

## **Case Report**

# **A Case Report of Edward Syndrome and Review of 152 Similar Cases Published in Various Journals**

**Satish Arakeri<sup>\*1</sup>, Uroos Fatima<sup>2</sup>, Ramkumar KR<sup>3</sup>, Noori Khalid<sup>4</sup>**

<sup>1</sup>Assistant Professor, Department of Pathology, DMWIMS, Naseera Nagar, Meppadi (PO), Kalpetta, Wayanad, Kerala -673577, India

<sup>2</sup>Associate Professor, Department of Pathology, DMWIMS, Naseera Nagar, Meppadi (PO), Kalpetta, Wayanad, Kerala -673577, India

<sup>3</sup>Professor & HOD, Department of Pathology, DMWIMS, Naseera Nagar, Meppadi (PO), Kalpetta, Wayanad, Kerala -673577, India

<sup>4</sup>Assistant Professor, Department of Obstetrics and Gynaecology, DMWIMS, Naseera Nagar, Meppadi (PO), Kalpetta, Wayanad, Kerala -673577, India

### **\*Corresponding author**

Dr. Satish Arakeri

**Email:** [drsatisharakeri@gmail.com](mailto:drsatisharakeri@gmail.com)

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**Abstract:** First described by Edward in 1960, this is the second most common autosomal trisomy after Down's syndrome. Incidence of Edward syndrome varies from 1 in 3500 to 1 in 7000. All the cases has been collected from internet published in various journals and are available as full article free to access and refer. One case is from our institution. Total 152 cases of Edward syndrome has been reviewed which are proved by cytogenetic study. The male to female ratio is 1.3:1. In more than 50% of cases, both maternal and paternal age is less than 30 years. Most common abnormality associated with cardiovascular system followed by extremities, urinary system, head and neck, gastrointestinal tract and genitals. In conclusion, Edward syndrome is a rare genetic disorder associated with multisystem involvement. Hence, data collection and frequent review of cytogenetically proven cases is must to study in detail about the association of genetic defect and organ system involved. It is also helpful to compare with other known genetic disorder.

**Keywords:** Edward syndrome, Review, Trisomy 18

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## **INTRODUCTION [1-3]**

First described by Edward in 1960, this is the second most common autosomal trisomy after Down's syndrome. Incidence of Edward syndrome varies from 1 in 3500 to 1 in 7000. Edward's syndrome is a condition which is caused by an error in cell division, known as meiotic disjunction.

### **Types of Trisomy 18**

#### **Full Trisomy 18**

The most common type of Trisomy 18 (occurring in about 95% of all cases) is full Trisomy. With full Trisomy, the extra chromosome occurs in every cell in the baby's body. This type of trisomy is not hereditary.

#### **Partial Trisomy 18**

Partial trisomies are very rare. They occur when only part of an extra chromosome is present. Some partial Trisomy 18 syndromes may be caused by hereditary factors. Very rarely, a piece of chromosome 18 becomes attached to another chromosome before or after conception. Affected people have two copies of

chromosome 18, plus a "partial" piece of extra material from chromosome 18.

#### **Mosaic Trisomy 18**

Mosaic trisomy is also very rare. It occurs when the extra chromosome is present in some (but not all) of the cells of the body. Like full Trisomy 18, mosaic Trisomy is not inherited and is a random occurrence that takes place during cell division.

As children with Down syndrome can range from mildly to severely affected, the same is true for children with Trisomy 18. This means that there is no hard and fast rule about what Trisomy 18 will mean for the child. However, statistics show that there is a high mortality rate for children with Trisomy 18 before or shortly after birth.

## **CASE REPORT**

One case is from our institution. The case detail as follows. A 36 year old primi gravida, conceived spontaneously after a married life of 6 months,

presented at 18 weeks with anomaly scan showing features suggestive of posterior urethral valve and borderline oligohydramnios. There was no history of consanguineous marriage. No history of chromosomal anomalies in the family. No history of any miscarriages.

