Splenogonadal fusion associated with Moebius and Poland syndromes: first case reported

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ABSTRACT

Introduction. Splenogonadal fusion is a rare congenital anomaly of unknown etiology caused by an abnormal fusion of the splenic tissue and the gonadal tissue.

Clinical case. 2-year-old patient with paralysis of the 6th, 7th, and 9th cranial nerves, tent-shaped mouth, cleft palate, right pectoralis major hypoplasia, disruptive defect of the right upper limb, and a mass located at the left inguinal region. At inguinal hernia repair surgery, a processus vaginalis with non-reducible content was observed. When opening the hernia sac, a descending segment of splenic tissue merging with the upper pole of the left testis was found. The patient was diagnosed with splenogonadal fusion. The splenic tissue merging with the testis upper pole was resected, and the remaining splenic tissue was reduced towards the abdominal cavity.

Discussion. Splenogonadal fusion is difficult to diagnose. Being familiar with it allows unnecessary orchiectomies to be prevented.

KEY WORDS: Splenogonadal fusion; Inguinal hernia; Orchiectomy; Moebius syndrome; Poland syndrome; Undescended testis.

FUSIÓN ESPLENOGONADAL ASOCIADA A SÍNDROME DE MOEBIUS Y POLAND: PRIMER CASO REPORTADO

RESUMEN

Introducción. La fusión esplenogonadal es una anomalía congénita rara, de etiología desconocida, causada por la fusión anormal entre el tejido esplénico y gonadal.

Caso clínico. Paciente de dos años, con parálisis del 6°, 7° y 9° nervio craneal, boca en tienda, paladar hendido, hipoplasia del músculo pectoral mayor derecho y defecto disruptivo de la extremidad superior derecha, masa visible en región inguinal izquierda. Al realizar la herniorrafia inguinal encontramos un conducto peritoneo vaginal con contenido no reductible y a la apertura del saco herniario observamos un segmento de tejido esplénico que desciende y se fusiona con el polo superior del testículo izquierdo. El paciente fue diagnosticado con fusión esplenogonadal. El tejido esplénico fusionado al polo superior del testículo es resecado y el resto del tejido esplénico se reduce hacia la cavidad abdominal.

Comentarios. La fusión esplenogonadal es una patología de difícil diagnóstico y su conocimiento puede evitar orquiectomías innecesarias.

PALABRAS CLAVE: Fusión esplenogonadal; Hernia inguinal; Orquiectomía; Síndrome de Moebius; Síndrome de Poland; Testículo no descendido.

INTRODUCTION

Splenogonadal fusion is a rare congenital anomaly of unknown etiology[(2)] caused by an abnormal fusion of the splenic tissue and the gonadal tissue[(1)]. It was first described by German pathologist Bostroem in 1883[(1,2)].

Splenogonadal fusion is usually asymptomatic, and it is incidentally found when exploring the inguinal canal, either for inguinal hernia or undescended testis repair purposes. However, at surgery, it can be mistaken for a testicular tumor. Therefore, if the surgeon is not familiar with it, an aggressive surgical approach may be adopted. This means unnecessary orchiectomies might be performed, potentially with endocrine and exocrine involvement, as well as psychological disorders for the patient[(1-3)].

Pediatric patients can have a scrotal mass and rarely acute scrotal pain as a result of torsion or splenic tissue involvement due to other pathologies such as parotitis, malaria, leukemia, infectious mononucleosis, and trauma[(4)].

CLINICAL CASE

2-year- and 9-month-old male patient with personal pathological history of low weight according to gestational age. He remained in hospital for 1 month following birth. He had familial –maternal– pathological history of *Helicobacter pylori* medical treatment in the first gestation weeks.

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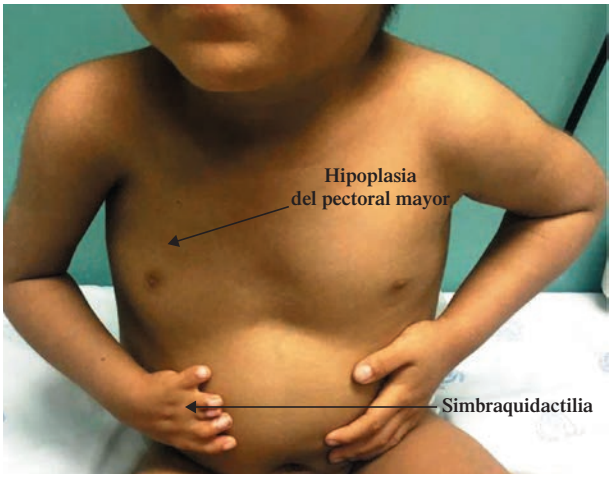

Figure 1. Right pectoralis major hypoplasia associated with a disruptive defect of the right upper limb.

