Ped Urol Case Rep 2017; 4(1):262-266 DOI: 10.14534/PUCR.2017124189

Ped Urol Case Rep

PEDIATRIC UROLOGY CASE REPORTS

ISSN 2148-2969

Journal homepage: http://www.pediatricurologycasereports.com

Splenogonadal fusion associated with delayed skeletal maturation: A case report and review of the literature

José Urbano de Medeiros Neto[1], Carlos Gustavo Hirth[1], Conceição Aparecida Dornelas[1], João Batista Gadelha de Cerqueira[2], Dennis Tomio Fujiike[2], Emília Tomé de Sousa[1]

[1]Faculty of Medicine, Department of Pathology, Federal do Ceara University, Fortaleza, Brasil
[2]Faculty of Medicine, Department of Urology, Federal do Ceara University, Fortaleza, Brasil

ABSTRACT

In this report of a 13-year old boy, we describe the first case of splenogonadal fusion (SGF) associated with growth hormone deficiency, delayed skeletal maturation and stunting, and provide a review of the literature on SGF based on a search in major medical indexes using the descriptors "splenogonadal", "splenogonadal fusion" and "congenital anomalies of the spleen". Predominant in males (15:1), SGF is a rare congenital anomaly with only around 200 cases described involving the presence of splenic tissue in the gonads and, in some cases, a fibrous cord connecting the two structures. SGF may be associated with severe limb anomalies, micrognathism and testicle cancer. Despite frequently increased testicle volume, the condition is usually asymptomatic. Knowledge of SGF is important in the differential diagnosis of malignancies, avoiding unnecessary orchiectomy. Nevertheless, SGF has been observed in association with malignant tumors, especially in patients with cryptorchidism.

Key Words: Spleen abnormalities; testicles abnormalities; splenogonadal fusion.

Corresponding Author: *Jose Urbano de Medeiros Neto, Faculty of Medicine, Department of Pathology, Federal do Ceara University, Rua Alexandre Baraúna, 949, Rodolfo Teófilo, CEP 60430-160, Fortaleza, Brasil*
Email: urbano_medeiros@hotmail.com
Accepted for publication: 28 November 2016

Introduction

Splenogonadal fusion (SGF) is a rare congenital anomaly resulting from a connection between the spleen and the gonad or mesonephric remnants during gestation. Both genders may be affected, but the condition is more often described in males probably because testicles are much easier to examine than ovaries. SGF was first reported by Boestrom in 1883, and a detailed report by Pommer followed in 1889. Putschar and Manion categorised splenogonadal fusion into continuous and discontinuous types depending on the anatomical continuity between the

principal spleen and the gonad [1]. In this report we present an incidental case of SGF in a 13-year old boy presenting with stunting at an endocrinology outpatient service.

Case report

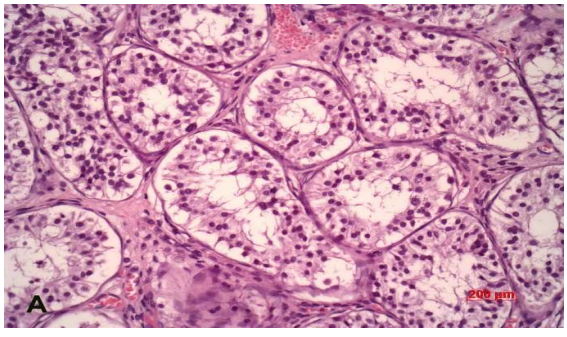
A 13-year old boy from Fortaleza (Northeastern Brazil) residing in Redenção presented with paleness and stunting as main complaints. According to the father, the boy was "pale and small" and had over the preceding months suffered from attention deficit and abdominal pain associated with diarrhea without blood or mucus. The patient had a history of spells of unconsciousness lasting less than an hour and coinciding with fever peaks. The height of the father and mother was 1.65 m and 1.60 m, respectively.

