

## **21 WEEKS FETUS WITH TRISOMY 18 (EDWARD SYNDROME), BONE MALFORMATIONS HIGHLIGHTED**

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Edwards syndrome also known as Trisomy 18 (T18) or Trisomy E is a genetic disorder caused by the presence of all or part of an extra 18th chromosome. 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; and in males, undescended testicles. The diagnosis of Trisomy 18 is by ultrasound and amniocentesis, the fetal DNA is examined for genetic abnormalities. Therapeutic abortion it can be done.

*Key words:* Micrognathia, malformed hands, ultrasound diagnostic cranial malformations, omphalocele, therapeutic abortion.

### **INTRODUCTION**

Edward syndrome is a trisomy of chromosome 18. Symptoms include mental retardation and numerous congenital anomalies causing serious health problems. It seems to affect females about three times more than males there is a case of births 5000–7000 about 1% of cases of spontaneous abortion are caused by trisomy 18.

Children born with trisomy 18, 50% do not live more than a week of extrauterine life and about 90% do not live more than one year. Most often the problem is present in excess chromosome in her ovule.

Most children with trisomy 18 die within the first year of life. Average lifespan is shorter than 2 months for 50% of children, 5–10% survive the first year after birth, but shows severe mental retardation. Edward Syndrome can not be prevented

### **Characteristics of trisomy 18**

A syndrome is a condition distinguished by a number of features that often occur together. The combination of features present in babies affected with trisomy 18 can lead to many different developmental and physical problems.

Symptoms may include:

1. Failure to grow and gain weight at the expected rate and severe feeding difficulties, diminished muscle tone and episodes in which there is temporary cessation of spontaneous breathing

2. Developmental delays and intellectual disability.

3. A prominent back portion of the head, low-set, malformed ears, an abnormally small jaw, a small mouth with an unusually narrow roof, an upturned

nose, narrow eyelid folds, widely spaced eyes, and drooping of the upper eyelids and undescended testes in boys.

4. Abnormalities in the bones of the hands and feet which may include overlapping flexed fingers, webbing of the toes, a deformity causing the heels to turn inwards and the soles flexed (clubfeet).

5. A small pelvis with limited hip movement and a short breastbone.

6. Kidney abnormalities and structural heart (cardiac) defects at birth such as an abnormal opening in the partition dividing the lower chambers of the heart and a persistence of the fetal opening between the two major arteries (aorta, pulmonary artery) emerging from the heart.

7. These congenital heart defects and respiratory difficulties may lead to potential life-threatening complications during infancy or childhood.

There may be a number of indications that there is an increased risk for the baby having trisomy 18 including:

- The mother's age
- A family history of trisomy 18
- The results of a screening test for this condition in pregnancy



Fetal karyotype with trisomy 18.

Edward's syndrome is something that may be detected prior to the birth of the child. Potential testing includes maternal serum alpha-fetal protein analysis or screening, amniocentesis, ultrasonography, and chorionic villus sampling. A woman who is pregnant with a child who has Edward's syndrome might have an uncommonly large uterus during the pregnancy because of the presence of

extra amniotic fluid. An unusually small placenta might be noted during the birth of the child. They also seem to have an increased risk for the development of a, "Wilms' tumor", a form of kidney cancer that affects children for the most part. It is recommended that children with Edward's syndrome have a routine ultrasound of their abdomen.

## RESULTS

In April of 2011 was admitted to the clinic "Prof.Dr.Panait Sârbu" a pregnant woman aged 33 years after repeated ultrasound examination we have established a number of fetal malformations including: malformations of the hand, forearm hiperflexia it on the abnormal digitationes, omphalocele, micrognathia, hypertelorism, chest atrophy and weakness of the pelvic bone.

In the pictures below will present alternative images of ultrasound and fetal after abortion to highlight specific relevance of ultrasound on bone changes in trisomy 18.

The diagnosis of trisomy 18 is made by amniocentesis and genetic analysis of karyotype. Amniocentesis for genetic diagnosis is usually done between weeks 15 to 20. In the United States, is the most common procedure used to diagnose fetal aneuploidy and other genetic diseases. Its safety has been confirmed by numerous multicenter study ultrasound guidance is used to pass a spinal needle 20 to 22 in diameter through the amniotic sac while avoiding the placenta, umbilical cord and fetus. Approximately 20 ml of fluid is collected for fetal cariotiparea needle is removed. Ultrasound to find uterine bleeding at puncture site, and fetal heart rate is noted at the end of the procedure.

