Perineal Groove: An Anorectal Malformation Network, Consortium Study

Inbal Samuk, MD[1,2], Eva E. Amerstorfer, MD[3], Maria Fanjul, MD[4], Barbara D. Iacobelli, MD[5], Gabriele Lisi, MD[6], Paola Midrio, MD[7], Anna Morandi, MD[8], Eberhard Schmiedeke, MD[9], Pernilla Stenstrom, MD[10], and Christien Sleeboom, MD[11]

Objective To review the Anorectal Malformation Network experience with perineal groove (PG) focusing on its clinical characteristics and management.

Study design Data on patients with PG managed at 10 participating Anorectal Malformation Network centers in 1999-2019 were collected retrospectively by questionnaire.

Results The cohort included 66 patients (65 females) of median age 1.4 months at diagnosis. The leading referral diagnosis was anal fissure (n = 20 [30.3%]): 23 patients (34.8%) had anorectal malformations. Expectant management was practiced in 47 patients (71.2%). Eight (17%) were eventually operated for local complications. The median time to surgery was 14 months (range, 3.0-48.6 months), and the median age at surgery was 18.3 months (range, 4.8-58.0 months). In the 35 patients available for follow-up of the remaining 39 managed expectantly, 23 (65.7%) showed complete or near-complete self-epithelization by a mean age 15.3 months (range, 1-72 months) and 4 (11.4%) showed partial self-epithelization by a mean age 21 months (range, 3-48 months). Eight patients showed no resolution (5 were followed for ≤3 months). Nineteen patients (28.7%) were primarily treated with surgery. In total, 27 patients were operated. Dehiscence occurred in 3 of 27 operated patients (11.1%).

Conclusions PG seems to be an underestimated anomaly, frequently associated with anorectal malformations. Most cases heal spontaneously; therefore, expectant management is recommended. When associated with anorectal malformations requiring reconstruction, PG should be excised in conjunction with the anorectoplasty. *(J Pediatr 2020;222:207-12)*.

Perineal groove (PG) is a congenital well-demarcated moist sulcus of the perineum lined with mucous membrane that extends from the posterior fourchette to the anterior margins of the anus (**Figure 1**).[1-5] These typically resolve by spontaneous epithelization by age 2 years; therefore, most reports support expectant management unless complications arise.[2,4-8] Others, however, advocate surgery on cosmetic grounds or to prevent infection of the groove or external genitalia.[1,9] Surgical intervention involves simple superficial excision of the mucus membrane with primary closure.

PG is considered to be a rare condition and was included in the group of rare miscellaneous anorectal anomalies in the international classification of anorectal anomalies proposed by Stephen.[10] PG is often misdiagnosed as anal fissure, perineal erosion/ulcer, traumatic tear, ulcerated hemangioma, diaper dermatitis, or even sexual abuse with suspected anal penetration and a tear extending to the vulva.[2,4,5,11-14] Consequently, many patients are subjected to unnecessary diagnostic procedures and treatments. The aim of this study was to review the experience with PG of participating centers of the Anorectal Malformation Network (ARM-Net), outline the clinical characteristics of this anomaly, and readdress general principles on approach and management.

Methods

The cohort included patients with PG who were managed at ARM-Net participating medical centers. PG is not recorded on the ARM-Net database. Therefore, data were collected retrospectively using an ad hoc questionnaire that was sent on-line in January 2019 to all members of the ARM-Net Consortium. The following variables were assessed: age at diagnosis, provisional diagnosis of referral, associated malformations, extension and limits of the groove, management strategies, indications for surgery, pathology, complications, and outcome.

ARM	Anorectal malformation
ARM-Net	Anorectal Malformation Network
PG	Perineal groove

From the [1]Department of Pediatric Surgery, Schneider Children's Medical Center of Israel, Petach Tikva, [2]Sackler Faculty of Medicine, Tel Aviv University, Tel Aviv, Israel; [3]Department of Pediatric and Adolescent Surgery, Medical University of Graz, Austria; [4]Department of Pediatric Surgery, Gregorio Marañón University General Hospital, Madrid, Spain; [5]Department of Medical and Surgical Neonatology, Newborn Surgery Unit, Bambino Gesù Children's Hospital-Research Institute, Rome, Italy; [6]Pediatric Surgery Department, G. d'Annunzio University of Chieti-Pescara, Chieti, Italy; [7]Pediatric Surgery, Mother and Child Department, Cà Foncello Hospital, Treviso, Italy; [8]Department of Pediatric Surgery, Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, Milano, Italy; [9]Department of Pediatric Surgery and Urology, Centre for Child and Youth Health, Klinikum Bremen-Mitte, Bremen, Germany; [10]Department of Pediatric Surgery, Skåne University Hospital Lund and the Institution of Clinical Research, Lund University, Lund, Sweden; and [11]Department of Pediatric Surgery, Emma Children's Hospital, Amsterdam UMC, University of Amsterdam and VU Amsterdam, Amsterdam, The Netherlands

