Case Report

# Omphalocele and Intrauterine growth restriction, an unusual association of two Congenital syndromic malformations

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## Abstract

We report a case of a 30 year old pregnant woman. Ultrasound examination showed severe intrauterine growth restriction (IUGR) and an omphalocele. Amniocentesis was performed and the fetal chromosomal analysis showed mosaic trisomy 18. Further genetic investigations were done. The pregnancy was terminated one week later. Autopsy confirmed the ultrasound images findings.

Our presentation is a very rare case report of mosaic trisomy 18, prenatal diagnosis, with important an anusual association of two congenital malformation, omphalocele and intrauterine severe growth restriction.

**Key words:** Omphalocele, Intrauterine Growth Restriction, Ultrasound, Amniocentesis, Mosaic Trisomy 18

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## Introduction

Omphalocele is a midline abdominal wall defect with extrusion of abdominal viscera, covered by a membranous sac, into the base of the umbilical cord and is the one of the most common congenital malformation of the anterior abdominal wall [1, 2]. Omphalocele is frequently associated with other congenital malformations [3, 4, 5, 6]. However, the frequency of the reported associated malformations for omphalocele ranges from 27% to 63% [7].

The objective of a presentation is to determine wether omphalocele and intrauterine growth restriction (IUGR) are syndromic [8,9,10,11,12,13,14] or non syndromic [15].

## Case Report

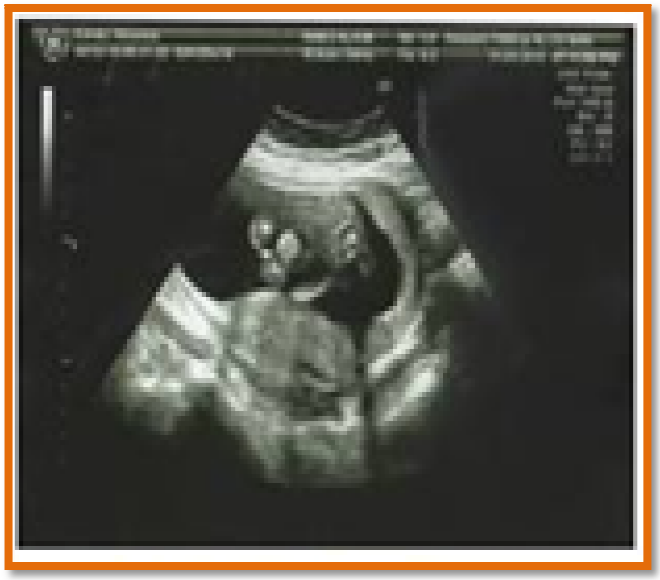
We report a case of a 30 year old Caucasian woman (Gravida 1, Para1), who was referred at 17 weeks' gestation for abnormal 2nd trimester prenatal scan. There was no family history of congenital malformations and chromosomal abnormalities. The couple had normal general health and was not consanguineous. Routine ultrasonography at 17 weeks of pregnancy, double and triple marker test (AFP, uE3 and hCG), selective ultrasonography for detection of fetal abnormalities, 3D and 4D live scan with Voluson Echograph E8, amniocentesis, fetal karyotype and OF-PCR were performed.
Double marker test was performed at 12w+3 days and was normal.

| Risc type | Calculated | Standard |
|---|---|---|
| **Age** | 1/1417 | |
| **Biochemical** | < 1/1000 | 1/380 |
| **Combinated 13/18** | 1/1000 | 1/380 |

Ultrasound evaluation revealed severe intrauterine growth restriction (IUGR): 16.4 weeks - biometrical age and 20.1 weeks - cronological age, normal cerebral and heart anatomy, normal nuchal fold 2,5mm.

**Case Report**

At the cord insertion on the anterior abdominal wall, we observed a 16 mm nonhomogenous medium echogenity image which suggested an omphalocele (Fig. 1, Fig. 2).



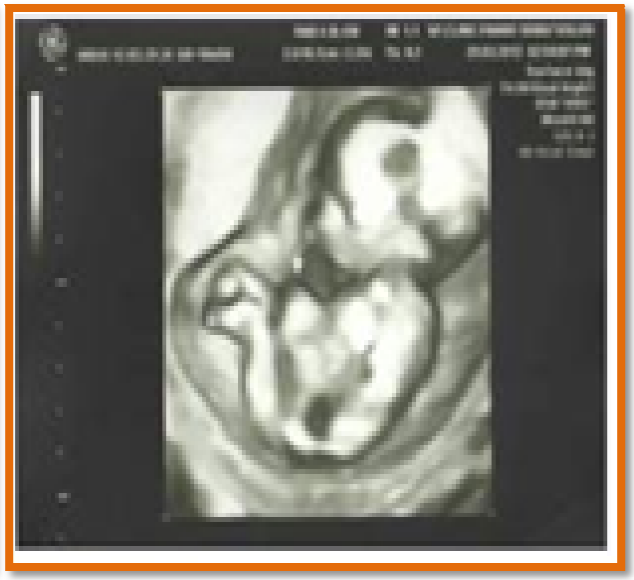


Fig. 1: 2D Mode & Fig. 2: 3D reconstruction showing 16 mm nonhomogenous medium echogenity image which suggested an omphalocele

