Indian Journal of Genetics and Molecular Research
Volume 2 Number 1, January - June 2013

Edward Syndrome

Sunil Mhaske*, Ramesh B. Kothari, Sandeep Deokate***, Ram Sethi***, Nishad Patil***, Pavan Suryawanshi***, Rahul Maski***, Neha Sharma******

Abstract

Edward syndrome is a genetic disorder caused by presence of all or part of an extra 18 chromosome. It is the 2nd most common autosomal trisomy after Down syndrome.

Keywords: Edward's syndrome; Trisomy18; TrisomyE.

Introduction

Edward syndrome occurs in around 1 in 6000 live births & around 80% of those affected are females. The majority of fetuses with the syndrome die before birth. The incidence increases as the mother's age increases. The syndrome has a very low rate of survival, resulting from heart abnormalities, kidney malformations & other internal organ disorders.

Case summary

20 years old rural habitat primi with 34 weeks gestation was admitted to our labour room with pain in abdomen since 1 day. Pain was intermittent in nature, radiating to back and medial aspect of thigh. She had history of threatened abortion for which she took treatment details of which are not available. She was referred to our hospital by a private practitioner.

According to her last menstrual cycle, her gestational age was 34 weeks. She was a registered case at PHC with 3 antenatal visits. She had taken iron and folic acid tablets with 2 doses of TT injection. She had a family history of 3rd degree consangious marriage. There was no any history of previous abortions.

On examination, she was an averagely built female with mild pallor. Pulse was 92/minute & Blood pressure being 130/80 mmHg. Per abdominal examination showed uterus of 34 weeks with fetus of breech presentation. Per vaginal examination showed 6 cm dilated, 60% effaced cervix with intact membranes without any PV bleeding.

Antenatal USG showed fetus with 34 weeks gestational age with breech presentation, massive polyhydramnios, IUGR with fetal oesophageal atresia. Mother's investigations revealed Hb - 9gm%, TLC - 15800/cumm, Platelet count - 2.24 lacs/cumm. Liver function tests and renal function tests were within normal limits. VDRL, HbsAg and Tridot were negative. Urine showed traces of albumin.

A male baby weighing 2 kgs was delivered by vaginal route with right medio lateral episiotomy. Baby did not cry immediately after birth. His heart rate was 134/minute. Respiration was non-spontaneous. Cyanosis was present. He was intubated with endotracheal tube of internal

diameter 3 mm & put on AMBU bag ventilation. Baby turned to pink color after 10 minutes of AMBU. Reflexes were absent. His

Author's Affiliation: *Professor & Head Dept of Peadiatrics; **Assistant Professor; ***Residents; ****Intern, Padmashri Dr. Vithalrao Vikhe Patil Medical College & Hospital, Near Govt. Milk Dairy, Vilad Ghat, Ahmednagar - 414111.

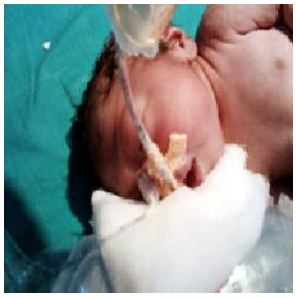
Corresponding Author: Dr. Sunil Mhaske, Professor & Head Dept of Peadiatrics, Padmashri Dr. Vithalrao Vikhe Patil Medical College & Hospital, Near Govt. Milk Dairy, Vilad Ghat, Ahmednagar - 414111. E-mail: sunilmhaske@rocketmail.com

length was 36 cm, head circumference – 28 cm (< 3rd percentile), chest circumference – 26 cm. Placenta and cord were normal. The baby was shifted to NICU & was kept on artificial respiration with IPPV mode of ventilation & FiO_2 being 100 %. Baby was treated with antibiotics, IV fluids and adrenaline as required. The baby died after 3 days being on ventilatory support despite of best resuscitative efforts.

Microcephaly

Simian crease

Mongoloid slant



Following investigations were done:

Hb – 14 gm/dl, TLC - 15600/cmm, DLC – P: 42, L: 54, E: 02, M: 02, B: 00.

Platelets – 2.5 lacs/cmm Blood group: B – Rh+ve.

USG Cranium: Choroid plexus cyst with underdeveloped brain.

USG Abdomen and Pelvis: Bilateral inguinal hernia undescended testis.