The authors declare no conflicts of interest.

https://doi.org/10.1016/j.jpeds.2020.03.026

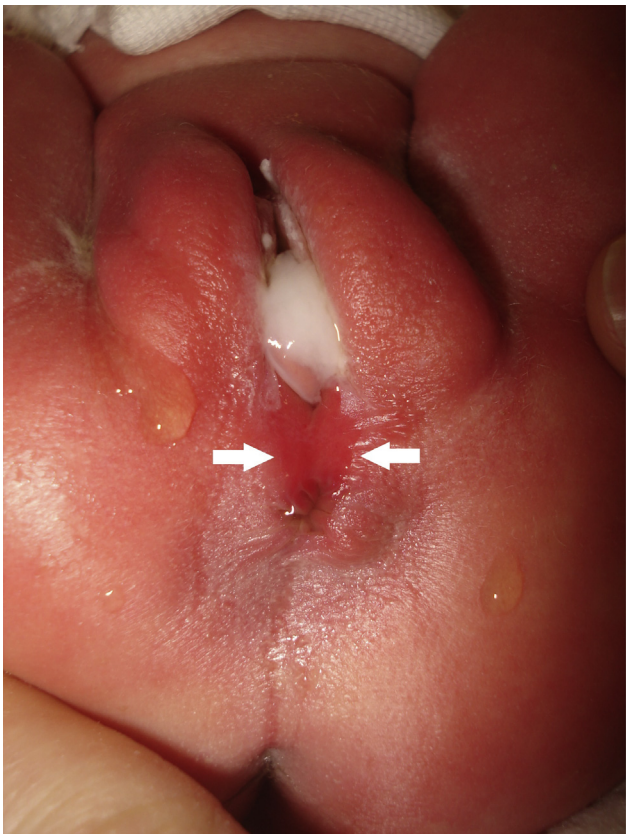

Figure 1. PG at birth between anus and fourchette.

Table I. Clinical characteristics of patients with PG

Characteristics	No. (%)
Sex	
Female	65
Male	1
Perineal extension of groove (F)	
Anus to posterior fourchette	35 (53.8)
Anus to midperineum	20 (30.7)
Fourchette to midperinum	10 (15.3)
Associated ARMs	23 (34.8)
Rectoperineal fistula	22
Anal stenosis	1
Associated genitourinary malformations	3 (4.5)
Referral diagnosis	
Anal fissure	20 (30.3)
The associated ARMs	11 (16.6)
Ectopic anus*	7 (10.6)
PG	6 (9)
Other†	2 (3)
"Bizarre looking anus"	2 (3)
Not specified	18 (27.3)
Management	
Expectant	47 (71.2)
Attempted expectant then surgery	8/47
Primary surgery	19 (28.7)
ASARP and excision of PG	8
Excision of PG alone	4
Y-V anoplasty and PG excision	4
Excision during other reconstruction‡	2
PSARP and excision of PG	1

ASARP, anterior sagittal anorectoplasty; *PSARP*, posterior sagittal anorectoplasty.
*Anus was normotopic.
†Noticed during clinical evaluation of unrelated condition.
‡Repair of urogenital sinus, approximation of divergent perineal muscles.

Results

The cohort included 66 patients (65 females) who were managed at 10 colorectal centers between 1999 and 2019. Of them, 34 were managed at a single center. Their characteristics are shown in Table I. The median age at diagnosis was 1.4 months (range, 1 day-58 months). Limits of the groove in the female patients were as follows: anus/perineal fistula to posterior fourchette in 35 patients (53.8%), anus to midperineum in 20 (30.7%), and fourchette to mid perineum in 10 (15.3%). In the male patient, the PG was located 1 cm ventral to the anus. Twenty-three patients (34.8%) had associated anorectal malformations (ARMs): rectoperineal fistula in 22 and anal stenosis in 1. Genitourinary malformations were recorded in 3 female patients and included vesicourethral reflux grade III, urogenital sinus, and imperforate hymen.

