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Sonographic imaging and differential diagnosis of fetal perineal masses

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ABSTRACT

Objectives: Congenital perineal masses are a rare finding that may originate from the soft tissues of the perineum itself (perineal lipomas, lipoblastomas, hemangiomas, hamartomas), the sacrum (sacrococcygeal teratomas), the external genitalia (accessory scrotum or bifid scrotum, penoscrotal transposition). The present study describes our experience with fetal perineal masses and presents a clinical-sonographic flowchart for prenatal workup.

Material and methods: Perineal masses were diagnosed in 16 cases throughout five years. All cases underwent a thorough sonographic morphologic fetal evaluation, and the parents were counseled

by a multidisciplinary team. Examinations were performed with E8 or E10 expert machines. Transvaginal and transabdominal transducers were used according to the gestational age and fetal presentation.

Results: A total of 16 cases were referred for a targeted scan for a perineal finding between 20 and 26 weeks of gestation. Definitive diagnosis was provided after 20 weeks, following visualization of the target sign. All couples were offered genetic counseling and amniocentesis. Genetic analysis revealed abnormal results in three cases — one case of down syndrome and two consecutive cases of Townes–Brocks syndrome in the same couple. Postnatal clinical examination of the neonate or the abortus in case of termination of pregnancy (4 cases) up confirmed the diagnosis in all cases.

Conclusions: Targeted, structured prenatal anatomical scan in fetuses presenting with perineal masses may aid in the prenatal differential diagnosis and enable appropriate genetic analysis, prenatal counseling, and postnatal treatment.

Key words: perineal masses; fetus; fetal diagnosis; ultrasound

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INTRODUCTION

Congenital perineal masses are a rare finding that may originate from the soft tissues of the perineum itself (perineal lipomas, lipoblastomas, hemangiomas, hamartomas), the sacrum (sacrococcygeal teratomas), the external genitalia (accessory scrotum or bifid scrotum, penoscrotal transposition). Findings associated with anorectal malformations and persistent cloaca may resemble a perineal mass, and therefore the differential diagnosis is essential. Most of the literature cases, specifically those diagnosed prenatally, are case reports [1–11]. Although the fetal perineum is not routinely assessed as a part of the fetal anatomical scan, we have implemented it to all fetal examinations referred for targeted scans in recent years. The present study describes our experience with fetal perineal masses and presents a clinical-sonographic flowchart for prenatal workup.

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MATERIAL AND METHODS

The local Ethical Committee approved the study. The committee provided a waiver of informed consent, as this was a retrospective study.

Perineal masses were diagnosed in fifteen cases throughout five years. All cases underwent a thorough sonographic morphologic fetal evaluation, and the parents were counseled by a multidisciplinary team including a neonatologist, a geneticist, and a pediatric surgeon and urologist. The fetal perineum was assessed in the axial plane to locate the anal mucosa and sphincter. A midsagittal plane assessed the spine, the conus medullaris, and the sacral vertebra.

Examinations were performed with E8 or E10 expert machines (GE Healthcare, Kretz Ultrasound, Zipf, Austria), equipped with an abdominal 4–8-MHz curvilinear transducer. Transvaginal and transabdominal transducers were used according to the gestational age and fetal presentation.

RESULTS

A total of 16 cases were referred for a targeted scan for a perineal finding between 20 and 26 weeks of gestation. Definitive diagnosis was provided after 20 weeks, following visualization of the target sign. All couples were offered genetic counseling and amniocentesis. Genetic analysis revealed abnormal results in three cases — one case of down syndrome and two consecutive cases of Townes–Brocks syndrome in the same couple. Postnatal clinical examination of the neonate or the abortus in case of termination of pregnancy (4 cases) up confirmed the diagnosis in all cases.

Prenatal sonographic findings, genetic workup, and final postnatal diagnoses are presented in Table 1 and Figures 1–3.