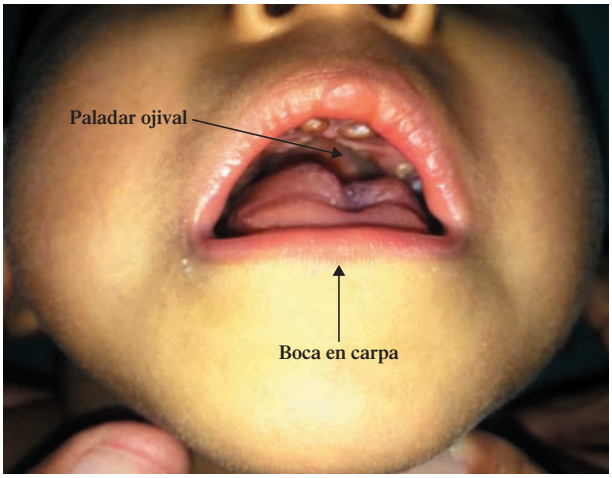

Figure 2. Tent-shaped mouth and cleft palate.

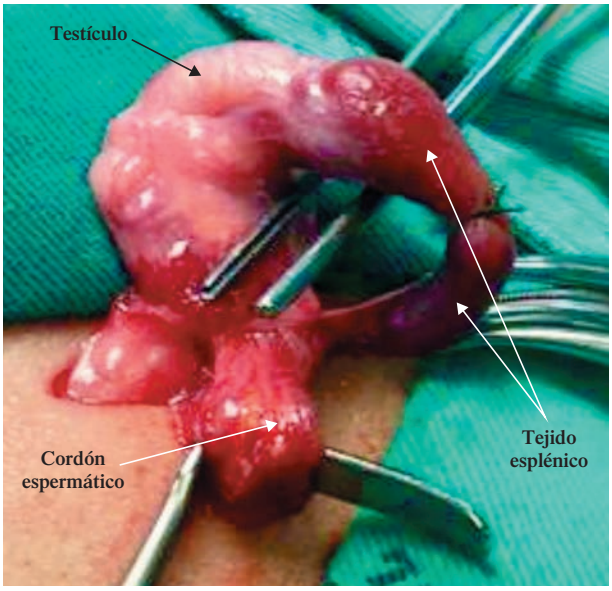

Figure 3. Testis, splenic tissue, spermatic cord, and splenic fibrous band.

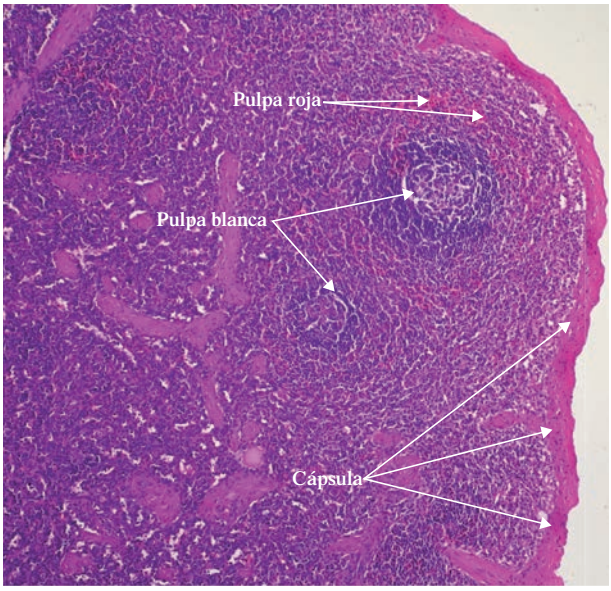

Figure 4. Histologic cut showing the white pulp, the red pulp, and the capsule.

