

# A monochorionic, monoamniotic twin pregnancy with one anencephalos fetus

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## ABSTRACT

*Even though presumably identical twins, arising from one single fertilized ovum, sharing the same genetic material and exposed to the same environment in the uterine life, we present a situation of striking morphological differences between such twins, an extremely rare situation, with only 8 prior citations in the literature.*

*A 37 years old woman, with a history of 7 spontaneous first trimester abortions, presents at emergency room for vaginal bleeding and low abdominal pain during the last 2 days after 8 weeks of amenorrhea. The ultrasound scan diagnosed a monochorionic, monoamniotic twin pregnancy.*

*The scan performed at 16 weeks gestational age shows the first twin developing more or less as expected, whereas the second twin has no identifiable bony cranial structure. Amniocentesis and the cytogenetic analyses found no chromosomal abnormalities. At 18 weeks gestational age the ultrasound scan shows no sign of cardiac activity in the anencephalos twin, followed two days later by the death of the other twin*

*Conclusions: Obviously, the development of the fetuses in a monozygotic pregnancy is determined by the genetic factors and environmental conditions, but it seems that there are more subtle elements that can modify this evolution and have to be taken into consideration.*

**Key words:** twin pregnancy, monochorionic pregnancy, anencephaly

**E**ven though presumably identical twins, arising from one single fertilized ovum, sharing the same genetic material and exposed to the same environment in the uterine life, we present a situation of striking morphological differences between such twins.

A 37 years old woman, with a history of 7 spontaneous first trimester abortions in the past 5 years, following a term delivery, presents at emergency room for vaginal bleeding and low abdominal pain during the last 2 days. Her last menstrual period was 8 weeks before.

Regarding her obstetric history, all pregnancies were singleton and no signs of fetal abnormalities were found. The term pregnancy was uneventful, ended with a vaginal spontaneous delivery of a 3400 grams, male, healthy

newborn. The post-partum period revealed no pathological events. Investigations made in order to assess infectious and immunological pathology found no significant results. The gynecological exam at the present admission revealed an enlarged uterus, concordant with pregnancy, a 50% shortened cervix, 2 cm open.

The ultrasound scan at admittance revealed the gestational sac within the uterine cavity measuring 59 mm in diameter and containing two embryos with crown – rump – lengths of 1.8 and 1.6 cm, respectively, without separating visible membrane. Cardiac activity was present in both embryos. Thus, the monochorionic, monoamniotic twin pregnancy diagnosis was made and action taken in order to assure a best possible pregnancy course.

Tocolytic therapy was indicated and the bleeding diminished progressively and stopped. The 3<sup>rd</sup> day, considering the local status, cervical cerclage was performed, without unexpected side events and the patient discharged after 2 more days.

At 12 weeks nuchal translucency is measured in both embryos, showing normal values, 2.8 mm and 2.6 mm, but the first alarm sign is drawn when cranial structure of one twin, situated at the fundus of the uterine cavity, is not properly visible, too soon though, to establish a diagnosis. Close observation, as well as further investigations are decided.

At 16 weeks the second trimester maternal serum "triple-test" is performed with the following results: the free oestriol and b human chorio gonado tropin were within normal range for gestational age, adapted for twin pregnancy but  $\alpha$ -fetoprotein value was extremely high. The scan performed at this time shows a visible difference in the development of the two fetuses. The first twin, in a lower position, appeared to develop more or less as expected, fetal biometry showing a 7-8 days delay compared to normal values for gestational age. The second twin, however, had no identifiable bony cranial structure and presented the same restriction in the uterine growth.

At this moment it appears clear that one of the twins is anencephalous, while the other has no visible morphological abnormalities. Considering the mother's strong wish to continue the course of pregnancy, amniocentesis is performed, followed by cytogenetic analysis, which found no chromosomal anomalies (46 XX).

At 18 weeks gestational age the ultrasound scan shows no sign of cardiac activity in the anencephalos twin, as well as an increasing delay in morphological development of both twins, followed two days later by the death of the other twin.

Abortion is induced with local prostaglandins and after a 7 hours labor the patient delivered two twins, no life signs present, one anencephalos, 160 grams and one apparently normal developed infant, 180 grams, with a large scalp haematoma. The histopathological exam, extremely difficult, due to tissue fragility, revealed no further anomalies in any of the twins.

The post-abortion period was uneventful and the patient was discharged after two days with antibiotic therapy and recommendation for a two weeks return visit. □



FIGURE 1. The anencephalos twin: no cranial bony structure visible



FIGURE 2. The two resulting twins: a striking difference



FIGURE 3. The apparently normal twin: large scalp haematoma



FIGURE 4. The anencephalos twin

## THEORETICAL BACKGROUND

Regarding the monozygotic pregnancy, as mentioned before, the genetic material is, theoretically, identical. However, considering the implications of perigenetic mechanisms, the events in the early conception, post-twinning period, genetic differences between twins are possible (1). Occasionally, monozygotic twins may be discordant for chromosomal and single gene disorders and there are quite numerous cases of cleft lip and cleft palate, renal and neurological inter-twin differences (1). From a research point of view these differences are of much importance, enabling the identification of certain mutations involved in the determination of a structural anomaly (1). Monoamniotic twins discordant for anencephaly are a rare situation, with only 8 prior citations in literature (2). Besides the etiology of the anomaly, which, certainly, is subject to many debates and involves epigenetic variability imprinting and variations in environmental exposure (1,3), a more practical aspect is related to the management of a such pregnancy.

The first step in defining the approach is assessing the chorionicity. Visualizing a single gestational sac in the early first trimester, with two embryos and one yolk sac, is indicating for a monochorionic, monoamniotic pregnancy. In the very few cases in which an important structural defect is identified in one twin, the other one being apparently normal, the management issue is not a simple one and it depends on the severity of the anomaly, the impact on the other twin and, of course, on the family wishes and believes. Alternatives include expectant management, selective termination of the anomalous fetus and termination of the entire pregnancy. Without considering the last one a real therapeutic option, expectant management is a risky choice, as preterm delivery, low birth weight, higher intrauterine death rate are complicating more than 20% of the cases (4). Moreover, in most of the cases, the polyhydramnios developed by the anencephalos twin, is adding to all these problems (4).

Selective termination of the affected fetus depends on the chorionicity. In a dichorionic pregnancy intracardiac injection of potassium chloride under ultrasonographic guidance is a relative simple and safe method (4). In a monochorionic pregnancy the method is prohibited, as it involves damage or even death of the

normal twin. Among the methods indicated in this case are percutaneous injection of sclerosants, as absolute alcohol, into the umbilical cord or intrahepatic vein (4), an older method, now abandoned for less invasive and more effective ones like fetoscopic cord ligation (5), ablation of the umbilical vessels (6), bipolar forceps cautery of the umbilical cord (6) or even more recent ones like fetoscopic section of the umbilical cord with ultrasonic harmonic scalpel (7) or radiofrequency thermablation (8). These techniques, apparently are associated with an improved outcome.

In a large multicenter study, involving 402 cases, the pregnancy loss rate was 8% before 24 weeks GA, 8% between 29-32 weeks and 17% between 33-36 weeks (9). In a different study, with a lower number of cases, 200, the overall pregnancy loss rate was as low as 4% for twin pregnancies (10). These results are quite encouraging when compared to the high rate of complications associated to expectant management and the general trend is to counsel patients for the selective termination of the affected fetus in order to protect the viable one, when the anomaly is severe enough. □

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