On physical examination, the patient measured 1.31 m (below the third percentile on the 2007 WHO height-for-age chart) and weighed 25.7 kg. The Tanner stage was G2P1. Upon palpation, the left testicle was larger than the right.

A parasitological examination of the feces revealed Strongyloides stercoralis. The blood count included: hemoglobin 13.9 g/dL (normal range [NR] 13.5-17.5), hematocrit 56.5% (NR 41-53), mean corpuscular volume 86.5 fL (NR 80-100), leukocytes 7,300/µL (NR 4,500-11,000), monocytes 7.96 (NR 2.0-10), eosinophils 6.21% (NR 0.0-4.4), basophils 0.89% (NR 0.0-1.0), and platelets 223,000 µ/L (NR 150,000-500,000). Hormonal findings included: insulin-like growth factor 1 (IGF-1) 68.9 ng/mL (NR 183.0-850.0), insulin-like growth factor-binding protein 3 (IGFBP-3) 2,920 ng/mL (NR 1,820-7,060), follicle-stimulating hormone (FSH) 1.1 mUI/mL (NR 0.02-3.00), luteinizing hormone (LH) 0.7 mUI/mL (NR 0.3-9.3), testosterone 2.5 ng/dl (NR <40), thyroid-stimulating hormone (TSH) 2.09 ng/dL (NR 0.34-5.60), and free thyroxine (T4) 0.81 ng/dL (NR 0.54-1.24). The insulin tolerance test revealed no increase in serum levels of IGF-1 and growth hormones (GH), indicating a deficiency in GH production.

Using the Greulich-Pyle method (hand radiography), the patient's age was estimated at 8 years. The skull was normal on nuclear MRI. On scrotal ultrasonography (US), an oval hypoechoic mass with regular outline measuring 1.5 x 0.9 cm was observed adjacent to the left epididymis. Further testing yielded: lactate dehydrogenase (LDH) 495 U/I (NR 230-480), alpha-fetoprotein 1.56 ng/mL (NR <10.9), and human chorionic gonadotropin (hCG) 0.31 mUI/mL (NR <3.0). Duplication of the epididymis was suspected. The patient was referred to the urology service and eventually submitted to surgical resection of the lesion and a segment of the left testicle.

Macroscopically, the surgical specimen appeared as a firm, grayish-brown node measuring 2.0 x 1.8 x 1.5 cm. Upon sectioning, the surface was compact and grayish-brown. Microscopically, testicle sections stained with hematoxylin and eosin (HE) displayed pre-pubertal seminiferous tubules lined with Sertoli cells and spermatogonia, with no production of spermatozoa. This was associated with splenic tissue containing hyperplastic white pulp and hypovascularized red pulp [**Fig. 1A,B,C**]. As a result, the patient was diagnosed with SGF.



