Perineal Groove: A Rare Congenital Midline Defect of Perineum

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Abstract

Perineal groove is a rare congenital malformation that is characterized by an exposed wet sulcus with nonkeratinized mucous membrane that extends from the posterior vaginal fourchette to the anterior ridge of the anal orifice. This condition is one of the uncommon anomalies of urogenital/anogenital region that is unknown to many clinicians. Although, this condition may be self-resolved before the age of 2 years, this nonepithelialized mucous membrane can pose the risk of local irritation and infection, urinary tract infection, and the possibility of nonself-resolved condition that eventually needs surgical correction. Only a few reported cases (n = 23) were found in current medical literatures. This lesion could be misdiagnosed as contact dermatitis, trauma, or even sexual abuse. Therefore, recognition of the congenital perineal groove at birth is important for the health care providers to deliver an appropriate parental counseling and appropriate follow-up.

Keywords

► congenital anomaly
► perineal groove
► perineal defect
► newborn

Congenital perineal groove is characterized by an exposed erythematous nonepithelized mucous membranes that resembles exposed wet erythematous sulcus, inflammation, or rupture of the midperineum area. This lesion extends vertically downward from the posterior portion of the vaginal fourchette to the anterior rim of the anus.1–3 This lesion resembles failure of mid-perineum fusion or failure of mid-perineum skin epithelialization.3

Case Presentation and Management

Case 1

At 37 weeks gestational age (GA) a white female with birth-weight (BW) of 3,450 g was delivered by cesarean section (CS) due to recurrent CS and placenta previa. Apgar scores were 8 and 9 at 1 and 5 minutes, respectively. Infant was admitted to the neonatal intensive care unit (NICU) for suspected imperforated anus. Infant’s mother was a 25-year-old gravida 3 and para 3 (G3P3) with good prenatal care. All prenatal tests were negative and there was no history of smoking, drinking alcohol, or abusing substances. Placental histopathology showed mild placentomegaly, acute chorioamnionitis without funisitis, and placenta previa. There was no family history of congenital anomalies.

Physical examination of the infant was normal except for the perineal area that showed a lesion, which was suspicious for congenital or iatrogenic perineal rupture or imperforated anus. The perineal defect was noted stretching vertically upward from 12 o’clock position of the anal rim toward the posterior vaginal fourchette, with a wet groove–like unkeratinized erythematous mucosal lesion (Fig. 1). Urethra and vaginal orifices were intact and within appropriate position. Infant did not show any signs of urination problem. The diagnosis of imperforated anus was ruled-out with normal anorectal examination and infant was stooling from the well-positioned anal opening located posterior to the lesion. The...
Congenital perineal groove is a rare and benign malformation. It is mostly presented in female infants as an isolated anomaly that could be associated with four-vessel umbilical cord and perineal groove anomaly.

Pathogenesis of perineal groove remains unclear. However, embryological mechanisms have been proposed as follows: (1) a relic of the open cloacal duct, (2) midline fusion failure of medial genital folds between the perineal raphe and the vestibule, and (3) urorectal septum developmental defect during cloacal embryological stages at 5th to 8th week of GA. Given that the perineal groove anomaly was also reported in one male infant, this congenital anomaly can be derived during the embryological development stage of external genitalia. External genitalia of both sexes are developed from the genital tubercle, genital folds, and labioscrotal folds. Therefore, failure of labioscrotal folds fusion to form perineal raphe will cause perineal groove to occur.

Histological reviews of the resected area varied from a nonkeratinized stratified squamous epithelium without sebaceous glands or sweat glands or hair follicles to a simple columnar or stratified columnar or cuboidal epithelium of a rectal type mucosa with intervening area of nonkeratinized stratified squamous epithelium. These findings resembled anorectal transitional zone epithelium, which implied that this congenital anomaly was underdiagnosed and underestimated as it may be unrecognized earlier at birth, and/or later misdiagnosed as a diaphragm, contact dermatitis, or trauma.

In 1968, Stephens described perineal groove as a congenital malformation that consists of three features: (1) a wet groove in midperineum between the fourchettes and the anus; (2) normal formation of the vestibule including the urethra and vagina; and (3) hypertrophy of the minoral tails that skirt the perineum and course posteriorly to join at the anus or to surround it.

Discussion

Congenital perineal groove is a rare and benign malformation. It is mostly presented in female infants as an isolated anomaly. So far only 23 cases have been reported in medical literatures. In rare cases, it may be associated with other regional anomalies of anogenital and/or urogenital system such as anteriorly placed anus, ectopic anus, prolapsed anus, or urinary tract malformation. Both of our patients were delivered within 12-month period and were diagnosed at birth. It is likely that the incidence of this congenital anomaly is underestimated as it may be unrecognized earlier at birth, and/or later misdiagnosed as a diaphragm, contact dermatitis, or trauma.

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Patients with this congenital malformation are at an increased risk of urinary tract infection, inflammation, or infection of the nonepithelialized mucosa, fecal continence, or incontinence and other associated regional anomaly. This lesion may be self-resolved and completely epithelialized by the age of 1 to 2 years. Surgical correction can be done.
mainly for cosmetic reason, if the lesion is not epithelialized by the age of 2 years. However, there are also some reports that the lesion may need surgical correction earlier than the age of 2 years for clinical reasons such as repeated inflammation or infection of the nonepithelialized area from exposure to mucous drainage, wet secretion from the urethra, vagina, or anal sites.

Because this lesion resembles rupture or inflammation of perineum region, failure to recognize this lesion at birth will lead to misdiagnosis such as infection, irritant dermatitis, ulcerated hemangioma, lichen sclerosis, trauma, or even sexual abuse.

Conclusion

This uncommon benign congenital malformation tends to be self-epithelialized. However, surgical referral is warranted if it involves other regional anomalies, recurrent infection, or ulceration of the perineal groove, failure of epithelialization after the age of 2 years or for a cosmetic reason. Early recognition of congenital perineal groove at birth is important for health care providers to deliver an informed parental counseling, appropriate follow-up, and to prevent an unnecessary surgical or medical intervention.

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Contributors’ Statement
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Permission for Photographs
All photographs provided in this case report have consent forms signed by patients’ parents or the legal guardians for the purpose of medical literature publication and education. This case report which included the photographs has received an Institutional Board Review approval from the University of Tennessee Health Science Center and the Regional One Health Hospital to conduct the study.

References