The leading referral diagnoses were anal fissure in 20 patients (30.3%) and ARM (without recognition of the PG) in 11 (16.6%). One patient misdiagnosed with an anal fissure received treatment with fitostimoline cream, an aqueous extract of *Triticum vulgare* to facilitate the wound healing processes. Only 6 patients (9%) were diagnosed correctly with PG by the primary care physician.

Five patients were lost to follow-up. In the 61 remaining patients, the mean duration of follow-up from presentation to the last clinic visit was 27.6 months (range, 1-219 months).

Expectant management was practiced in 47 patients (71.2%). Eight (17%) eventually underwent excision of the PG because of persistent mucosal discharge with local irritation. Surgery was performed after a median follow-up time of 14 months (range, 3.0-48.6 months) from presentation, at a median patient age of 18.3 months (range, 4.8-58.0 months). Surgery consisted of removing the strip of mucosa and suturing the perineal skin in the midline. Of the remaining 39 patients, 35 were available for follow-up: 23 (65.7%) showed complete or near-complete self-epithelization by a mean age of 15.3 months (range, 1-72 months), and 4 (11.4%) showed partial self-epithelization by a mean age of 21 months (range, 3-48 months) (Figure 2). No resolution was observed in 3 patients with a mean follow-up of 33 months (range, 12-48 months) and in another 5 patients who had been followed for only 3 months or less (mean, 2.6 months) at the time the study was conducted.

Nineteen patients (28.7%) were primarily managed by surgery. The median age at surgery was 6.9 months (range, 0.1-58.0 months). The PG was associated with rectoperineal fistula in 14 patients (73%) and with urogenital sinus in 1 patient. Indications for surgery included excision of PG in conjunction with anorectoplasty in 13 patients (68.4%),