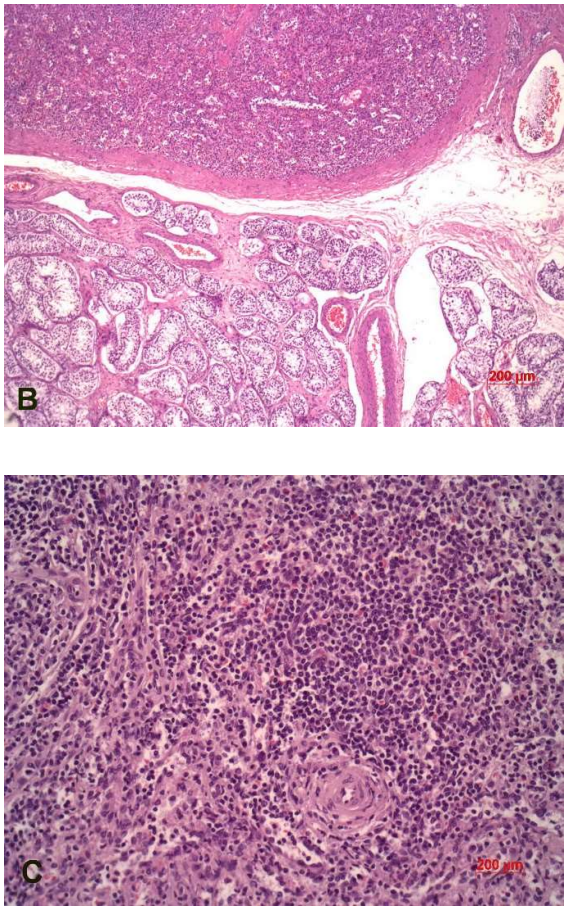


Fig. 1A,B,C. Displayed pre-pubertal seminiferous tubules lined with Sertoli cells and spermatogonia, with no production of spermatozoa. This was associated with splenic tissue containing hyperplastic white pulp and hypovascularized red pulp (H&E).

Discussion

SGF, a rare anomaly with only around 200 cases described, is predominant in males (15:1) [2]. In a case report of a 36-year old woman with urinary complaints, Arancio et al [3] stated that only ten cases of SGF in females are known. This is likely because testicles are much easier to examine than ovaries.

In 1956, Putschar and Manion [1] classified SGF into two types: continuous and discontinuous. In the former, a cord of splenic or fibrous tissue connects the upper pole of the spleen to the upper pole of the gonad or mesonephric remnants. In the latter, no such connection exists [1].

The etiology of SGF remains unknown. The embryonic development of the spleen and gonads begins in the 5th week of gestation. At this time, the spleen is located in the dorsal mesogastrium, but between the 6th and 8th weeks, due to intestinal rotation, it migrates to the left side of the abdominal cavity, approaching the developing gonad [4]. Presumably, SGF occurs at this stage of embryonic development triggered by an unknown agent. The use of phenothiazine derivatives by pregnant women has been proposed as a risk factor for SGF [5].

A number of theories have been proposed to explain SGF. The earliest of these in 1913 suggests that inflammation between the peritoneal surfaces over the gonadal ridge and spleen could produce an adhesion or partial fusion of the two organs [6]. Another theory postulates the existence of a retroperitoneal pathway allowing communication between the splenic tissue and the gonad or mesonephric remnants [7]. On the other hand, Cortes et al [8] believe that splenic tissue can migrate through the diaphragmatic ligament, from the cranial portion of the mesonephros to the testicle. These theories may explain cases of continuous SFG in which a fibrous cord persists between the structures. However, discontinuous SFG is a rare instance of accessory spleen, thus necessarily of different pathophysiology [9].

SGF may be associated with other congenital anomalies. Cryptorchidism results from the interference of the fibrous cord with gonadal descent [8]. Micrognathism and malformation of the upper and lower limbs may also be present. Embryonic development of the limbs and Meckel's cartilage (which forms the mandibular arch) starts between the 5th and 8th

weeks of gestation, i.e., concurrently with that of the spleen and gonads, suggesting that the malformation of all these structures may be triggered by the same agent [1]. Other reported associated malformations include cleft palate, cardiac defects, spina bifida, craniosynostosis and anal malfunction [2]. These changes are usually associated with the continuous form of SGF, but cases of discontinuous SGF associated with cardiac defects (n=3) and micrognathism and limb malformation (n=1) have been reported [10].

Importantly, SGF in cryptorchidic testicles has been associated with malignant tumors, such as carcinoma in situ [11], nonseminomatous germ cell tumors [12], mixed germ cell tumors [13] and anaplastic seminoma [14].

This is the first report of a patient with SFG associated with growth hormone deficiency, delayed skeletal maturation and stunting. This association cannot be explained in light of current knowledge.

SGF may be diagnosed at any age, but most reported cases involve young men under 20 and affect the left testicle. The condition is usually asymptomatic, but testicular pain may occur under the influence of mononucleosis, mumps and leukemia [10]. Diagnosis is most often incidental upon investigation of cryptorchidism, inguinal hernia, scrotal mass [1], testicular torsion [15] and infertility [2], or established upon the histopathological evaluation of orchiectomy.

