# Edward Syndrome (Trisomy 18): A Case Report

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***Abstract-*** To assess a case of Edward's Syndrome (trisomy18). Review of literature shows only few series evaluating such cases stimulating us to publish this case. The data reviewed does not appear to have any adverse effect on the mother or fertility.

## I. INTRODUCTION

**Edwards syndrome** (also known as **Trisomy 18** [**T18**]) is a genetic disorder caused by the presence of all or part of an extra 18th chromosome. This genetic condition almost always results from nondisjunction during meiosis. It is named after John Hilton Edwards, who first described the syndrome in 1960.[1] It is the second most common autosomal trisomy, after Down syndrome, that carries to term.

Edwards syndrome occurs in around one in 6,000 live births and around 80 percent 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, and other internal organ disorders.

It occurs in 1 in 2500 pregnancies and 1 in 6000 pregnancies in the US. Reports say that a baby boy has higher still birth rates than a baby girl. Very small number of people with this condition live till 20's to 30's with multiple abnormalities.

Here we are presenting a case of a patient with fetus detected with Edward's Syndrome (Trisomy18).

## II. CASE REPORT

Our patient 45 year female Gravida four, Para one, living one, abortions two with 14.3 weeks B/D and 15.0 weeks B/S with previous LSCS (i/v/o transverse lie) came for medical termination of pregnancy i/v/o fetal chromosomal anomaly that is trisomy18 as mentioned in the scan and referral notes.

Menstrual history: patient attained menarche at 14yrs of age which regular cycles in the past of around 28 to 30 days lasting for 4-5 days with moderate flow not associated with pain.
LMP: 26/07/13
EDD: 03/05/14

Obstetric history: G4P1L1A2
P1L1: Male baby/15yrs/LSCS i/v/o transverse lie.
A1: Two months/ spontaneous/ 14yrs back/ D and C done.
A2: Two months/ spontaneous/ 7yrs back/ D and C done.
G4 :Present pregnancy.
No history of contraception.
Past history: No history of diabetes mellitus, hypertension, bronchial asthma, tuberculosis or fever.
No history of blood transfusion.
No history of any major medical or surgical illness.

Personal history: sleep and apetite are normal.
Bowel and bladder habits are regular.
No addictions.

Family history: not significant.

On examination: GC fair
P=92/min
BP= 120/80 mm of hg
No pallor/ no edema.
RS/CVS: NAD
P/A: uterus just palpable
Soft/no guarding/ no rigidity.
L/E: NAB.

## III. INVESTIGATIONS

Hb= 12.1 gm%
TLC= 9200
PLT= 2.1 lac
S.Creat= 0.9
BT/CT= 1'20"/3'15"
Beta HCG= 10.65 mg/dl
TSH= 1.5
HIV/HBsAG/VDRL= Non reactive

FLUORESCENCE IN SITU HYBRIDIZATION(FISH) was performed on the interphase with uncultured cells of chorionic villi using the **Vysis AneuVysion DNA Probe kit** for chromosome 13, 18 and Y.

This test indicated a high risk for trisomy 13 and 18.

Our patient was explained about the risks and complications associated with this condition and the need for termination of pregnancy.

Patient was induced with tablet misoprost 400 microgram per vaginally at 12.30pm followed by 200 microgram per vaginally after six hours at 6.30pm. Patient delivered at 10.15 pm.
The abortus and the placenta together weighed around 125 gms.

## IV. DISCUSSION

Humans are usually born with 46 chromosomes, which are arranged in 23 pairs. The chromosomes are numbered from 1 to 22 and the last pair, known as X and Y, determine whether we are a boy (XY) or a girl (XX). One of each pair of chromosomes comes from our mother in the unfertilized egg and the other of the pair comes from our father in the sperm. Very occasionally, a baby is born with an extra copy of chromosome number 18. This

condition is known as Edward's syndrome. Edward's syndrome happens more often in girls than boys, but it is not known why.

When Edward's syndrome is caused by an entire extra chromosome 18 this is called a "primary trisomy" (as shown opposite). This is a non-inherited version of Edward's syndrome. An alternative (but rare) cause of Edward's syndrome 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'. This can be discussed in a genetics clinic.

It is possible to distinguish between the 2 causes of Edward's syndrome by looking at the chromosomes of the baby. This can be done during pregnancy, or after pregnancy by testing the baby's blood, or blood from the umbilical cord. If the baby's chromosomes have not been checked then it is possible to look at the chromosomes ofthe parents to see if either of them carries a balanced translocation.

About 1 in 3000 pregnancies/babies are diagnosed as having an extra copy of chromosome 18. Edward's syndrome is named after Dr. John Edwards who discovered that the extra chromosome causes the condition. The condition is also known as Trisomy 18 because there are 3 copies of chromosome 18.

Edward's syndrome is almost always caused by a primary trisomy and therefore it is very unlikely that a future pregnancy will have this condition. However some parents do choose to have a test in a future pregnancy to check the chromosomes of the baby. The need for testing and how this is carried out can be discussed at the genetics clinic.

Sometimes Edward's syndrome can be suspected during an ultrasound scan, where the main structures of the baby are looked at to pick up any potential problems. Detecting the condition early can help some parents to prepare themselves for how the condition will affect their lives. It also gives them the opportunity to make the personal choice of whether or not to continue with the pregnancy.There are many things that could be seen during an ultrasound scan that increase the baby's chance of having Edward's syndrome. Some of these signs would suggest serious abnormalities. Other signs are not actually harmful on their own but do occur more often in babies with conditions like Edwards.

Here are a few of the more common signs that suggest a baby has Edwards syndrome:

## V. Raised nuchal thickness

A scan may be done at 12 to 14 weeks to look for a small collection of fluid on the back of the baby's neck. The thickness of this fluid filled area is measured ("nuchal translucency" or "nuchal thickness") and if it is larger than average ("raised") it suggests the baby might have a condition such as Edward's syndrome.In most cases, the extra chromosome is present due to a "genetic mistake" that occurred in either the egg or the sperm that went to make that baby. The parents usually have normal chromosomes themselves. It is not known why the "genetic mistake" happens, but it is slightly more likely to occur in babies of older mothers. Edward's syndrome is a serious condition and affected babies/pregnancies can have a range of severe medical problems. Sadly, most babies with Edward's syndrome die before the end of pregnancy or are stillborn. Of the babies that are born alive, about half survive the first month of life and less than 10% live longer than a year. For the most part they require specialised nursing in a hospital or hospice. However there are some infants who can live at home and be cared for by their parents. Babies with Edward's syndrome tend to have a low birth weight, a small head ("microcephaly"), a small jaw Mosaic Edward's syndrome is a rare form of the condition where some cells in the body have 2 copies of chromosome 18 and others have 3 copies of 18. Mosaic Edward's syndrome is very varied. Some babies are only mildly affected, while others have as many problems as babies with the "full" form. How a child/person is affected depends on how many cells have three copies of chromosome 18. If a couple have had one baby with mosaic Edward's syndrome, they are very unlikely to have another child with the same condition. ("micrognathia"), malformations of their heart and kidneys,clenched fists and malformed feet. They also characteristically have feeding and breathing problems in infancy and severe learning disability.

So here, we present rare case of Edwards Syndrome.

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