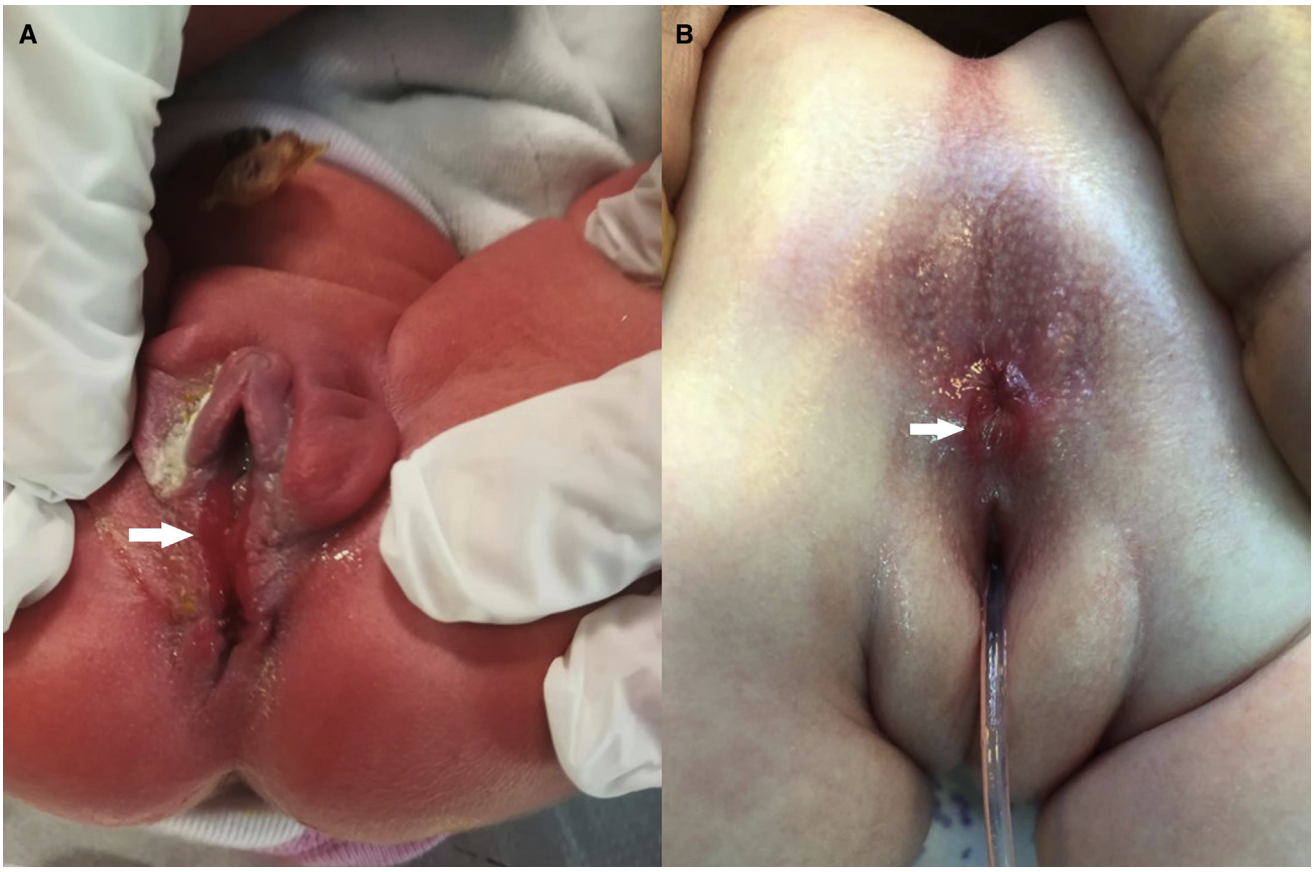

Figure 2. Patient with perineal fistula and PG. **A,** At birth. **B,** At age 48 days, before mini-posterior sagittal anorectoplasty. The PG shows nearly complete epithelization.

persistent mucosal discharge with local irritation in 4 (21%), and excision during other reconstructive surgery in 2 (10.5%): repair of the urogenital sinus (n = 1) and approximation of divergent parasagittal muscle fibers noted on muscle stimulation (n = 1). Types of surgical procedures are listed in **Table I**.

Because 34 of the 66 patients (51%) were managed at a single center, the type of management was further analyzed between these patients and the other 32 patients managed at different centers. We found that 12 of the 34 single-center patients (35.2%) underwent primary operative management: 8 during reconstructive surgery and 4 in an isolated procedure. The other 22 (64.7%) were managed expectantly, of whom 8 (36.3%) eventually required surgery for local complications, mainly during the first one-half of the study period. Among the 32 patients managed at different centers, 7 (21.9%) were operated primarily, in conjunction with reconstructive surgery, and 25 (78.1%) were managed expectantly, none of whom required surgery at any time during follow-up.

Of the total 27 patients in the cohort treated surgically, 3 had postoperative dehiscence: 1 after anterior sagittal anorectoplasty with excision and 2 (initially managed expectantly) after excision alone performed at mean age of 25.1 months.

The pathology report was available for 14 operated patients. Findings included nonkeratinized squamous epithelium in 8, keratinized squamous epithelium in 3, both keratinized and nonkeratinized squamous epithelium in 2, and squamous epithelium (further description not available) in 1.

Of 9 patients without ARM, for whom we had data on constipation, only 2 had chronic constipation that required stool softeners. Continence data were available for only 3 patients without associated ARM who reached toilet training age and all had voluntary bowel movements with no soiling.

Discussion

Our search of the English literature from 1968 to 2019 yielded 21 articles describing 68 cases, including a single series of 26 cases[15] (**Table II**). According to the literature, many patients with PG are misdiagnosed early during their course and some are subjected to unnecessary diagnostic tests and medical interventions, including skin biopsies, corticosteroids, barrier creams, antifungal creams, and even laser treatment for suspected ulcerated hemangioma.[2,4,13] In the present cohort, the leading referral misdiagnosis was anal fissure (in 30.3% of patients); 1 patient with a misdiagnosed anal