DISCUSSION

This study reports our experience with the prenatal diagnosis of fetal perineal masses. We propose a clinical,

Table 1. Prenatal sonographic findings, genetic workup, and final postnatal diagnoses

Case	Prenatal sonographic findings	Genetic workup	Postnatal final diagnosis
1	Non-visualization of the target sign Perineal lump Tethered cord	Normal CMA	Anal atresia
2	Non-visualization of the target sign Perineal lump Single kidney	Normal CMA	Anal atresia
3	Non-visualization of the target sign Perineal lump Single kidney, single umbilical artery. Tetralogy of Fallot with absent pulmonic valve	Normal CMA	Anal atresia
4	Non-visualization of the target sign Perineal lump	Normal CMA	Anal atresia
5	Non-visualization of the target sign Perineal lump	Trisomy 21	Anal atresia
6 + 7	Non-visualization of the target sign Perineal lump hypoplastic-dysplastic kidneys bifid thumb, accessory tragus (Two consequent pregnancies)	Townes Brocks syndrome (OMIM #107480)	Anal atresia
8	Sacrococcygeal teratoma (type 1)	Normal CMA	Sacrococcygeal teratoma (type 1)
9	Sacrococcygeal teratoma (type 1)	Normal CMA	Sacrococcygeal teratoma (type 1)
10	Sacrococcygeal teratoma (type 4)	Normal CMA	Sacrococcygeal teratoma (type 3)
11	Double penis	Normal CMA	Double penis
12	Infra scrotal, perineal mass	Normal CMA	Perineal lipoma
13	Infra scrotal, perineal mass	Normal CMA	Perineal lipoma
14	Infra scrotal, perineal mass	Normal CMA	Perineal lipoma
15	Infra scrotal, perineal mass	Normal CMA	Perineal lipoma
16	Infra scrotal, perineal mass	Normal CMA	Perineal lipoma

CMA — chromosomal microarray analysis

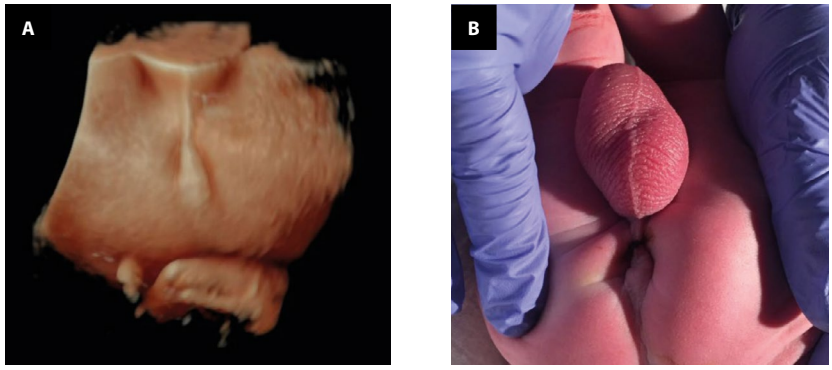

Figure 1. Pre- and postnatal imaging of a fetus at 30 weeks of gestation. The target sign was not visualized, consistent with anal atresia; **A**. 3D rendering of the fetal perineum demonstrating a perineal mass caudal to the scrotum and **B.** its corresponding postnatal image

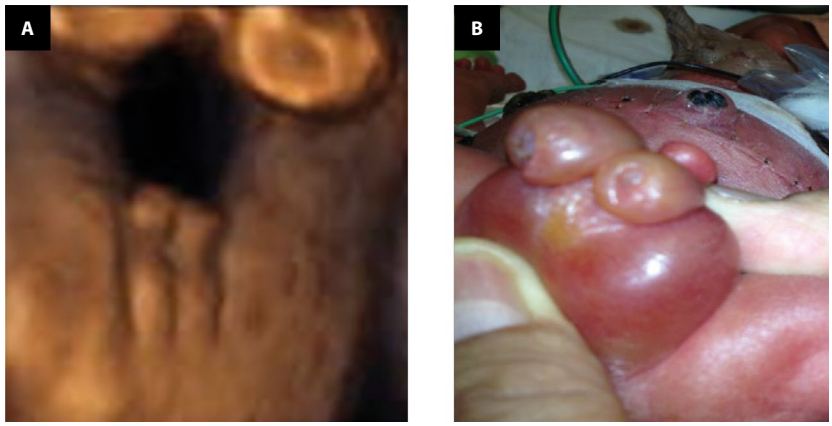

Figure 2. 3D rendering of the fetal perineum obtained at 22 weeks of gestation and its corresponding postnatal image in a case of a double penis

sonographic assessment flow chart based on prenatal and postnatal findings (Fig. 4).

The target sign, representing the fetal anal musculature and mucosa, plays a critical role in the clinical-sonographic differential diagnosis approach to fetal perineal lumps. According to our results, non-visualization of the target sign associated with a midline perineal lump located below the scrotum seems to represent poor development of the perineum and is related to high anorectal malformations in male fetuses. It should be noted that perineal masses can be diagnosed during the early second trimester; however, the target sign is not visualized before 20 weeks of gestation [12].