Immediate amniocentesis was performed. It revealed the elevation of the AFP 19848 IU/ml in the amniotic fluid (median deviation 11800 IU/ml). The Fluorescence in situ hybridization (FISH) analysis showed a trisomy 18 mosaicism, with 85% of the chromosomes affected. (Fig 3).

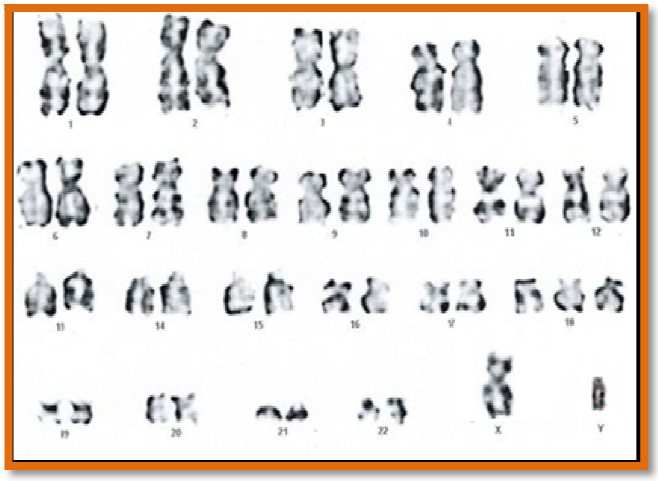


Fig 3: Kariotype 47,XY, +18

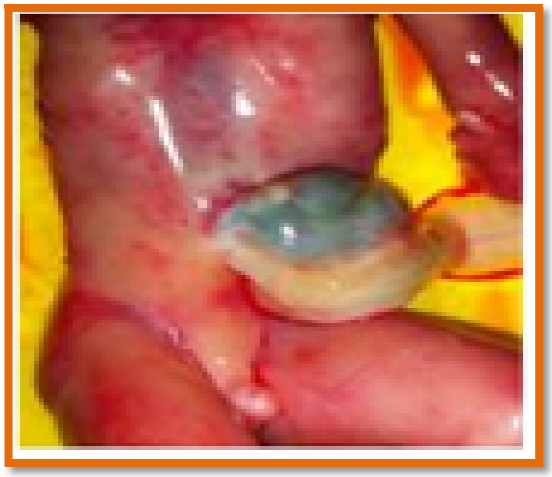


Fig 4: After fetal demise omphelocele is visible

The parents decided to terminate the pregnancy one week later. Autopsy revealed the ultrasound images (Fig. 4).

## Discussion

Chromosomal mosaicism is the presence of more than one cell line in the same individual, and it occurs in approximately 5% of trisomy 18 cases [16, 17]. These individuals carry both a trisomy 18 and an euploid cell line. Their clinical findings are highly variable, from the absence of dysmorphic features to the complete trisomy 18 syndrome [18].

Fluorescence in situ hybridization (FISH) provides a rapid and accurate technique for detecting chromosomal aneuploidy. It is an excellent method for identifying mosaicism in placental tissues following prenatal diagnosis (19).

This study demonstrates the usefulness of amniocentesis and FISH analysis for the prenatal detection of this rare case of trisomy 18 mosaicism.

The two major anomalies, omphalocele and intrauterine severe growth restriction, were diagnosed using 3D ultrasound examination in the second trimester of pregnancy. Further evaluation of both the parents and future pregnancies should be assessed.

Prenatal ultrasound examination and genetic diagnosis was very useful in the management of a fetus with a unusual association of two congenital syndromic malformations.

## Conclusions

Our presentation is a very rare case report of mosaic trisomy 18, prenatal diagnosis, with important an anusual association of two congenital malformation, omphalocele and intrauterine severe growth restriction.

In the same time, this case provides further evidence for the benefit of advanced imaging for prenatal diagnosis and the understanding of fetal condition. More important for our patient was her better understanding of fetal disorder due to 3D findings.

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**Permission from IRB:** Yes

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