AN UNUSUAL CASE OF EXOMPHALOS WITH SIRENOMELIA, IMPERFORATED ANUS AND OTHER MULTIPLE MALFORMATIONS, DETECTED BY MEANS OF REAL-TIME ULTRASONOGRAPHY IN A TWIN PREGNANCY WITH ANOTHER SYMPODIC PAPYRACEUS FETUS

P. MARTINELLI (*), A. DI LIETO (**), D. CATALANO (**), M. LOCCI (*)

(*) Obstetric and Gynecological Physiopathology
(**) Antenatal Care
2nd School of Medicine, University of Naples (Italy)

SUMMARY

In this paper it has been reported an unusual case of fetal multiple malformations with the concomitance of exomphalos, sirenomelia, imperforated anus and indistinct external genitalia, detected in a twin pregnancy, with another papyraceus sympodic fetus. The diagnosis of fetal multiple anomalies occurred by means of real-time ultrasonography, evaluating all the direct and indirect U/S findings.

Real-time ultrasound scanning detects many important data about fetal behaviour and normal or abnormal fetal anatomy.

Several single or multiple fetal defects have been discovered in the recent cases. Actually it is not only possible to diagnose accurately some defects as those of the neural tube, but also many abdominal defects, diaphragma defects and limbs anomalies. An unusual case of fetal exomphalos with sympodia and other multiple defects, occurring in a twin pregnancy with another papyraceus fetus is discussed in this issue.

CASE HISTORY

The mother, 26-years old, gravida III, para 0, with 2 previous cesarean sections, had no medical history or familiar dispositions to malformations. The first ultrasound examination was performed at the 24th week of gestation since she didn't feel any fetal movements, and a clinical examination showed uterine size/date discrepancy.

The real-time scanning showed a living fetus in transverse position, an anterior placenta and a marked oligoamnios. BPD was 5.2, corresponding to the 20th week of gestation. It was very difficult to show the spine and the chest was very small indeed (Fig. 1). The abdominal transverse scanning showed an irregular mass with solid and cystic structures inside, instead of normal abdominal shape (Fig. 2). During a 30 min examination no limb movements were observed.

The following U/S examinations confirmed a marked BPD growth retardation (Fig. 3), while the abdominal mass expanded in the small volume of amniotic fluid (Fig. 2).

It was thus evident the presence of severe multiple malformations, such as exomphalos and limb anomalies.

The patient was informed about the severity of the diagnosis, but we decided to wait for the spontaneous termination of pregnancy because of previous hysterotomies.

The pregnancy lasted 39 weeks, at that time the fetus died and the labor started. Premature rupture of membranes occurred and some fetal viscera appeared in vagina, with the fetus in transverse position. The patient delivered by cesarean section a stillborn fetus, of 1800 gr, and a papyraceus sympodic fetus. The first stillborn fetus showed an unusual concomitance of exomphalos, sirenomelia, imperforated anus and indistinct external genitalia (Fig. 4).