Clinical examination revealed uterine size corresponding to 18 weeks. Level ii scan done showed single live fetus of 17 weeks 5 days, distended bladder and posterior urethra giving keyhole appearance, possibility of posterior urethral valve. Amniotic fluid index was 8 cm.

A level iii targeted scan was advised which revealed multiple fetal anomalies in the form of diaphragmatic hernia, posterior urethral valve.

Fetal karyotyping was done after amniocentesis which revealed trisomy 18 (Edwards’s syndrome).

Autopsy of the fetus has been done. It shows the following gross features.

- Low set ears, Micrognathia
- Left sided poster lateral diaphragmatic defect with herniation of intestine, spleen and left lobe of liver into the chest cavity.
- Mediastinal contents shifted to right side of the thoracic cavity.
- Gross abdominal distension present
- Bilateral kidneys grossly normal
- Bilateral dilated tortuous ureter
- Dilated urinary bladder with marked thinning of its wall and contained straw colored fluid.
- Posterior urethral valve present
- Bilateral testis identified in the lower part of abdomen.
- Imperforate anus
- Left foot shows talipes valgus defect



**Fig. 1: Fetus with low set ears with cubitus valgus defect left foot**



**Fig. 2: Fetus showing diaphragmatic defect with herniation of intestine in left sided chest cavity and distended urinary bladder**

**CASE REVIEW**

All the cases has been collected from internet published in various journals and are available as full article free to access and refer. One case is from our institution.

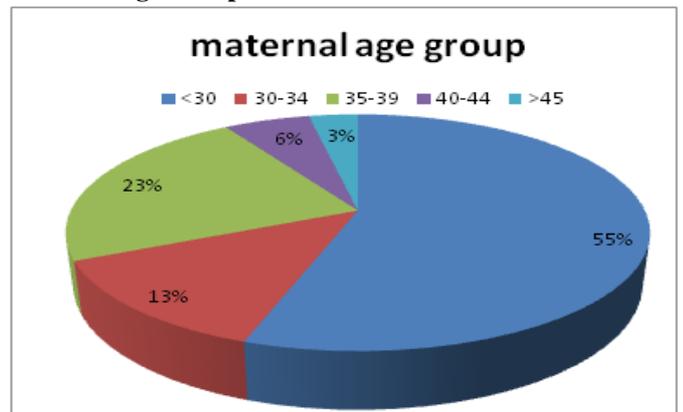
**OBSERVATION [4-9]**

Total number of cases reviewed: 152 cases

**Table 1: Details of number of cases reviewed from various authors article**

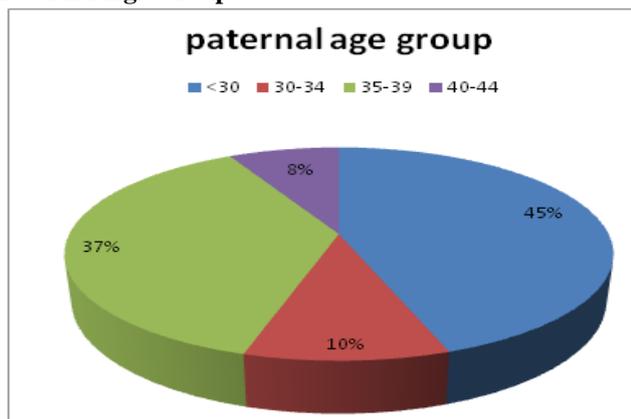
Authors	Number of cases
Naquin KK <i>et al.</i> [5]	118
Taylor AI [4]	27
Bhat BV <i>et al.</i> [6]	03
Barani K and Padmavathy R [7]	01
Patra S <i>et al.</i> [8]	01
Bhanumathi B <i>et al.</i> [9]	01
Our institution case	01
Total	152

