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Edward's Syndrome-A Case Report

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Abstract:

Edward's syndrome is commonly known as the trisomy 18 syndrome. It is a devastating genetic disorder that can be characterized by multiple congenital anomalies. About 50% babies with this syndrome do not survive beyond one week of age and approximately 95% does not survive past the first year of life. The feature of Edward's Syndrome involves in craniofacial, skeletal, cardiovascular, central nervous system and genitourinary malformations. The mortality rate among infants with trisomy 18 is high as a result of cardiac and renal malformations, feeding difficulty, sepsis and central nervous system defects. Here we report a neonate with Edward's syndrome.

Keywords: *Edward's syndrome; Trisomy 18; Genetic disorder; Mortality rate.*

Introduction:

Edward's syndrome is a rare chromosomal disorder due to presence of extra chromosome 18 that effects on multiple organs.[1] Trisomy 18 is the second most common autosomal trisomy syndrome after trisomy 21. It is well known that trisomy 18 pregnancies have a high risk of fetal loss and stillbirth.[2] The live birth prevalence is form 1/3600 to 1/1000, female to male ratio is approximately 3:1.[3-4] It is the second most common autosomal trisomy that carries to term. Edward's Syndrome is a genetic condition almost always results from non-dysfunction during meiosis. It was first diagnosed in 1960 by Edwards et al. and Smith et al. by specific dysmorphic features.[5] In Edward's syndrome, most common abnormalities were found in cardiovascular system, upper and lower extremities, urinary system, head and neck, gastrointestinal tract and genitals. The cardiovascular system associated with ventricular septal defect, atrial septal defect and patent ductus arteriosus.[6] The survival rate of Edward's syndrome was a very low which results from heart abnormalities, kidney malformations, and other internal organ disorders.

Case Report:

A one day old female newborn, first issue of her non-consanguineous parents presented in the neonatology department of Chattagram Maa-O-Shisu General Hospital with the complaints of small for gestational age and breathing difficulty since birth. Regarding maternal history, mother's age was 24 year, she was on regular antenatal checkup and immunized. There was no history of any abortion, gestational diabetics-mellitus, hypertension fever, rash and jaundice. Baby was delivered at term by caesarian section uneventfully. On examination, baby was lethargic, dyspneic, dysmorphic in appearance like small face, small chin, low set ear, microophthalmia. Vitals were normal except respiratory rate which was 68 breaths/ min. Birth weight was 1800 gm. which corresponded below 10 centile, occipital frontal circumferences was 32 cm, which corresponded below 10 centile. Nasal and oral cavity was normal. There was overlapping of index finger over the 3rd finger of hands, Rocker bottom feet was present. On chest examination, small and wide apart nipple, short sternum, intercostal recession was present. Apex beat was situated in left 4th intercostal space along the midclavicular line, S_1 and S_2 were audible in all four areas. There was pansystolic murmur over the whole precordium but best heard at left lower sternal border, grade 2/5, no

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Fig.-1: *Female baby with trisomy 18 small face, low set ear, microphthalmia, microgan- thia, clenchedfist, hypertonic cris-cross and rockerbottom feet.*

Fig.-2: *Trisomy18; Overlapping of fingers and hypoplastic nails.*

radiation. Breath sound was vesicular in nature. Abdominal examination, size and shape of abdomen was normal; umbilicus was healthy and centrally placed. There was no organomegaly, genitalia was normal and anus was patent. On locomotors examination there were limited abduction of hip, hypertonic crisscross and rocker bottom feet, back and spine was normal with preserved reflexes.