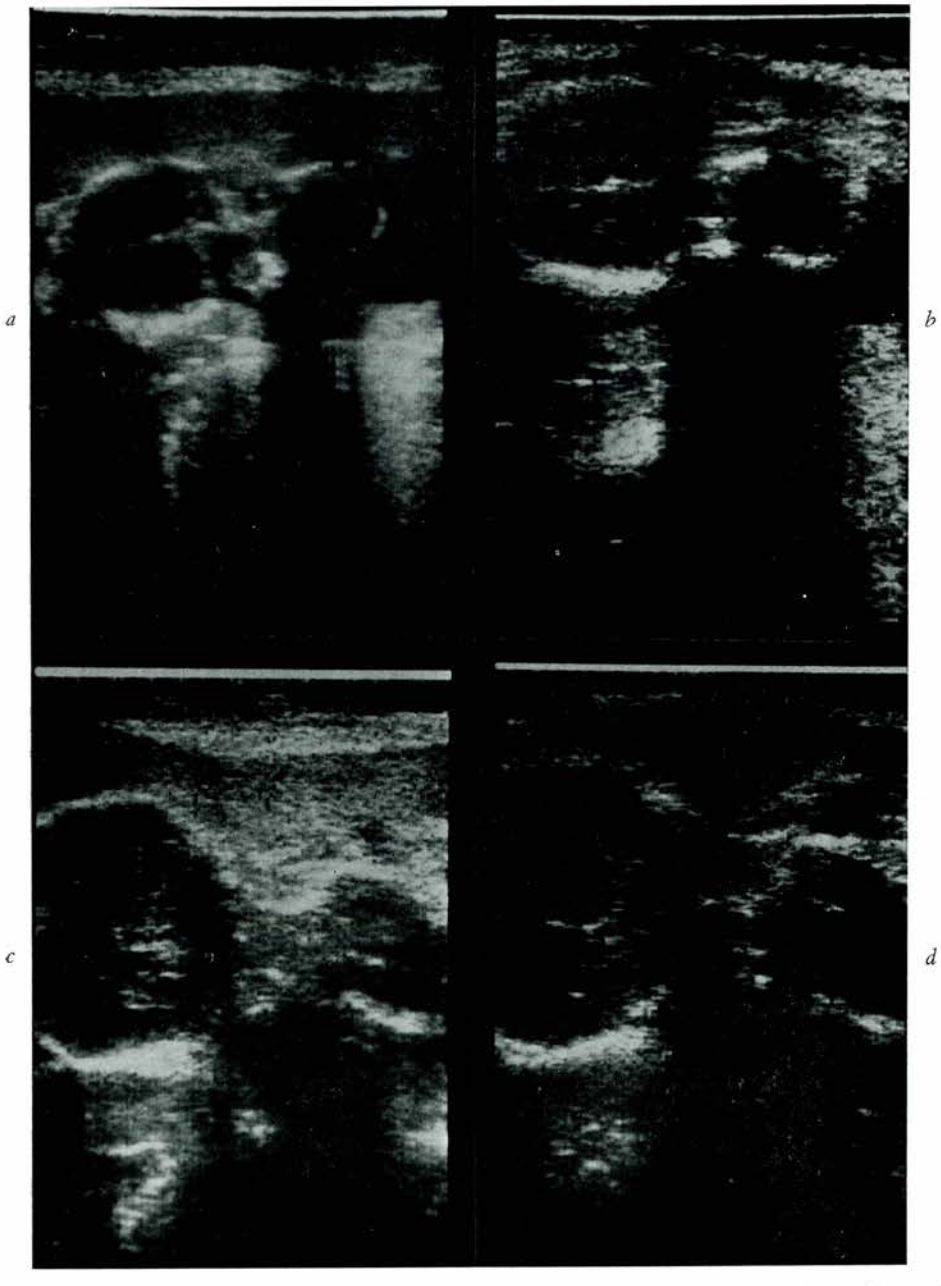

Fig. 1. — Longitudinal scans of the fetal head and chest, performed respectively: at the 24th week (*a*); 31st week (*b*); 34th week (*c*); and 37th week of gestation (*d*).

