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Short Commentary

Perineal Groove: More than a Rare Anomaly Review of the Literature

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ABSTRACT

The perineal groove is a rare congenital malformation that mainly affects females. Stephens and Smith first described it in 1968 as a moist groove between the vulvar fork and the anus without alteration of the vestibule, urethral or vaginal introitus, and hypertrophy of the labia minora bordering the groove posteriorly to join or surround the anus. Perineal groove is usually asymptomatic and with spontaneous resolution. Due to its lack of knowledge and scarce documentation in the literature, its diagnosis is often erroneous and patients and their families are subjected to stressful events, unnecessary studies and treatments. A literature review is conducted to increase awareness of the existence, treatment, and prognosis of this medical condition.

INTRODUCTION

The perineal groove is a rare congenital malformation that mainly affects females. Stephens and Smith first described it in 1968 as a moist groove between the vulvar fork and the anus without alteration of the vestibule, urethral or vaginal introitus, and hypertrophy of the labia minora bordering the groove posteriorly to join or surround the anus [1].

The perineal groove is classified as complete if it covers the entire perineal surface from the vulvar region to the anal sphincter. If the groove starts from the vulvar area but does not extend to the anus (superior) or from the anus and does not reach the vulva (inferior), it is considered partial. This classification is based on previous research [2]. Several etiopathogenic hypotheses suggest that the sulcus may result from defects in the uroanal septum, failure to fuse the medial genital folds, or the presence of an open remnant of the cloacal duct. Additionally, abnormal expression of several genes (SHH, Gli2, Gli3, Hoxa-13, Fgf10, and BMP4) could play a role [3-5]. In most cases, the sulcus is asymptomatic and its diagnosis is clinical. However, because of its infrequency, it is often misdiagnosed and treated unnecessarily, causing great stress to the family, especially in the face of the anxiety of possible surgical

intervention. A literature review is conducted to increase awareness of the existence, treatment, and prognosis of this medical condition.

SEARCH STRATEGY

An UpToDate, DynaMed, Europe PMC, Biblioteca Virtual en Salud (BVS), Google Scholar and PubMed search was carried out with the Medical Subject Headings terms "perineal groove" and "congenital perineal groove". All relevant and non-duplicated articles published in English and Spanish until July 2022 were included in this review.

RESULTS AND DISCUSSION

The perineal groove is a rare congenital anomaly occurring predominantly in girls, with a female:male ratio of 30:1 according to the 62 cases documented in the literature [2-21]. (Table 1) shows the sociodemographic and clinical data of the patients included in these studies. The cause of the perineal groove remains uncertain, and although no definitive pathological association has been described, cases have been reported in relation to gestational diabetes mellitus (GDM; 4/62) and Carbohydrate Intolerance (CHI), thalassemia, preeclampsia, placenta previa, group B streptococcus infection, Patent Ductus Arteriosus (PDA), among others. Statistical evidence for these associations is limited due to the small number of cases [5,9,10,19].

The perineal groove is typically an asymptomatic anomaly. However, nearly 20% of reported cases have exhibited symptoms, with the most common being constipation (16.5%) and pain (25%), particularly during defecation. It is important to note that this malformation may cause discomfort in some patients, despite its mostly benign nature. Its presentation is generally isolated, although there have been reported cases associated with hypospadias, bifid scrotum [7], ectopic or imperforate anus (3%) [3,8], DAP (5%), laryngeal atresia [12] and anomalies of the urinary tract, especially when it occurs in males [4,15]. Similarly, one of the most recent published cases was associated with an anterior anus [2]. Notably, no association between this condition and low birth weight or preterm delivery has been identified.

A thorough clinical diagnosis is required, including meticulous examination of the perineal area, for accurate characterization of the lesion and to rule out other conditions such as dermatitis, anal fissure, ectopic anus, perineal trauma, lichen sclerosus, anorectal and genitourinary malformations, since up to 5% of cases are initially considered to be sexual abuse [11,17,18] or are subjected to invasive procedures under suspicion of other congenital malformations with worse prognosis [4,14]. Only twelve individuals (19%) received a diagnosis at birth, with an average age of diagnosis being 14.6 months and most receiving diagnoses from specialists other than the one who initially detected the alteration [4-6,13-18]. This may indicate insufficient awareness regarding its existence.