Assessment of the genitalia should follow the visualization of the target sign. As reported by others and demonstrated in the present series, perineal findings may resemble a male phallus. Application of Doppler flow and imaging of fetal urination through the normal penis with no urination through the perineal mass may aid in the differential diagnosis between rare cases of penoscrotal transposition, double penis, and perineal lipomas or other masses originating from the perineal soft tissues (Fig. 5). Sacrococcygeal teratomas originate from the coccyx and sacrum, and assessment of the sacrum is the third step in the prenatal workup of perineal masses, as they may be associated with sacral malformations and sacral agenesis.

Accurate prenatal diagnosis of perineal lumps is essential for proper prenatal counseling regarding the postnatal course and targeted genetic analysis. Our series demonstrates that anal atresia is associated with trisomy 21 and other syndromes. Similarly, urogenital malformations, precisely a penoscrotal inversion, were associated with a distal deletion of 13q [13]. Genetic syndromes associated with sacrococcygeal teratomas include autosomal dominant Currarino syndrome (OMIM #176450). The genotype--phenotype correlation is variable, although traditionally reported as a triad of a presacral mass, sacral agenesis, and anal atresia. For example, in a series by Kochling et al. [14], ten out of 23 patients from 9 families with mutations in the homeobox gene HLXB9 were asymptomatic. The radiologic

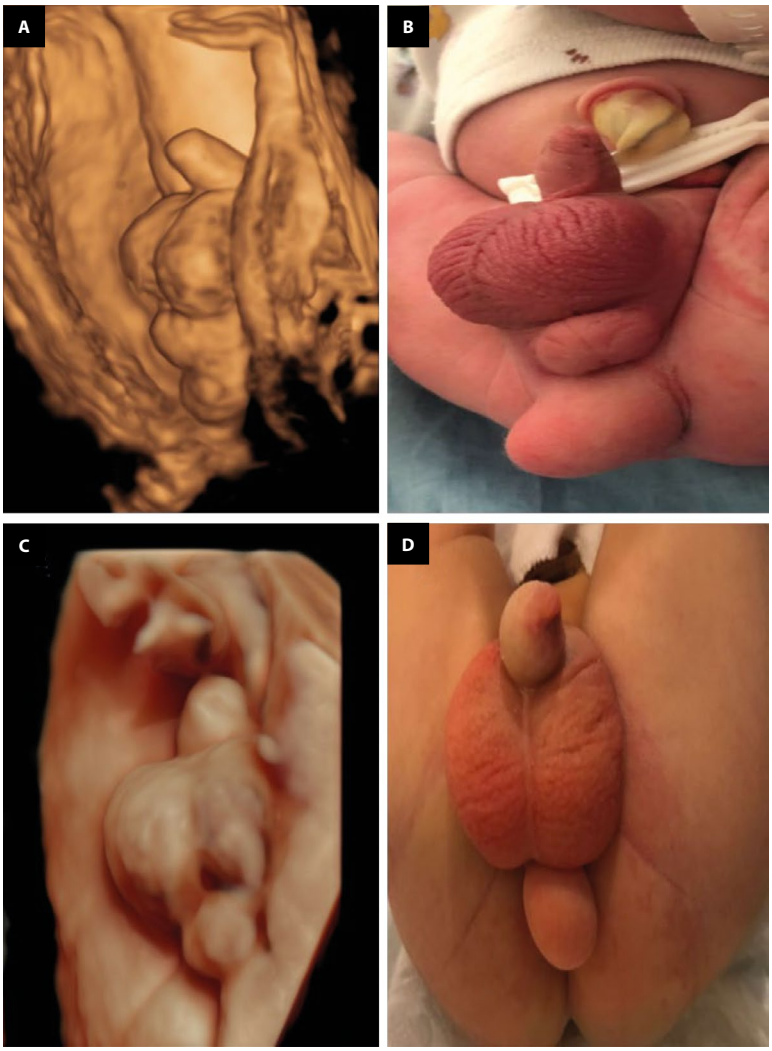

Figure 3. 3D rendering of the fetal perineum and its corresponding postnatal image of perineal lipomas; **A**, **B.** Two lipomas located below the scrotum (arrows); **C**, **D.** One lipoma located below the scrotum (arow)

