

Perinatal diagnosis of congenital urogenital sinus abnormality

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Objective. Anomalies of the urogenital sinus, which is a transient feature of early human embryological development, are rare birth defects that most commonly occur within the context of congenital adrenal hyperplasia. These anomalies are characterized by the confluence of the urethra and vagina that creates a common channel; in this anomaly, the urogenital tract and the anorectal canal drain through separate perineal orifices. Urogenital sinus abnormalities commonly present as pelvic mass, hydrometrocolpos or ambiguous genitalia. Here, we report the case of a female newborn with congenital urogenital sinus abnormalities diagnosed soon after birth.

Materials and Methods. This female newborn was delivered at 38 weeks gestational age by vaginal delivery. Pregnancy was unremarkable, however prenatal ultrasound revealed

the presence of an abdominal mass of unknown origin. At delivery, the Apgar score was 8 at 1 minute and 9 at 5 minutes of life. Birth weight was 2890 gr (10-25th percentile). Based on the prenatal ultrasound finding, the presence of urogenital sinus abnormality was first hypothesized. Family history was negative for congenital malformations and disorders.

Results. By employing many postnatal imaging modalities (pelvic ultrasound, cystourethrography and genitography), the urogenital sinus abnormality was confirmed and the patient underwent surgery.

Conclusions. Given the normal adrenal function, the non-ambiguous genitalia and the absence of associated syndromes or malformations, we assume that the urogenital sinus abnormality of this patient can be ascribed to an arrest of normal cloacal development.