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Diversity of fetal sirenomelia depending on gestational age

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Objectives

Sirenomelia is an extremely rare congenital malformation characterized by varying degrees of lower limb fusion and associated visceral anomalies. This study explores the morphologic diversity and sonographic presentations of sirenomelia across different gestational ages (GA).

Methods

A retrospective analysis of five cases of sirenomelia was performed, including 1st and 2nd-trimester examinations. Detailed ultrasound (US) findings, associated anomalies, and diagnostic conditions were reviewed. GA-related differences in morphology, limb fusion pattern, and associated abnormalities were compared.

Results

No. 1: 11+4 gestational weeks (wks), sirenomelia type VI, omphalocele (OC), cleft lip/palate, single umbilical artery (SUA), bilateral renal agenesis (RA), oligohydramnios.

No. 2: 18+1 wks, sirenomelia type II/III, frontonasal dysplasia, OC, bilateral RA, anhydramnios, SUA, skeletal abnormalities (SA).

No. 3: 16+1 wks, oligohydramnios, intrauterine demise. Diagnosis of sirenomelia type I was made postnatally.

No. 4: 11+6 wks, sirenomelia type V, SUA, unilateral RA, contralateral multicystic dysplastic kidney, SA.

No. 5: 13+1 wks, dichorionic-diamniotic twin gestation, sirenomelia type V in one fetus, SUA, facial clefting, ventriculomegaly, SA, dextrocardia.

In three cases, a human genetic analysis was performed, and two fetuses were autopsied. All pregnancies were terminated, in case of gemini selective fetal reduction was performed.

Conclusion

Sirenomelia spectrum demonstrates distinct GA-related variability in sonographic appearance. Recognizing these developmental and diagnostic differences enhances early detection, classification, and counseling.



Figure 1. 3D volume of case no. 2 with sirenomelia type II/III



Figure 2. 3D volume of case no. 4 with sirenomelia type V.

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