The prognosis of the perineal groove is excellent and spontaneous epithelialization occurs in more than 70% of patients around one year of age [2-21], although cases of natural closure have been documented up to 4 years of age [3,8]. Conservative treatment is recommended and involves local hygiene, stool softeners to prevent constipation, sitz baths, and symptom management. Surgical correction entails excising the groove and performing primary closure. This procedure is indicated if epithelialization fails to occur by two years of age, for esthetic reasons, or if symptoms are unresponsive to optimal medical treatment [3-5]. Esposito et al. suggest using liquid adhesives on the skin suture line to decrease the likelihood of fecal contamination and secondary dehiscence[15]. In the review we conducted, 80% of patients received conservative treatment and 17% underwent surgery. The spontaneous healing rate was only 38%, which we attribute to biases in data collection and follow-up. As such, it is advisable to wait for the natural healing process of the groove, which occurs within 1-2 years of life.

The clinical data reviewed suggest a possible causative relationship with alterations in carbohydrate metabolism (GDM, CHI), but further studies are needed to establish an association [5,10,14,19]. The purpose of this publication is to raise awareness of this malformation's existence to prevent misdiagnosis, avoid superfluous tests and interventions, and enrich the scientific literature.

CONFLICT OF INTEREST

None to declare

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Table 1: Cases of perineal groove reported in the literature and their main clinical features.

Author (year)	Cases (f/m)	Age at diagnosis	Symptoms	Mother's disease	Newborn disease	Treatment	Outcomes
Moreno et al [2]	1 (f)	Birth	No	No	No	Conservative	Healing
Ihn et al [3]	26 (1 m)	1.5 M (mean)	Pain (1)	No	Imperforate anus (1)	Conservative	Healing (10)
Boutsikou et al [4]	4 (f)	Birth	No	IgM (+) CMV (1)	No	Conservative (3), surgery (1*)	Healing (1*), ND (3)
Cheng et al [5]	2 (f)	Birth	No	GDM	PDA (2), DRS (2), ICH (1)	Conservative	ND
Wokciechowski et al [6]	1 (f)	1 M	No	ND	No	ND	ND
Garcia et al [8]	5 (f)	14 M (mean)	Constipation (1)	No	Imperforate anus, hemangioma, myelomeningocele and vesicorenal anomalies (1)	Conservative	Partial healing (2)
Hunt et al [9]	1 (f)	Birth	No	GBHS infection	No	Conservative	Healing
Harsono et al [10]	2 (f)	Birth	No	GDM and preeclampsia (1)	PDA y IAC (1), umbilical cord with 4 vessels (1)	Conservative	Healing
Senanayake et al [11]	1 (f)	26 M	No	ND	No	Conservative	Suspected sexual abuse
Pastene et al [13]	2 (f)	18 D (1*), 8 Y (1**)	Pain (1**)	No	No	Conservative	Healing (1*), ND (1**)
Diaz et al [14]	2 (f)	4 M (1*), 6 M (1**)	No	GDM (1**)	No	Conservative	Partial healing
Carrera [17]	1 (f)	30 M	Pain, constipation	ND	No	Conservative	Suspected sexual abuse
Esposito et al [15]	6 (f)	3 M (mean)	Recurrent infection	ND	No	Surgery (6)	Dehiscence (2), Healing (4)
González et al [18]	1 (f)	2 M	No	ND	No	Conservative	Suspected sexual abuse
Verma et al [20]	1 (f)	30 M	No	ND	No	Conservative	ND
Sekaran et al [19]	1 (f)	Birth	No	GDM, consanguinity	No	Conservative	Healing
Mullassery et al [16]	1 (f)	6 M	Aesthetic	No	No	Surgery	ND
Kanamori er al [12]	1 (f)	Birth	No	No	*CHAOS*, 5p deletion	Conservative	ND
Chatterjee et al [7]	1 (m)	7 A	Mild fecal incontinence	ND	Penoscrotal hypospadias, bifid scrotum	Surgery	Healing
Abdel et al [21]	2 (f)	ND	ND	ND	Ectopic anus (1)	Surgery (2)	ND
Total	62 (2 m - 3.2%)	14.6 M (mean)	12 (19.3%)	7 (11.3%)	9 (14.5%)	Conservative: 50 (80.6%). Surgery: 11 (17.7%)	Healing: 24 (38.7%)

(): Number of cases, M: Male, F: Female, ND: No Data, M: Months, D: Days, Y: Years, */**: Corresponds to the patient, CMV: Cytomegalovirus, GDM: Gestational Diabetes Mellitus, GBHS: Group B beta-Hemolytic Streptococcus infection, CHI: Carbohydrate Intolerance, PDA: Patent Ductus Arteriosus, RDS: Respiratory Distress Syndrome, ICH: Intracerebral Hemorrhage, IAC: Inter-Atrial Communication.

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