Complications are rare and include transient spotting or amniotic fluid leakage in 1–2% and 0.1% in amniocorion. Fetus with needle trauma are rare. Fetal cells obtained during amniocentesis frequently grow in culture. However, this is possible if the fetus is abnormal. Digital PCR chorionic villous tissue amniocitelor or uncultivated give rapid aneuploidy detection and seems to be promising in expanding medical practice.

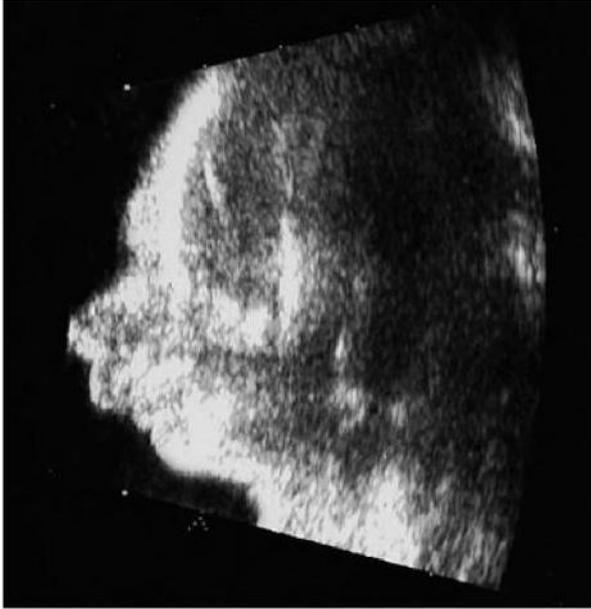


Fig.1. Micrognathia and occipital bone hypertrophy



Fig. 2 Clinodactilie, abnormal insertion of fingers.



Fig. 3 Hiperflexia of the hand.

The second quarter amniocentesis is associated with the lowest incidence of unknown results - up to 0.8%. In these circumstances, these findings are rarely true fetal mosaicism, but instead indicate a new or pseudomosaicism placental mosaicism.



Fig. 5. Abortion and placenta delivered spontaneously.



Fig. 4. Omphalocele.

## DISCUSSION

Following scans were able to detect a number of fetal abnormalities, including bone resulted in suspicion of trisomy 18. This was confirmed by amniocentesis and thus could establish effective therapeutic procedure.

Ultrasound elements that raised suspicion of trisomy 18:

1. Appearance of the fetal skull, occipital bone hypertrophy, micrognathia, small orbits spaced (hypertelorism), low insertion ears, palate hypoplasia, narrow nose.

2. Such skeletal malformations, it's a narrow thorax, prominent calcaneus, hands hiperflex, overlapping fingers over thumb, fingers glued planting.

3. Other anomalies associated with this genetic abnormality, omphalocele, polyhydramnios, choroid plexus cysts.

These anomalies detected by fetal ultrasound, the genetic analysis of karyotype required to perform amniocentesis and fetal cell harvesting. This is the only method that can definitely diagnose trisomy 18.

An important issue is how to present therapeutic abortion is caused in our clinic. So when it was established genetic diagnosis and pregnant women called for abortion was done at the start of abortion by administering prostaglandin (misoprostol 200 $\mu$ g) both vaginally and orally. Pursuing the uterine tone, frequency of uterine contractions, blood pressure, pulse, urine output, general condition of the patient. Operational administration of this treatment can be done over a period of 48–72 hours, possibly oxiton to support management of uterine contractions and

administration of medicinal products for increasing estrogen and oxytocin receptor maturation.

After abortion expects placental expulsion if do not produce it, they practice controlling instruments for its extraction. Usually after abortion and placenta delivery its practiced cavity control to check whether it is free to prevent bleeding or infectious complications.

The advantages of this procedure are: maintaining integrity uterine cavity, perforation and bleeding risks significantly lower risk of uterine synechiae low and not least producing a similar miscarriage miscarry naturally produced (fig.5).

### CONCLUSIONS

Highlighting ultrasound or other bone abnormalities is one way that can be suspected Trisomy 18. Amniocentesis and amniotic fluid extraction is the most common way of diagnosing fetal trisomy 18 by the genetic analysis of cells. Outbreak therapeutic abortion using prostaglandins provide a favorable prognosis on reproductive function of women compared with conventional methods of abortion.

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