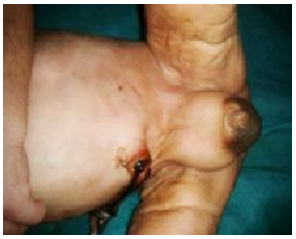
2D-ECHO revealed moderate VSD with left to right shunt.

Clinical features are listed as follows:

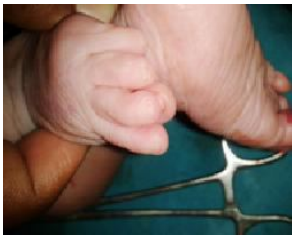
Clinical features	Edward Syndrome	Down Syndrome	Our patient
- Microcephaly	+	+	+
- Hypertelorism	+	-	+
- Mongoloid slant	-	+	+
- Low set ears	+	+	+
- Microphthalmia	-	-	+
- Depressed nasal bridge	-	-	-
- Simian crease	+	+	+
- Clenched hands with index finger overlapping 3rd finger & 5th finger overlapping 4th finger	+	+	+
- Short dorsiflexed 1st toe	+	-	-
- Short sternum	+	-	+
- Rocker bottom feet	+	+	+
- Inguinal hernia	+	+	+
- Oesophageal atresia	+	+	+
- Congenital heart defect (VSD, PDA, ASD)	+	+	+
- Cleft lip, cleft palate	+	-	-
- Hypotonia or Hyperflexibility	-	-	+
- Micropenis	-	+	+
- Mental subnormality	+	+	-
- Open mouth with protruding tongue	-	+	-
- IUGR	+	+	+

Micropenis

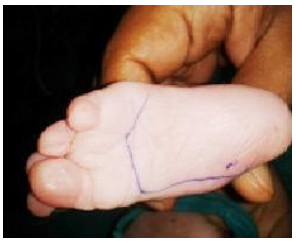
Hypotonia or Hyperflexibility



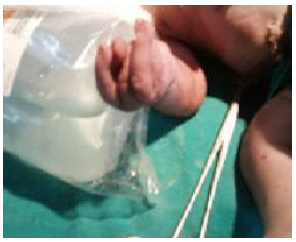
Overlapping of toes



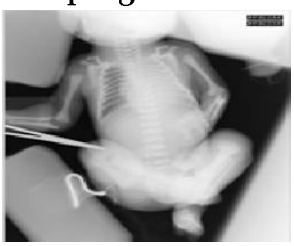
Short 1st toe



Clenched hands with index finger overlapping 3rd finger & 5th finger overlapping 4th finger



Esophageal atresia



Discussion

It is named after Dr. John H. Edward who 1st described the syndrome in 1960. Edwards served as professor of human genetics at Birmingham University from 1969 to 1979 and at Oxford University from 1979 to 1995. He was the author of *Human Genetics* (1978) as well as numerous papers on a variety of topics in the field.

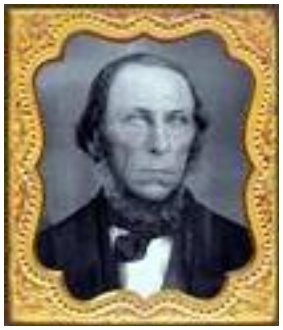
Signs and symptoms

Children born with Edward syndrome may have some or all of the following characteristics:

Kidney malformations, Structural heart defects at birth (i.e., ventricular septal defect, atrial septal defect, patent ductus arteriosus), Intestines protruding outside the body (omphalocele), Esophageal atresia, Mental retardation, developmental delays, growth deficiency, feeding difficulties, breathing difficulties, and arthrogryposis (a muscle disorder that causes multiple joint contractures at birth).

Some physical malformations associated with Edwards syndrome include small head (microcephaly) accompanied by a prominent back portion of the head (occiput); low-set, malformed ears; abnormally small jaw (micrognathia); cleft lip/cleft palate; upturned nose; narrow eyelid folds (palpebral fissures); widely spaced eyes (ocular hypertelorism); drooping of the upper eyelids (ptosis); a short breast bone; clenched hands; choroid plexus cysts; underdeveloped thumbs and or nails, absent radius, webbing of the second and third toes; clubfoot or Rocker bottom feet; and in males, undescended testicles.

Dr. John H. Edward



In utero, the most common characteristic is cardiac anomalies, followed by central nervous system anomalies such as head shape abnormalities. The most common intracranial anomaly is the presence of choroid plexus cysts, which are pockets of fluid on the brain. These are not problematic in themselves, but their presence may be a marker for trisomy18. Sometimes excess amniotic fluid or polyhydramnios is exhibited.