Table II. Summary of cases of PG reported in the English literature

Cases	Source	Year	No. and sex	Age at diagnosis	Associated anorectal condition/anomaly	Associated genitourinary features/anomalies	Management	Outcome/follow-up
1	Stephens[16]	1968	4 F	NA	–	Hypertrophy of mineral tails	Expectant (n = 1) NA (n = 3)	Healed*/9 mo NA (n = 3)
2	Gellis et al[17]	1977	4 F	NA	Anterior mucosal prolapse (n = 2)	–	NA	NA
3	Shigemoto et al[10]	1978	1 F	4 y	Anterior mucosal prolapse	Hypertrophy of mineral tails	Expectant	NA
4	Kadowaki et al[5]	1983	1 F	2 mo	Anterior mucosal prolapse	–	Expectant	Healed*/1 y
5	Abdel Aleem et al[18]	1985	2 F	†	Rectoperineal (n = 1)	–	Surgery	NA
6	Chatterjee et al[6]	2003	1 M	7y	Missing musculature of anterior anal canal with anterior mucosal prolapse	Penoscrotal hypospadias, bifid scrotum	Surgery	Healed/12 mo
7	Aslan et al[19]	2005	1 M	Newborn	Anal atresia associated with Currarino syndrome	Penoscrotal inversion; hypospadias; right renal agenesis	Surgery	Healed/21 mo
8	Mullassery et al[1]	2006	1 F	6 mo	–	–	Surgery‡	Healed/NA
9	Sekaran et al[20]	2009	1 F	Newborn	–	–	Expectant	Healed*/12 mo
10	Verma and Wollina[2]	2010	1 F	2.5 y	–	Prominent labia majora	Expectant	NA
11	Esposito et al[21]	2011	6 F	3 y median	–	–	Surgery§	3 dehiscence
12	Siruguppa et al[9]	2012	1 F	Newborn	Anterior anus	–	Expectant	Epithelized starting at age 2 mo
13	Diaz et al[13]	2014	2 F	4 mo, 6 mo	–	–	Expectant	Stable/12 mo (n = 1) Partially healed/8 mo (n = 1)
14	Senanayake et al[14]	2014	1 F	26 mo	–	–	NA	NA
15	Hunt et al[8]	2016	1 F	Newborn	–	–	Expectant	Healed*/6 mo
16	Harsono and Pourcyrous[11]	2016	2 F	Newborn	–	–	Expectant	Stable/4 mo (n = 1) Healed*/1 y (n = 1)
17	Garcia-Palacios et al[4]	2017	5 F	Mean 14 mo	Pelvis syndrome (n = 1)	External genitalia malformation (n = 1) Vesicorenal abnormalities (n = 1)	Expectant (n = 5)	Mean FU 7 mo Stable (n = 3) Partial epithelization (n = 2)†
18	Cheng et al[3]	2018	2 F	Newborn	–	–	Expectant	NA
19	Wojciechowski[22]	2019	1 F	Newborn	–	–	Expectant	Stable/1 mo
20	Boutsikou et al[12]	2019	4 F	Newborn	–	–	Expectant (n = 3) Surgery (n = 1)¶	1-Signs of epithelization/1 wk; 2- NA; 1 healed (surgery)
21	Ihn et al[15]	2019	25 F / 1 M	1.5 m median	"Imperforate anus" (n = 1)		Expectant	Partial healing in 71% of patients >2 y of age
Total		–				68 (n = 65 F, 3 M)		

NA, not available; *FU*, follow-up.
*Healed fully epithelialized.
†During the first year of life.
‡For cosmetic reasons.
§For recurrent infection of PG (n = 6), external genitalia (n = 3), urinary tract infection (n = 1).
¶Suture by obstetrician for a misdiagnosis of perineal tear.

fissure had been treated with topical preparation with active ingredients in an attempt to accelerate healing. A rapid and correct diagnosis of PG during the first week of life was achieved in only 9% of patients, all by neonatologists trained in a center with a dedicated colorectal service. This further emphasizes the importance of raising awareness of PG among primary care physicians and neonatologists through workshops and courses on ARMs.

Stephens described three major features common to PG: normal formation of urethra and vagina, hypertrophic minoral tails surrounding the sulcus, and a wet sulcus (either deep or shallow) in the perineum between the fourchette and the anus.[16] PG may be complete, extending from the posterior fourchette to the anterior margins of the anus, or incomplete, ending in the midperineum from either the anus or vagina.[3,15] In the present study, 53.8% of the cases were complete and the rest were incomplete, mostly from anus to midperineum.

In the English literature, only 3 cases of PG have been reported in male patients, in association with hypospadias in 2 of them.[6,15,19] The male patient in our cohort did not have hypospadias.