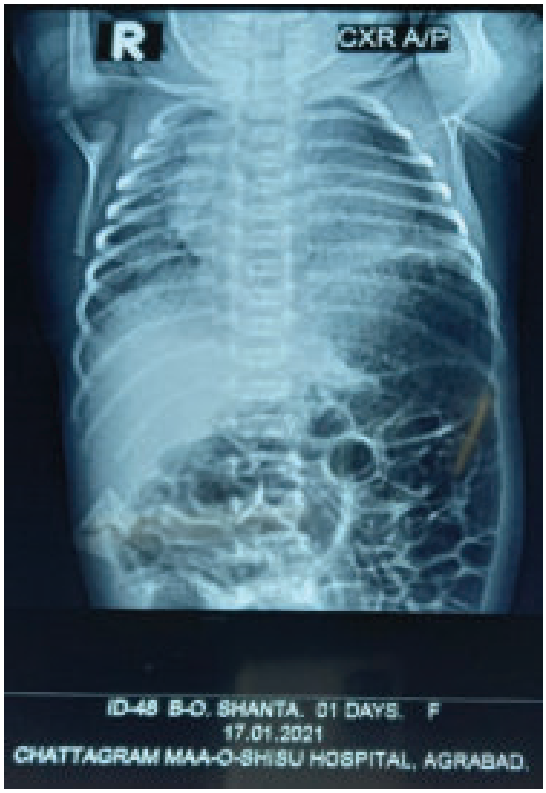

Fig.-3: *Chest X-ray shows Cardiomegaly*

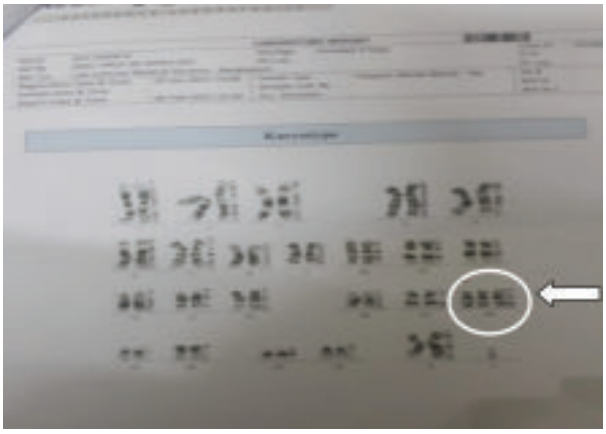

Fig.-4: *karyotype reveals 47 chromosomes instead of 46, with an extra chromosome in pair18.*

Table-I
Investigation Profile of the infant

Test	Result
Hb%	16.3gm/dl
Total count	13,900/cmm
Differential count	
Neutrophil	77%
Lymphocyte	18%
Platelet count	1,50000/cmm
C-reactive protein	<5mg/L
Serum Electrolyte:	
Sodium	140mmol/L
Potassium	4.0mmol/L
Chloride	105mmol/dl
HCO_3	28.5mmol/dl
Random blood sugar	150mg/dl
Serum Calcium	7.7mg/dl
Serum creatinine	0.7mg/dl
Blood culture	No growth
Arterial blood gas analysis	Normal
Chest X-ray	Cardiomegaly
Echocardiography	Large ASD with moderate PDA with moderate VSD with moderate to severe pulmonary hypertension.
USG of abdomen	Normal abdominal scan
Karyotyping	47,XX,+18(Trisomy 18)

Discussion:

Edward's syndrome is the second most common autosomal trisomy in live born children after trisomy 21. The exact cause is of trisomy 18 is not known, the incidence increases as the mother's age increases. In this case maternal age was found to be less than 25yrs. This syndrome also has maternal history of polyhydramnions and less fetal movements. Trisomy 18 is classified into three possible types, non-disjunction, mosaicism and unbalanced translocation. When Edward's syndrome is caused by an entire extra chromosome 18 this is called a "primary trisomy", which is a non-inherited version of Edward's syndrome. The another rare type is an "unbalanced translocation", this happens when an extra portion of chromosome 18 is attached to part of another chromosome. This can occur because one of the baby's parents carries what is known as a 'balanced translocation'.[7] This type has an extra chromosome in all body cells and occurred at the time of fertilization.[8] In non-disjunction type, the cell division have occurred unequally, eggs of sperms' chromosome 18 did not separate correctly. The deficiency probably occurred before conception during the formation of the egg or sperm and is therefore not thought to result from any environmental impacts during pregnancy. There are no associations found with alcohol, tobacco or drug use before or during pregnancy.

Edward's Syndrome has some common physical features in all babies but the presence of some features and congenital anomalies varies. Infants with trisomy18 are usually small for gestational age has typically recognizable physical findings. They may have microcephaly, short stature, mental retardation, cranio-facial abnormalities such as a small face, prominent occiput, microganthia, cleft lip/cleft palate, upturned nose, narrow palpable fissures, small mouth; limb abnormalities as well as clenched fist with index finger overlapping third finger fourth finger and small fingernails and toe nails, under developed or altered thumbs, absent radius, short sternum, narrow hip with limited abduction, webbing of second and third toes, club foot or rocker-bottom feet.[9,10] Although the prognosis of the disease do not depend upon the physical findings, they do provide clues that suggest the diagnosis. In 97% of trisomy 18 cases, structural disorders are found at least in three organs.[11] More than 90% cases, there is associated occurrence of a congenital heart defect. Most infants have a ventricular septal defect, or atrial septal defect or patent ductus arteriosus. About 10% have a double outlet right ventricle and a hypo plastic left heart.[6] In about 50% cases, there is under development of reproductive organs, horseshoe kidney in 32%, omphalocele in 14%, diaphragmatic hernia in 11% babies and esophageal atresia has been reported with a rate of 11%.[12]

In this reported case, facial dysmorphism, hyper tonicity, deformities of fingers and limb, cardiovascular abnormalities were present. Bowel bladder abnormalities were absent. Survival in trisomy 18 is related to the severity of congenital malformations and to some extent, the accessibility of pediatrics care.

Subsistence into childhood or beyond is rare. In few children who do survive past early infancy, profound mental deficiency, limited social response, significant developmental delay are present. Prenatal screening should be offered, especially after the birth of an affected child.

Conclusion:

Edward's syndrome is a rare chromosomal disorder. The clinical features and karyotyping were essential for diagnosis and prognosis. Different congenital anomalies should be searched out to manage accordingly.

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