Physical exploration demonstrated bilateral paralysis of the sixth cranial nerve, bilateral facial paralysis, glossopharyngeal nerve paralysis, tent-shaped mouth, cleft palate, right pectoralis major hypoplasia, disruptive defect of the right upper limb, and a visible mass in the left inguinal region increasing and decreasing in size according to physical activity and rest. Clinical diagnosis was left inguinal hernia and Moebius syndrome associated with Poland syndrome (Figs. 1, 2).

When repairing the left inguinal hernia, a processus vaginalis with non-reducible content was found. The hernia sac was opened, and a cord-like segment of splenic tissue descending through the sac and merging with the upper pole of the left testis was observed (Fig. 3). The segment was resected, and the remaining splenic tissue was reduced towards the abdominal cavity. The patient was discharged on the same day with diagnostic suspicion of splenogonadal fusion. Subsequently, the histopathological report confirmed the presence of splenic tissue (Fig. 4) made up of strongly-stained lymphocyte cell aggregates surrounding small central arteries (white pulp), and on the outside, large areas of lighter cells (red pulp). Along with surgery findings, this allowed splenogonadal fusion diagnosis to be confirmed.

DISCUSSION

Splenogonadal fusion is a rare entity, with less than 200 cases published up until now[5-7]. In 1956, Putschar and Manion[6] classified splenogonadal fusion into 2 types –continuous and discontinuous. Continuous-type splenogonadal fusion occurs when the spleen in a normal position merges with the gonad by means of a discrete cord that may be: a) Entirely made up of splenic tissue; b) Made up of multiple interconnected splenic tissue nodules in the shape of a rosary; c) Made up of fibrous tissue[4,6]. In the discontinuous type, a splenic segment is fully detached, which means the splenic tissue merges with the gonad but is completely separated from the spleen – this type can be regarded as a rare variant of accessory spleen[4,6]. Ours was a continuous type case, and the cord was fully made up of splenic tissue.

Splenogonadal fusion, and especially so the continuous type, may be associated with other abnormalities or found when assessing them. These abnormalities include peromelia (limb deficiency syndrome-splenogonadal fusion), micrognathism, congenital heart disease, aortic coarctation, microgastria, cleft palate, craniosynostosis, osteogenesis imperfecta, spina bifida, diaphragmatic hernia, intestinal malrotation, anorectal abnormalities, inguinal hernia, hypospadias, cryptorchidism, crossed testicular ectopia, persistent Müllerian duct syndrome, Roberts syndrome, Moebius syndrome, and Potter syndrome[1-12]. Continuous-type splenogonadal fusion can also be associated with intestinal obstruction[8,9]. Our case was associated with other malformations and syndromes such as Moebius and Poland syndromes.

Association of splenogonadal fusion with testicular malignancy is weak. In the literature, there are approximately seven cases of splenogonadal fusion and neoplastic changes, but in all of them, malignancy occurred in adults with undescended testes[4]. This should be kept in mind, since splenogonadal fusion can be mistaken for a tumor mass at surgery, which means orchiectomy is unnecessary – as it was the case in 37% of the instances reported[5].

According to some authors, continuous-type splenogonadal fusion represents 55-58% of the total instances. As it was the case with our patient, it mostly occurs on the left side – 98% of times according to the series published[1-6,10]. Male-female ratio is 16:1. However, this may not be entirely true, since the ovary is difficult to reach, and most of these cases are asymptomatic, which means splenogonadal fusion incidence in female patients may be underestimated[1,4,6-8]. Indeed, splenogonadal fusion in women has usually been described as an incidental finding at autopsy or laparotomy, whereas in men, it is associated with the search for inguinal hernia, undescended testis, supernumerary testis, or scrotal mass, as in our case[5,11].

Splenogonadal fusion is believed to occur between gestation weeks 5-8, when the splenic blastema is located in close proximity to the left urogenital fold, which makes them partially merge[4,10,11]. At intrauterine week 8, the testis starts to descend from its initial embryological position between the dorsal mesogastrium and the mesonephros, which results in the spleen partially descending[4]. The limbs and Meckel's cartilage – which will give rise to the mandibular arch – also develop actively at this time. This explains why development abnormalities, both limb and facial anomalies, are so frequent[10,11].

