was diagnosed antenatally with USG and amniocentesis was reported by Mudaliyar et al in 2017. The authors therefore recommend an active foetal surveillance to recognise the defects as early as possible so that medical termination of pregnancy can be conducted in the initial phase. [14] Our cases mentioned above were referred to us after birth with severe defects in multiple systems and conservative management in the form of tube feeding and mechanical ventilatory support was administered to both the children and one child was offered paediatrics surgery for immediate management of omphalocele. However, survival duration was limited to one and three days respectively rendering it difficult for any other management options.

CONCLUSION:

Most of the cases of Edward syndrome are diagnosed prenatally. However, in cases where antenatal diagnosis is missed, careful clinical examination along with karyotyping is useful in diagnosis and intervention. Microcephaly, short stature, mental retardation, cranio-facial abnormalities such as a small face, prominent occiput, micrognathia, cleft lip/cleft palate, ocular hypertelorism, small mouth; limb abnormalities including overlapping fingers, camptodactyly, nail hypoplasia, hip abnormalities, short sternum, underdeveloped thumbs, absent radius, webbing of the second and third toes, clubfoot, rocker bottom feet, CVS defects are commonly associated with Edwards syndrome. Majority of neonates born with Edward syndrome die in early infancy. We also recommend for thorough antenatal scans as there is a need for early recognition in such cases and genetic counselling may be needed if such parents opt to conceive again.

Conflicts of interest: Nil

Acknowledgement: We thank Dr Surbhi Rathi (HOD Pediatrics) and Dr Pravin Rathi (Dean) for supporting us.

Table 1: comparison of 2 cases

Finding	Case 1	Case 2
Abnormal antenatal scan	Yes	Yes
Required resuscitation at birth	Yes	Yes
Low birth weight	Yes	Yes
Hypotonia	Yes	Yes
Dysmorphism facies	Yes	Yes
Microcephaly	Yes	Yes
Cardiovascular malformation	Yes	Yes
CNS malformation	Not detected	Yes
GI malformation	Yes	No
Undiscendend testis	Yes	Yes
Limb and finger malformation	Yes	Yes
Contractures of fingers	Yes	No

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