PG is considered to be generally asymptomatic and self-resolving.[3,4] Therefore, expectant management is recommended for uncomplicated cases until the sulcus heals by self-epithelization, usually by 2 years of age, but sometimes as late as age 4 years of age.[3-5,9-11,20] However, in the present study, 18% of patients had symptoms, suggesting that symptomatic PG is more common than previously presented. Moreover, this rate might be an underestimation, because data on symptoms were lacking in patients who were primarily operated for associated ARMs. According to some reports, the development of local complications including persistent inflammation, mucous discharge, and infection, in addition to increased risk of urinary tract infection will facilitate a surgical procedure.[4,8,20,21] This finding was also true in the present cohort, in which attempts at expectant management were terminated in 8 patients when local complications, mainly unremitting mucosal discharge with persistent local irritation, were identified. Nevertheless, in 65.7% of our patients, complete or near-complete self-epithelization occurred by a mean age of 15.3 months, and only 1 patient reached 4 years of age with no signs of resolution. Because 62% of the patients with no resolution had a very short follow-up, we could not calculate the true rate of complete resolution in our cohort.

PG is commonly reported as an isolated condition that coexists with a normotopic anus.[6] ARM was reported in 2.9% of cases in the English literature (**Table II**). In the present series, however, the rate of associated ARMs was 34.8%. This finding may partially represent a selection bias because patients were managed by pediatric surgeons in dedicated ARMs centers. The malformations were almost exclusively rectoperineal fistulas. This association affected the management strategy, because 59% of the PGs associated with rectoperineal fistula were excised in conjunction with anorectoplasty, without a prior attempt at expectant management.

When the anterior wall of the anus is continuous with the mucosal lining of the groove, patients may be at risk of prolapse with straining.[5,10] Anterior mucosal prolapse was reported in 7.4% of cases in the literature.[5,6,10,17] occasionally as a consequence of missing musculature in the anterior anal canal (**Table II**). This occurrence was not seen in any of our cases.

Some authors reported that surgery was performed because of inflammation/infection of the groove or at the request of parents on cosmetic grounds.[1,9,20] However, in the present cohort, primary excision of the PG was usually part of the repair of an associated ARM. In cases of isolated PG, surgical intervention consisted of a simple superficial excision of the mucus membrane with primary closure. The most worrisome complication of surgery was perineal dehiscence. Esposito et al described 6 female patients who underwent PG excision of whom 3 had complications of dehiscence.[21] Dehiscence was reported in 3 of 27 operated patients (11.1%) in our cohort, 2 of whom underwent excision alone. Both were >2 years of age at surgery, which may have been a contributory factor.

The embryonal origin of PG is not fully understood, and various theories have been offered over the years. In our cohort, histologic study of the resected area revealed almost exclusively squamous epithelium, either keratinized or nonkeratinized. Based on a study of the perineum in premature babies, Stephens suggested that defects of development of the uroanal septum, in addition to imperfect medial migration of the genital folds, may lead to the formation of PG.[16] The perineum remains cleft, with the floor being the septum and the sides, the genital folds. This theory is supported by reports of the formation of PG in the presence of a smaller uroanal septum, which is not overlaid by inner genital folds.[10,23] Mullassery et al showed that the groove contains both squamous epithelium and a rectal-type mucosa, resembling the anorectal transition zone suggesting that the source is an embryonal remnant such as the urorectal septum.[1] Harsono and Pourcyrous reviewed the histologic results of the resected area, which varied from nonkeratinized squamous epithelium to columnar or cuboidal epithelium.[11] They proposed that the resemblance to anorectal transitional zone epithelium supports an association of PG with an embryonal defect during urorectal septal development. Others proposed that PG forms as a consequence of failure of the midline fusion of the medial genital folds, or as a relic of the open cloacal duct.[18,24] Failure of midline development may explain the shortened distance between the anus and phallus and hypospadias in males and between the anus and urogenital sinus in females.[6,9]

This study has several limitations. We were unable to perform a retrospective database analysis or extrapolate the estimated prevalence because PG is not listed as an anomaly in the ARM-Net database, and many participating centers either do not have specific records of PG or they have records

from recent years only. In addition, the high proportion of patients treated at a single center probably impacted the evaluation of the management strategies; as compared with the other centers, this center applies a more liberal approach toward surgery in patients with isolated PG, especially when there are local complications. In addition, a high percentage of the patients in whom the groove failed to heal had a very short follow-up, such that the ultimate rate of spontaneous epithelization may be higher than reported. Further follow-up is needed to determine this value. ■

Submitted for publication Nov 14, 2019; last revision received Mar 11, 2020; accepted Mar 13, 2020.