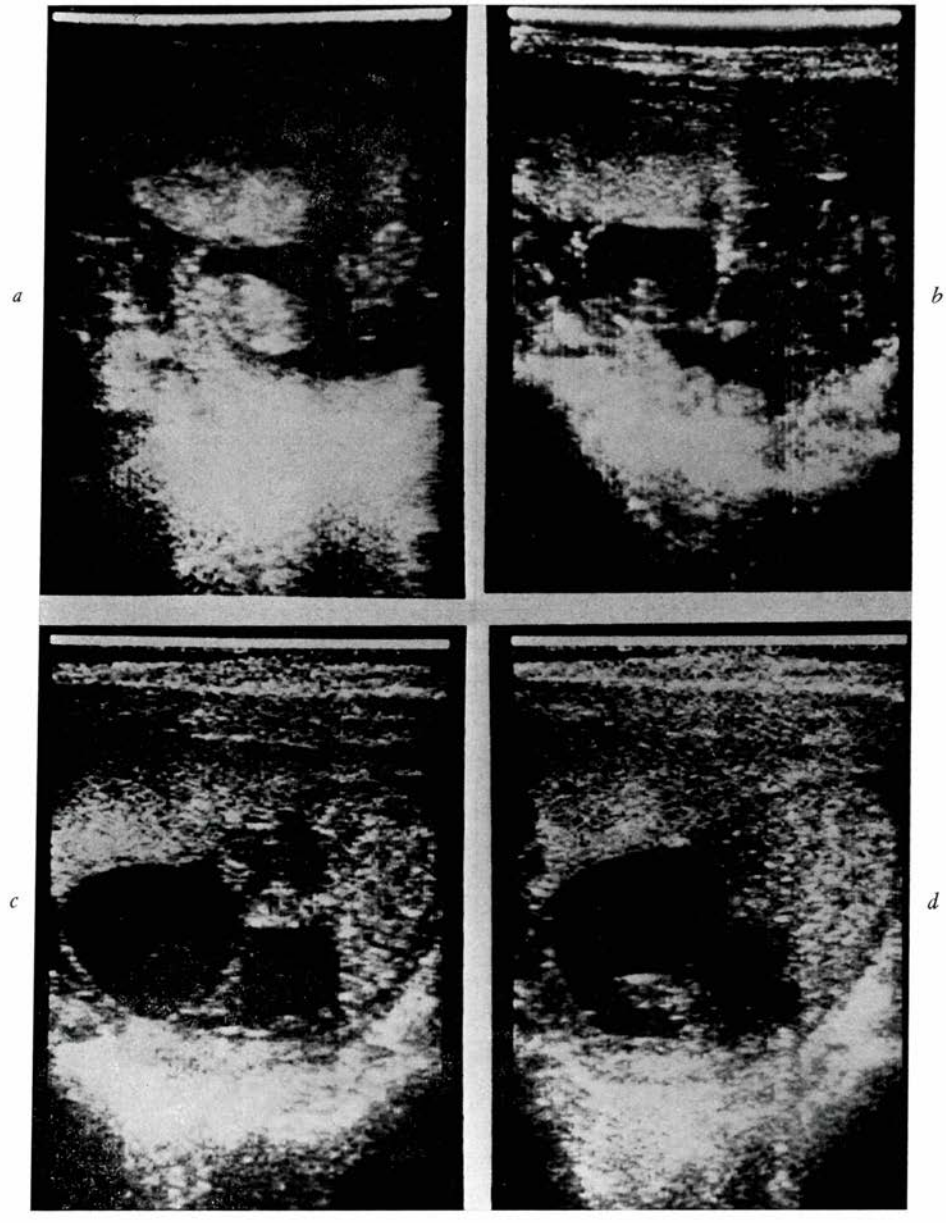

Fig. 2. — Transverse scans of fetal herniated abdominal viscera (exomphalos) performed respectively: at the 24th week (*a*); 31st week (*b*); 34th week (*c*); and 37th week of gestation (*d*).

