SIRENOMELIA WITH AGENESIS OF CORPUS CALLOSUM

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Sirenomelia is a very rare anomaly presented with fusion of the lower limbs. Genitourinary, neural tube, and vertebral anomalies are found in most cases. We report a case of sirenomelia with agenesis of corpus callosum, which has not been reported previously.

Keywords: Corpus callosum • mermaid • sirenomelia

Introduction

Sirenomelia (mermaid syndrome) is a rare anomaly of caudal region of the body, presented with fusion of the lower limbs.1 Renal agenesis, imperforated anus, as well as vertebral and neural tube defects have association with this ailment.2 Herein, we report a patient with sirenomelia and agenesis of corpus callosum.

Case Report

A 23-year-old pregnant nulliparous woman was referred to our hospital for ruling out intrauterine growth retardation of her fetus. The maternal medical and obstetric histories were uneventful, as well as the family history. She had a nonfamilial marriage. The ultrasonography was performed at 27 weeks of gestation, revealing oligohydramnios, opposition of lower extremities, agenesis of kidneys and bladder, and gross disparity between the biparietal diameter and the femoral length. The head circumference was 25 cm and crown-rump length was 30 cm, with an estimated weight of 825 g. The diagnosis of mermaid syndrome was made and the pregnancy was terminated for presence of congenital malformations incompatible with life. The fetus was evaluated by spiral computerized tomography (CT) scan and magnetic resonance imaging (MRI), after evacuation of the uterus. The fetus had Potter facies, imperforated anus, ambiguous external genitalia, single umbilical artery, and total fusion of the hypoplastic lower extremities. Both legs were in supination. The fetus had eight rudimentary fingers (Figure 1). The spiral CT scan revealed left fibular hemimelia and fused maldeveloped pubis bone. The ribs, scapulae, clavicles, and dorsal vertebrae were normal (Figure 2). The MRI showed that the lateral ventricles were separated by an abnormally high third ventricle, compatible with agenesis of corpus callosum (Figure 3). The kidneys, urinary bladder, and the internal genitalia were also absent. Karyotyping was not possible, since the fetus was immersed in formalin immediately after termination of pregnancy. The parents did not consent to autopsy the fetus.

Figure 1. Mermaid fetus, showing fused lower limbs, rudimentary foot, and deformed ears.
Sirenomelia with agenesis of corpus callosum

Discussion

Sirenomelia is a rare anomaly with an incidence of 1/100,000 births.³ It is characterized by fusion of the lower limbs; however, in most cases, renal, vertebral, anorectal, or neural tube defects also exist. The anomaly is more common in 46-XY fetuses and in one of two monozygotic twins.⁴ Maternal diabetes mellitus has an association with this congenital anomaly. In our case, however, there was no evidence of hyperglycemia in the mother before or during pregnancy. In the normal fetus, there are two umbilical arteries originating from the iliac arteries, which return blood to the placenta. In sirenomelia, like what we observed in our case, there is a single umbilical artery returning blood originating from the abdominal aorta. The aorta distal to this artery is rudimentary.⁵ The above-mentioned arterial anomaly, with distal hypoxia, could explain renal agenesis, sacro-vertebral, and lower limb malformations, but does not explain cardiac, cranial, and radial malformations commonly found in this anomaly.⁶ The renal and pubic bone defects in our case can be explained by this theory, but agenesis of corpus callosum could not. Another theory suggests that during the third week of gestation, a teratogen interferes with the formation of notochord, ensuing malformation of the caudal structures. Since notochord has an inducing effect for differentiation of the neural tube, its defect may lead to cranial defects.³ Hydrocephaly, cebocephaly, holoprosencephaly, and anencephaly have been noted in previous cases of sirenomelia. Our fetus had agenesis of corpus callosum (Figure 3). To the best of our knowledge, so far, agenesis of corpus callosum has not been reported with sirenomelia.

Early prenatal diagnosis of sirenomelia could be suspected by ultrasonography. In the case of severe oligohydramnios, MRI may reveal fetal anomalies precisely.

References