**Maternal Age Groups**



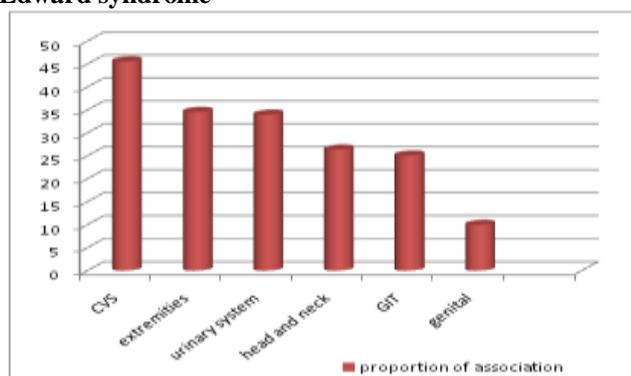
**Fig. 3: Pie chart of various maternal age groups**

**Paternal Age Groups**



**Fig. 4: Pie chart of various paternal age groups**

**Most common abnormalities associated with Edward syndrome**



**Fig. 5: Bar chart of various abnormalities associated with Edward syndrome**

**DISCUSSION**

Total 152 cases of Edward syndrome has been reviewed which are proved by cytogenetic study. The male to female ratio is 1.3:1. In more than 50% of cases, both maternal and paternal age is less than 30 years. In 3/4<sup>th</sup> of cases, Edward syndrome is associated with primigravida.

Most common abnormality associated with cardiovascular system followed by extremities, urinary system, head and neck, gastrointestinal tract and genitals. Most common cardiovascular abnormalities are ventricular septal defect, atrial septal defect and patent ductus arteriosus. Most common abnormalities associated with extremities are calcaneo-valgus defect, hip abduction and finger deformity. Most common eye abnormalities are microphthalmos, epicanthal fold and ocular hypertelorism. Most common ear abnormality is low set ears. Micrognathia and short neck is also a common finding. Skull shows elongation defect with microcephaly. Mental retardation associated with both hyper and hypotonia is a common findings. In gastrointestinal tract, diaphragmatic hernia, umbilical hernia and pyloric stenosis are routinely seen. . In urinary system, hydronephrosis, hydroureter with posterior urethral valve is commonly found.

Undescended testis is commonly associated with this syndrome.

**CONCLUSION**

In conclusion, Edward syndrome is a rare genetic disorder associated with multisystem involvement. Hence, data collection and frequent review of cytogenetically proven cases is must to study in detail about the association of genetic defect and organ system involved. It is also helpful to compare with their known genetic disorder.

**REFERENCES**

1. What is Trisomy 18? Trisomy 18 foundation. Available from [http://www.trisomy18.org/site/PageServer?PageName=whatisT18\\_whatis](http://www.trisomy18.org/site/PageServer?PageName=whatisT18_whatis)
2. Edwards JH, Harnden DG, Cameron AH, Crosse VM, Wolff OH; A new trisomic syndrome. *Lancet*, 1960; 1 (7128):787-790.
3. Smith DW, Patau K, Therman E, Inhorn SL; A new autosomal trisomy syndrome: multiple congenital anomalies caused by an extra chromosome. *J Pediatr.*, 1960; 57: 338-345.
4. Taylor AI; Autosomal Trisomy Syndromes: A Detailed Study of 27 Cases of Edwards' Syndrome and 27 Cases of Patau's Syndrome. *Journal of Medical Genetics*, 1968; 5(3): 227-252.
5. Naguib KK, Al-Awadi SA, Moussa MA, Bastaki L, Gouda S, Redha MA *et al.*; Trisomy 18 in Kuwait. *International Journal of Epidemiology*, 1999; 28(4): 711-716.
6. Bhat BV, Usha TS, Pourany A, Puri RK, Srinivasan S, Mitra SC; Edward syndrome with multiple chromosomal defects. *The Indian Journal of Pediatrics*, 1989; 56(1): 137-139.
7. Barani K, Padmavathy R; Trisomy 18- a case report. *Internet Journal of Pathology*; 2013; 15(1). Available from <http://ispub.com/IJPA/15/1/2950>
8. Patra S, Garg A, Krishnamurth S, Aneja S; Edward's syndrome with a novel karyotype. *Bangladesh Journal of Medical Science*, 2011; 10(3): 211-212.
9. Bhanumathi B, Goyel NA, Mishra ZA; Trisomy 18 in a 50-year-old female. *Indian Journal of Human Genetics*, 2006; 12(3): 146-147.