Imaging is a helpful diagnostic tool, which may prevent unnecessary orchiectomy. By marking the cells of the reticuloendothelial system, technetium-99m sulfur colloid scintigraphy provides the most specific results [16]. Scrotal US associated with abdominal US is unspecific but is a widely accessible tool which allows to visualize the fibrous cord, if any, and the mass adhering to the gonad. US of patients with continuous SGF also allows to visualize movements of the upper pole of the topic spleen when traction is applied to the testis [16].

Our patient was submitted to scrotal US with visualization of an oval structure adjacent to the left epididymis, leading to the hypothesis of duplication of the epididymis. The diagnosis of SGF was established following histopathological examination of the resected lesion and testicle segment.

Although cases of malignancy have been observed (especially in cryptorchidism), the gonadal mass observed in SGF is considered essentially benign. Thus, proper recognition and diagnosis of SGF is important to keep surgery as conservative as possible.

Acknowledgements

The author(s) declare that they have no competing interest and financial support.

References

[1]Putschar WG, Manion WC. Splenic-Gonadal Fusion. Am J Pathol. 1956;32(1):15–33.

[2]Kumar S, Jayant K, Agrawal S, Parmar KM, Singh SK.. A rare case of continuous type splenogonadal fusion in a young male with primary infertility. Case Rep Urol. 2014; 2014:796761.

[3]Arancio M, Marchetti C, Landi G, Mina A, Delsignore A, Marcato M, Martinengo C. [A rare case of female splenogonadal fusion]. Urologia. 2009; 76(2):137-9.

[4]Varga I, Galfiova P, Adamkov M, Danisovic L, Polak S, Kubikova E, Galbavy S. Congenital anomalies of the spleen from an embryological point of view. Med SciMonit. 2009;15(12):RA269-76.

[5]Pauli RM, Greenlaw A. Limb deficiency and splenogonadal fusion. Am J Med

Genet. 1982; 13(1):81-90.

[6]Sneath WA. An Apparent Third Testicle consisting of a Scrotal Spleen. J Anat Physiol. 1913;47(Pt 3):340-2.

[7]von Hochstetter, A. Milzgewebeimlinken Ovarium des linken Individual teiles Einesmenschlichen Thoracopagus. Virchows Arch. f. path. Anat. 1953-54, 324, 36-54.

[8]Cortes D, Thorup JM, Visfeldt J. The pathogenesis of cryptorchidism and splenogonadal fusion: a new hypothesis. Br J Urol. 1996;77(2):285-90.

[9]Le Roux PJ, Heddle RM. Splenogonadal fusion: is the accepted classification system accurate? BJU Int. 2000; 85(1):114-5.

[10]Khairat AB, Ismail AM. Splenogonadal fusion: case presentation and literature review. J Pediatr Surg. 2005;40(8):1357-60.

[11]Lopes RI, de Medeiros MT, Arap MA, Cocuzza M, Srougi M, Hallak J. Splenogonadal fusion and testicular cancer: case report and review of the literature. Einstein (Sao Paulo). 2012;10(1):92-5.

[12]Imperial SL, Sidhu JS. Nonseminomatous germ cell tumor arising in splenogonadal fusion. Arch Pathol Lab Med. 2002;126(10):1222-5.

[13]Thomsen BM, Wierod FS, Rasmussen KC. Combined malignant testicular tumor and splenogonadal fusion. A case story. Scand J Urol Nephrol. 1997;31(4):393-5.

[14]Falkowski WS, Carter MF. Splenogonadal fusion associated with an anaplastic seminoma. J Urol. 1980; 124(4):562–4.

[15]Karaman MI, Gonzales ET Jr. Splenogonadal fusion: report of 2 cases and review of the literature. J Urol. 1996;155(1):309-11.

[16]Patel RV. Splenogonadal fusion J Pediatr Surg. 1995; 30(6):873-4.

Access this article online

http://pediatricurologycasereports.com

Quick Response Code