Reprint requests: Inbal Samuk, MD, Department of Pediatric Surgery, Schneider Children's Medical Center of Israel, 14 Kaplan Street, Petach Tikva 4920235, Israel. E-mail: Inbal.samuk@gmail.com

References

1. Mullassery D, Turnock R, Kokai G. Perineal groove. J Pediatr Surg 2006;41:e41-3.
2. Verma SB, Wollina U. Perineal groove–a case report. Pediatr Dermatol 2010;27:626-7.
3. Cheng H, Wang Z, Zhao Q, Zhu H, Xu T. Perineal groove: report of two cases and review of the literature. Front Pediatr 2018;6:227.
4. Garcia-Palacios M, Mendez-Gallart R, Cortizo-Vazquez J, Rodriguez-Barca P, Estevez-Martinez E, et al. Perineal groove in female infants: a case series and literature review. Pediatr Dermatol 2017;34:677-80.
5. Kadowaki H, Nakahira M, Yamada C, Takeuchi S, Tamate S, Shiokawa C. Perineal groove and perineal canal Jpn. J Surg 1983;13:216-8.
6. Chatterjee SK, Chatterjee US, Chatterjee U. Perineal groove with penoscrotal hypospadias. Pediatr Surg Int 2003;19:554-6.
7. Pastene SC, Rojas MF. Perineal groove in pediatric gynecology: a report of 2 cases. Rev Chil Pediatr 2014;85:486-90.
8. Hunt L, Srinivas G. Newborn with a perineal lesion. Pediatr Rev 2016;37:e1-3.
9. Siruguppa K, Tuli SS, Kelly MN, Tuli SY. Newborn female with a midline perineal defect. Clin Pediatr (Phila) 2012;51:188-90.
10. Shigemoto H, Endo S, Kimoto M, Horiya Y, Isomoto T, Yamamoto Y, et al. Perineal groove. Case report and embryology. Cong Anom 1978;18:85-7.
11. Harsono M, Pourcyrous M. Perineal groove: a rare congenital midline defect of perineum. AJP Rep 2016;6:e30-2.
12. Boutsikou T, Mougiou V, Sokou R, Kollia M, Kafalidis G, Iliodromiti Z, et al. Four cases of perineal groove- experience of a Greek maternity hospital. Medicina 2019;55:488.
13. Diaz L, Levy ML, Kalajian A, Metry D. Perineal groove: a report of 2 cases. JAMA Dermatol 2014;150:101-2.
14. Senanayake SM, Tennakoon UA. Perineal groove leading to a suspicion of child sexual abuse. Ceylon Med J 2014;59:147-8.
15. Ihn K, Na Y, Ho IG, Oh JT. Clinical characteristics and conservative treatment of perineal groove. J Pediatr Surg 2019 [Epub ahead of print].
16. Stephens FD. The female anus, perineum and vestibule. Embryogenesis and deformities. Aust N Z J Obstet Gynaecol 1968;8:55-73.
17. Gellis S, Feingold M. Picture of the month. Am J Dis Child 1977;131:921-2.
18. Abdel Aleem A, el Sheikh S, Mokhtar A, Ghafouri H, Saleem M. The perineal groove and canal in males and females–a third look. Z Kinderchir 1985;40:303-7.
19. Aslan A, Karagüzel G, Mihci E, Melikoğlu M. Currarino syndrome associated with penoscrotal inversion and perineal fissure. Pediatr Surg Int 2005;21:733-5.
20. Sekaran P, Shawis R. Perineal groove: a rare congenital abnormality of failure of fusion of the perineal raphe and discussion of its embryological origin. Clin Anat 2009;22:823-5.
21. Esposito C, Giurin I, Savanelli A, Alicchio F, Settimi A. Current trends in the management of pediatric patients with perineal groove. J Pediatr Adolesc Gynecol 2011;24:263-5.
22. Wojciechowski M, Van Mechelen K, Van Laere D. Arch Dis Child 2019;104:286.
23. Jin ZW, Jin Y, Li XW, Murakami G, Rodríguez-Vázquez JF, Wilting J. Perineal raphe with special reference to its extension to the anus: a histologic study using human fetuses. Anat Cell Biol 2016;49:116-24.
24. Van der Putte SC. Normal and abnormal development of the anorectum. J Pediatr Surg 1986;21:434-40.