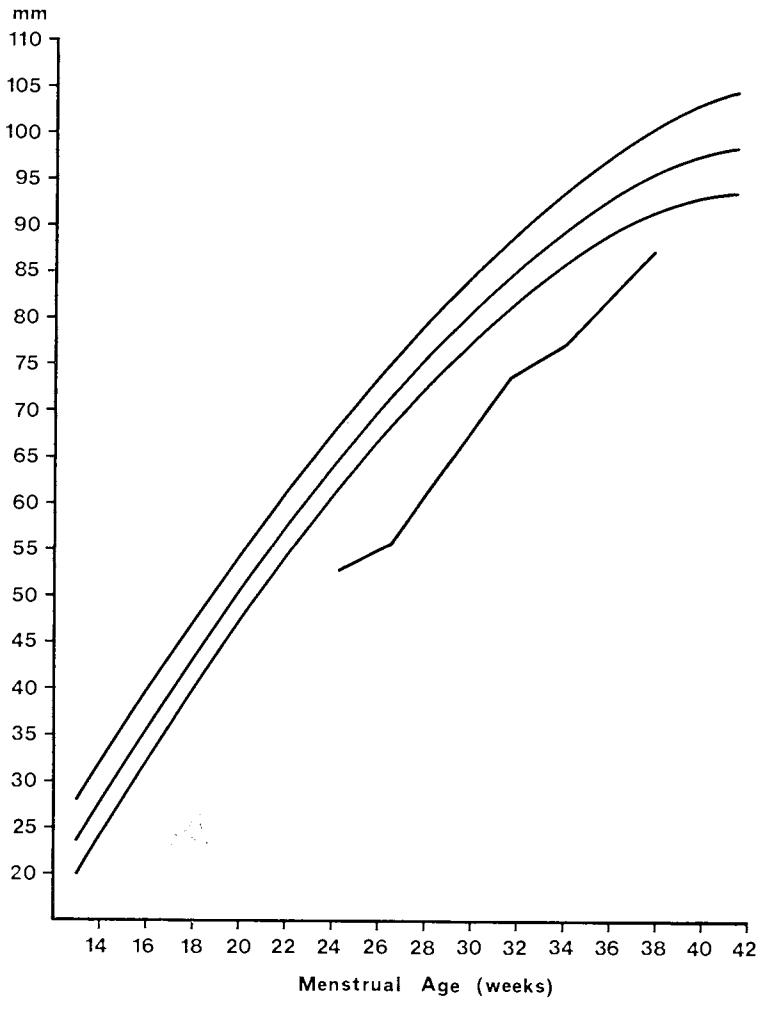

Fig. 3 — The BPD growth curve.

COMMENT

Sirenomelia or sympodia is a very serious malformation that represents a fusion of limbs at their fibular norges, ever associated to pelvis defects and often to imperforated anus. It occurs in about 1/60,000 newborns, and in identical twins ([2]).

On the contrary, the exomphalos is a viscera herniation into the base of umbilical cord, so that at birth, abdominal contents are covered by a membranous sac of peritoneum-amnion. Its incidence varies from 1/3,200 to 1/10,000 newborns, and is related to older maternal age and multiparity ([3]).

Colombani and Coll. ([1]) described many associations of exomphalos and other serious abnormalities such as Meckel syndrome, diaphragma defects, central ner-

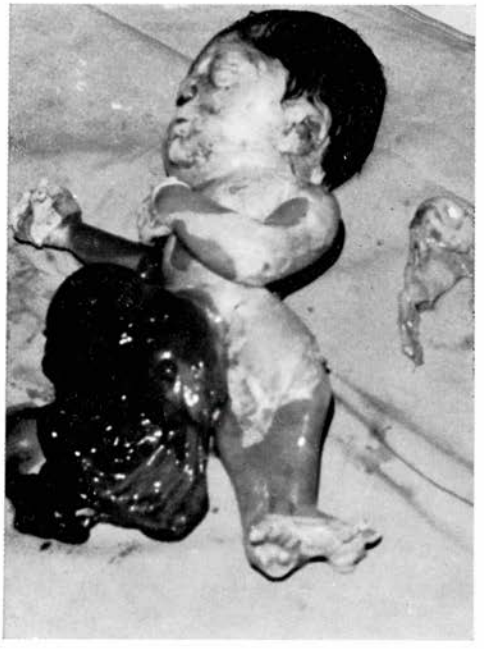

Fig. 4 — The fetus with exomphalos and sirenomelia, the sympodic papiraceus fetus immediately after delivery.

vous system, cardiovascular system and genitourinary tract defects. The peculiarity of the described case is the very rare concomitance of exomphalos and sirenomelia.

The evaluation of all direct and indirect signs, the exomphalos, the marked IUGR, the reduced development of chest, the oligoamnios and the absence of any fetal movements, allowed us to diagnose the most important defects of the fetus.

Prenatal U/S scanning permits to evaluate the severity of fetal anomalies, so that the diagnosis in early pregnancy allows to interrupt it. When in late pregnancy fetal malformations are detected, it's very important to know their severities to select for fetuses which could be saved with an accurate neonatal intensive care at birth. In case of multiple fetal malformations many questions about obstetrical behaviour arise.

In the reported case it was not possible to interrupt the pregnancy for the previous hysterotomies, but the prenatal diagnosis permitted to prepare the mother to the event accurately.

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