Three hypotheses have been proposed to explain the development of splenogonadal fusion[11]:

- Sneath suggested that the inflammation occurring between the gonadal ridge and the spleen results in both of them adhering to each other. However, this does not explain right side cases or the presence of intra-ovarian or intra-testicular splenic tissue.
- Von Hochstetter proposed a retroperitoneal approach allowing splenic tissue and gonadal tissue to communicate.
- Putschar and Manion believed splenogonadal fusion could be caused by the fact the splenic tissue is surrounded by the tunica albuginea of the gonad.

As previously stated, splenogonadal fusion can be associated with other malformations, and also with other syndromes. Buccoliero et al. (in 2011)[12] and Lammens et al. (in 1998)[13] reported 1 case of splenogonadal fusion associated with Moebius syndrome each, similarly to ours. However, our case report is all the more interesting as our patient also had Poland syndrome, which has not been associated with splenogonadal fusion in the literature up until now.

CONCLUSION

Splenogonadal fusion is a rare congenital malformation which can be diagnosed at inguinal region surgery. Clinically speaking, it is difficult to distinguish from other inguinal canal disorders. Therefore, being familiar with it helps prevent unnecessary orchiectomies.

REFERENCES

1. Slongo J, Cardona-Grau D, Swana HS, Rich MA. Splenogonadal fusion: An important differential diagnosis for testicular masses. Pediatr Urol Case Rep. 2019; 6(3): 55-8.
2. Granados Navas FJ, Castro M, Padrón Arredondo G. Splenogonadal Discontinuous Fusion as a Transoperative Finding in Inguinal Hernia Surgery in children (Granado's Hernia). Clinical Case. Mathews J Case Rep. 2019; 4(1): 44.
3. Essaoudi MA, Lamalmi N, Rouas L. An Uncommon Cause of Testicular Lump: Discontinuous Type Spleno-Gonadal Fusion. SM J Pediatr Surg. 2018; 4(3): 1070.
4. Jamal YS, Raml EI, Kurdi MO, Aljahdali EA, Jamal AY. Discontinuous type splenogonadal fusion in abdominoscrotal hydrocele: First reported case. JKAU Med Sci. 2018; 25(1): 45-9.
5. Srinivasa Rao RC, Radhakrishna V, Rao N, Rakshit S. Torsion of a Splenule in a Case of Splenogonadal Fusion Mimicking a Strangulated Inguinal Hernia. J Indian Assoc Pediatr Surg. 2018; 23(2): 100-2.

6. Shakeri A, Rasolmali R, Shakeri S. A case of splenogonadal fusion accompanied by accessory spleen in a 4-year-old boy. Urol Ann. 2018; 10(4): 406-8.
7. Abokrecha A, Almatrfi A. discontinued splenogonadal fusión and bilateral empty scrotum in an 18-month-old boy. Eur J Pediatr Surg Rep. 2017; 5(1): e1-e3.
8. Al-Salem AH. The Spleen and Splenic Disorders. En: Al-Salem AH, Editor. Atlas of Pediatric Surgery. Switzerland: Springer; 2020. p. 246-8.
9. Skandalakis LJ, Gray SW, Ricketts R, Skandalakis JE. The Spleen. En: Skandalakis JE, Gray SW, eds. Embriology for Surgeons. Second Edition. Baltimore: Williams & Wilkins; 1994. p. 338-43.
10. Abdel-Latif M, Sameh Shalaby M. Spleen. En: Carachi R, Doss SHE, eds. Clinical Embriology. Switzerland: Springer; 2019. p. 374-5.
11. Malik RD, Liu DB. Splenogonadal Fusion: An Unusual Case of an Acute Scrotum. Rev Urol. 2013; 15(4): 197-201.
12. Buccoliero AM, Messineo A, Castiglione F, Rossi Degl'Innocenti D, Santi R, Martin A, Taddei GL. Splenogonadal fusion: exceptional association with Moebius syndrome and intestinal intussusception. Fetal Pediatr Pathol. 2011; 30(4): 220-4.
13. Lammens M, Moerman Ph, Fryns JP, Schröder JM, Spinnewyn D, Casaer P, Dom R. Neuropathological findings in Moebius syndrome. Clin Genet. 1998; 54(2): 136-41.