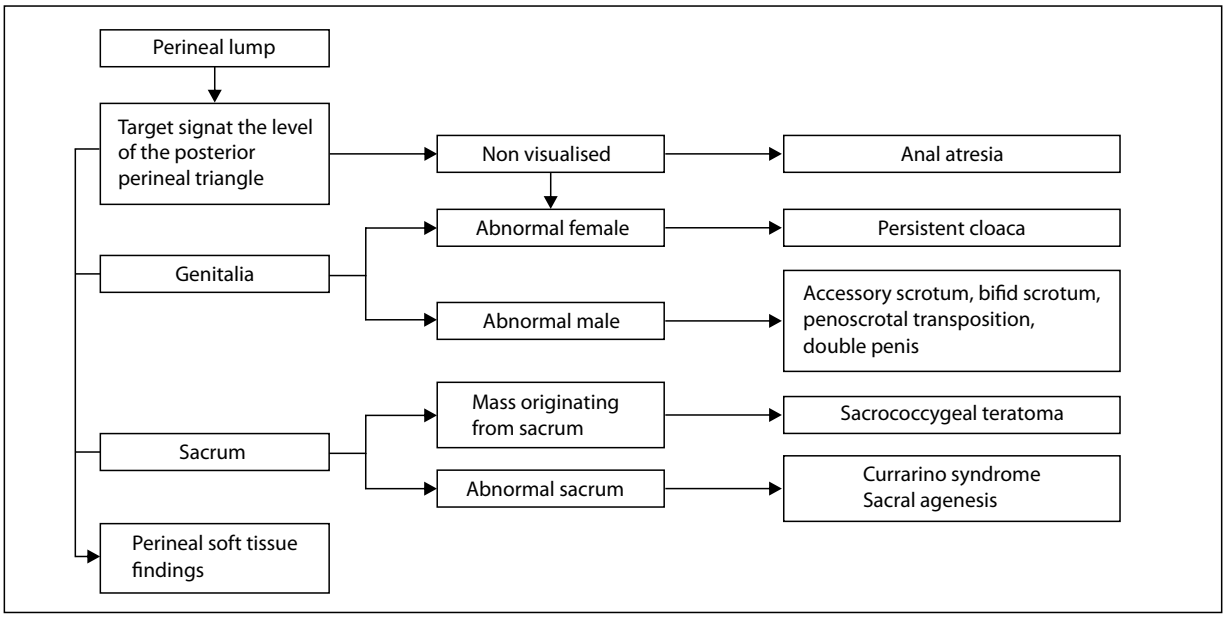

Figure 4. A flowchart for prenatal sonographic workup of a perineal mass

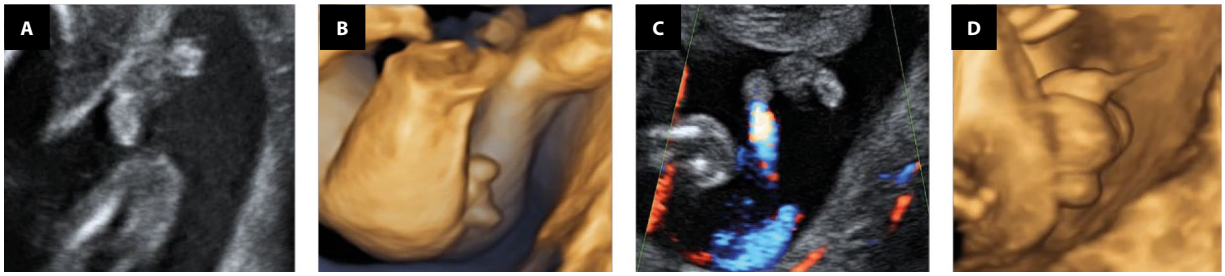

Figure 5. A clinical, sonographic approach to a fetal perineal lump. The target sign was visualized at the posterior perineal triangle; **A**. 2D midsagittal view demonstrating the scrotum and upper and lower findings that resemble a male phallus (*); **B**. 3D rendering of the fetal perineum demonstrating the findings described in **A**; **C**. Doppler flow demonstrates urination from the upper mass, i.e., the penis, while no flow is demonstrated originating from the lower mass; **D**. 3D rendering demonstrating fetal urination as described in; the final diagnosis was a perineal lipoma

investigation did reveal characteristic phenotypic features in all patients; however, the complete triad was found only in eight of the 23 patients.

CONCLUSIONS

To summarize, the diversity of fetal perineal malformations requires an accurate diagnosis based on a targeted, structured anatomical scan of the perineum to provide counseling regarding the postnatal outcome.

Conflict of interest

All authors have declared all potential conflicts of interest

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