Genetics

Edward syndrome is a chromosomal abnormality characterized by the presence of an extra copy of genetic material on the 18th chromosome, either in whole (trisomy 18) or in part (such as due to translocations). The additional chromosome usually occurs before conception. The effects of the extra copy vary greatly, depending on the extent of the extra copy, genetic history, and chance. Edward syndrome occurs in all human populations but is more prevalent in female offspring.

Trisomy 18 (47, XX, +18) is caused by a meiotic nondisjunction event. With nondisjunction, a gamete is produced with an extra copy of chromosome 18; the gamete thus has 24 chromosomes. When combined with a normal gamete from the other parent, the embryo has 47 chromosomes, with three copies of chromosome 18.

A small percentage of cases occur when only some of the body's cells have an extra copy of chromosome 18, resulting in a mixed population of cells with a differing number of chromosomes. Such cases are sometimes called mosaic Edward syndrome. Very rarely, a piece of chromosome 18 becomes attached to another chromosome (translocated) before or after conception. Affected individuals have two copies of chromosome 18 plus extra material from chromosome 18 attached to another chromosome. With a translocation, a person has a partial trisomy for chromosome 18, and the abnormalities are often less severe than for the typical Edwards syndrome.

Diagnosis

Edward syndrome may be diagnosed at birth by the physical abnormalities characteristic to the syndrome. In addition, physical examination of the infant may show arched fingerprint patterns, while x-rays may show a short sternum. Definitive diagnosis is achieved through karyotyping. Using special stains and microscopy, individual chromosomes are identified, and the presence of an extra chromosome 18 is revealed.

Edward syndrome can be detected before birth. If a pregnant woman is older than 35, has a family history of genetic abnormalities, has previously conceived a child with a genetic abnormality, or has suffered earlier miscarriages, she may undergo tests to determine whether her child carries genetic abnormalities. Tests include maternal serum alpha-fetal protein analysis or screening, ultrasonography, amniocentesis, and chorionic villus sampling.

In addition, a pregnant woman carrying a child with Edward syndrome may have an unusually large uterus during pregnancy, due to the presence of extra amniotic fluid. In addition, an unusually small placenta may be noted during birth.

Treatment

There is no cure for Edward syndrome. 90 to 95 % of all babies born with it die within a year of birth. The few infants that do survive need special treatment ranging from muscular

therapy to nervous system and skeletal corrections for their various handicaps.

Prognosis

In case of Edward syndrome, major causes of death include apnea and heart abnormalities. It is impossible to predict an exact prognosis during pregnancy or the neonatal period. Half of infants with this condition do not survive beyond the first week of life. The median lifespan is 5–15 days. About 8% of infants survive longer than 1 year, One percent of children live to age 10, typically in less severe cases of the mosaic Edward syndrome.

References

1. Edward syndrome (John Hilton Edwards). WhoNamedIt.com. Retrieved 2008-07-24[abc] "Health: Edward syndrome". BBC. Retrieved 2012-01-04.
2. What is Trisomy 18? Trisomy 18 Foundation. Retrieved 2008-07-24.[b]"Trisomy18". Medline. Retrieved 2008-07-24.
3. Hurt K, Sottner O, Záhumenský J, *et al.* Choroid plexus cysts and risk of trisomy 18. Modifications regarding maternal age and markers (in Czech). *Ceska Gynekol.* 2007; 72(1): 49–52. PMID 17357350.
4. Chen, MD, Harold. Introduction to Trisomy 18. *EMedicine.* Retrieved 2008-07-24.
5. For a description of human karyotype see Mittleman, A. (editor). 1995. An International System for Human Cytogenetic Nomeclature. Retrieved 2006-06-04.
6. National Down Syndrome Cytogenetic Register Annual Reports 2008/09.
7. Trisomy 18: MedlinePlus Medical Encyclopedia. Nlm.nih.gov. 2011-12-14. Retrieved 2012-01-04.
8. Rodeck, Charles H, Whittle, Martin J. *Fetal Medicine: Basic Science and Clinical Practice.* Elsevier Health Sciences. 1999. ISBN 0-443-05357-X.
9. Mitchel L. (March 1, 2003). "Trisomy 13 survival can exceed 1 year". OB/GYN News. Retrieved 2008-07-24.