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LETTER TO THE EDITOR

Body stalk anomaly: Management of two dichorionic–diamniotic pregnancies

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The anterior wall defects are a heterogeneous group of fetal malformations, essentially represented by omphalocele, gastroschisis, bladder exstrophy, cloacal exstrophy, and body stalk anomaly. Body stalk anomaly is a sporadic, rare and lethal condition, characterized by the presence of a large abdominal wall defect, severe kyphoscoliosis, rudimentary umbilical cord, and limb deformities. The abdominal organs lie outside of the abdominal cavity in the amnio-mesoderm sac, which is limited by the placenta on one side and the amnion on the other (extracelomic space). The umbilical cord is rudimentary or absent, with only one umbilical artery [1]. Associated anomalies are neural tube defects, intestinal atresia, anal atresia, agenesis of the colon, exstrophy of the cloaca, absence of diaphragm, diaphragmatic hernia, genitourinary malformations, craniofacial defects, and anomalies of the pericardium, heart, liver and lungs [1–5].

The prevalence of this syndrome is about 1 per 14 000 births, but the real incidence is 1 per 7500 pregnancies because of spontaneous miscarriage in the first trimester [1,2,4–7]. The pathogenesis of this lethal syndrome has not been completely clarified. There are three main physiopathological hypotheses: early amnion rupture before obliteration of the celomatic cavity with amniotic band syndrome, abnormal embryonic folding when the tri-laminar embryo is transformed into a cylindrical embryo during the first 4 weeks of development, and finally a generalized impairment of embryonic blood flow [1,2,4–7]. We report two consecutive cases of dichorionic–diamniotic pregnancies with one fetus affected by body stalk anomaly.

A 26-year-old woman, gravida 2, para 1, with a dichorionic–diamniotic pregnancy, was referred to our department with a diagnosis of exomphalos of one twin at 22 gestational weeks. A detailed ultrasound scan revealed a morphological normal twin with appropriate growth and regular amniotic fluid volume, and an affected fetus with large anterior abdominal wall defect, thoracic hypoplasia, severe kyphoscoliosis, clubfeet, left heart ventricular hypoplasia and right atrium dilatation, and a very short umbilical cord. Because of the presence of a healthy fetus, an expectant management was decided. Serial ultrasound scans were planned. At 27 gestational weeks the patient was admitted because of the presence of hydramnios in the affected fetus and an evacuative amniocentesis (approximately 1000 mL) was effectuated. At 30 gestational weeks the affected fetus died and four days later an emergency cesarean section was performed for abruptio placenta, resulting in the birth of a healthy baby girl weighing 1347 g. Postmortem examination of the affected fetus confirmed the sonographic diagnosis of body stalk anomaly.

A 29-year-old woman, gravida 3, para 2, with a dichorionic–diamniotic pregnancy, was referred to our department at 23 gestational weeks with an uncertain diagnosis of myelomeningocele or sacrococcygeal teratoma associated with exomphalos. The ultrasound scan revealed a dichorionic–diamniotic pregnancy with one normal fetus and an intrauterine growth-restricted fetus characterized by the presence of relevant structural anomalies: kyphoscoliosis, abdominal wall defect, thoracic hypoplasia, very short umbilical cord, and limb deformities. Similarly

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to the first case, expectant management instead of feticide of the abnormal fetus was preferred. At 28 gestational weeks the affected fetus presented moderate hydramnios. The patient was admitted to our unit at 32 gestational weeks because of the preterm delivery. Tocolysis and fetal lung maturity therapy were administered. An ultrasound scan revealed a moderate hydramnios in the affected fetus. Three days later the patient had a spontaneous premature rupture of fetal membranes. An iterative cesarean section was performed resulting in the birth of a healthy baby girl weighing 1460 g. The affected twin weighed 890 g, presented body stalk anomaly, and died after a few minutes. Postmortem evaluation confirmed the prenatal diagnosis of body stalk anomaly.

The management of body stalk anomaly in cases of singleton pregnancy is obvious because of the lethality of this rare condition, and termination of pregnancy should be considered when the diagnosis is made. In the case of dichorionic–diamniotic pregnancies with one fetus affected, the management is complicated by the contemporary presence of a healthy fetus, therefore a conservative management is advisable. The management should be focused on the well-being of the unaffected twin, taking into account the poor prognosis of the body stalk anomaly. Nonetheless, serial ultrasound scans must be performed because of the risk of fetal demise and preterm delivery. Weekly assessment of amniotic fluid volume and Doppler ultrasounds should be carried out, and fetal growth and eventually cervical length measured once every two weeks.

Body stalk anomaly is a rare and sporadic malformation. It should always be considered in the differential diagnosis of fetal abdominal wall defects, and because of its poor prognosis an aggressive clinical management is advisable unless it occurs in the presence of particular conditions such as dichorionic–diamniotic pregnancies with only one fetus affected, where a conservative management is mandatory.

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