# Splenogonadal fusion-limb deformity syndrome: a rare but important cause of undescended testis

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***Background:*** **Splenogonadal fusion is a rare congenital anomaly which is characterized by fusion formation between the spleen and gonad.**

***Methods:*** **We report a case of a 14-month boy with spleongonadal fusion-limb deformity syndrome focusing on the importance of awareness of this syndrome.**

***Results:*** **The patient was admitted to our clinic because of a left undescended testis, and preoperative diagnosis was not made. During the operation, "spleen-like" tissue attached to the gonad induced splenogonadal fusion, which was confirmed by laparoscopy. The patient also had a short right femur, hip dysplasia and a syndromic face.**

***Conclusion:*** **Splenogonadal fusion anomaly should be considered in the evaluation of undescended testis, especially in patients with facial and limb deformities.**

***World J Pediatr 2016;12(2):246-248***

***Key words:*** **diagnostic laparoscopy; splenogonadal fusion; undescended testis**

Case report

## Introduction

Splenogonadal fusion-limb deformity syndrome (SGFLD) is a rare form of splenogonadal fusion anomaly with a great deal of interesting embryological defect hypotheses. We report a 14-month-old boy with continuous splenogonadal fusion anomaly who was admitted to our clinic because of a left undescended testis, recognized with splenic tissue attached to the testis during the operation and confirmed by laparoscopy. The boy also had facial and limb deformities which met the criteria of SGFLD.

## Case report

A 14-month-old boy was referred to our clinic from the Department of Pediatric Endocrinology of this hospital for the evaluation of a left undescended testis. Human chorionic gonadotropin treatment was applied and no benefit was achieved. He was also under evaluation for a short right femur and hip dysplasia. He had a syndromic face and a left testis in the inguinal canal close to the external ring. The patient was suspected of a genetic syndrome. Ultrasonography in the newborn period showed nothing abnormal including undescended testis or any abnormality of the spleen. Physical examination revealed an ordinary undescended testis, so he was scheduled for a routine orchiopexy. The long loop vas was identified during the dissection and the testis was liberalized. Further dissection revealed a spleen-like tissue attached to the upper pole of the testis, and laparoscopy was performed. Complete exploration of the abdominal cavity revealed a continuous type of splenogonadal fusion, an open internal ring and no other abnormalities (Fig. 1). After the left inguinal incision, the testis and spermatic bundle were isolated, the splenic tissue was dissected, and orchiopexy was carried out (Fig. 2). During the dissection, a gross arterial connection which restricted the descent was identified but ligated after manifestation of an independent testicular artery. No abnormalities were found with gross inspection and palpation of the testis, and no vascular insufficiency occurred after surgery.

Follow-up for 4 years showed bilateral well grown testes. At the age of two years, the patient lost an important part of his front teeth and was operated upon thrice for hip dysplasia. He is still under evaluation and close follow-up of endocrinology for growth retardation (with low insulin-like growth factor), but has no delay in mental development.

## Discussion

Splenogonadal fusion anomaly was first described by

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doi: 10.1007/s12519-015-0046-y
Online First November 2015

Bostroem in 1883.[1] As a rare congenital anomaly, more than 150 cases in 16 reviews have been reported since then.[1,2] A review of 137 cases in 1996 revealed that most of the cases are not reported. Moreover, the prevalence of the anomaly in females is underestimated because of inaccessibility to the female gonad on physical examination. The male/female ratio is 16:1 in overall and 13:8 in the cases diagnosed by autopsies.[1]

Splenogonadal fusion anomaly was reviewed and described in detail by Putschar and Manion in 1956 and classified into two types: continuous and discontinuous.[3] In the continuous type, there is a cord-like band or fibrous or splenic tissue linking the spleen to the gonad. In the discontinuous type, there is no connection. The higher frequency of associated anomalies in the continuous type built up the idea that it results from an earlier embryological defect. Besides, both Putschar and Manion and later reports concluded that SGFLD is a syndrome which should be distinguished from simple splenogonadal fusion without limb defects.

SGFLD was first described in 1889 by Pommer in an infant with limb deformities, micrognathia and anal atresia.[1] Since then, associated anomalies were well established and 34 cases were reported with a significantly high portion of terminated pregnancies because of severe anomalies.[1,4-6]

The exact pathogenesis of splenogonadal fusion anomaly is unknown; however there have been some reasonable hypotheses. The spleen has an interesting embryological development process. Despite most of the intraabdominal organs, it is not a derivate of the digestive tract. Clusters of mesenchymal cells on the dorsal mesogastrium are rapidly fused and vascularized. The rotation of the stomach and the growth of the dorsal mesogastrium 6-7 weeks cause the translocation of the spleen from the midline to the left side of the abdominal cavity where a significant proximity to the primordial gonad is obtained. Therefore, splenogonadal fusion anomaly is considered to happen during 5-8 weeks of gestation. Three main mechanisms such as simple adhesion, inflammatory processes and teratogens are attributed to a long time; however there are some other suggestions in the later reports.[7] Cortes et al[8] hypothesized that splenogonadal fusion is a result of migration of splenic cells into the left diaphragmatic ligament which inhibits its involvement and therefore the formation of the splenic cord. They found an undescended testis ratio of 31%, more than half of them bilateral; they found that noninvolvement of the ligament is the cause of the disruption in the descent of the testes. Also they argue that associated extremity anomalies indicate an earlier embryological, probably a midline defect.[8]

McPherson et al[4] argued that SGFLD is probably a developmental field defect with a hit occurring during blastogenesis and may be a polytopic field defect. They also stated that the earlier the abnormal event happens, the greater the number of anomalies that are produced as other developmental defects. Loomis et al[9] observed 351 embryos and found that the development of extremities is most intense in Carnegie phases 15-16 and that the developing spleen has a proximity to left mesonephros in phases 17-18. They suppose that SGFLD is therefore an early embryological defect and fusion without late occurence of limb defects.[9] An association with the group of oromandibular-limb-hypogenesis syndromes or Hanhart complex also has been proposed because of the high frequency of orofacial abnormalities.[1]

The probable early embryological defect and multiple abnormalities are related to a genetic defect; however, to our knowledge, none of them was characterized. Our patient fulfilled the criteria of SGFLD with splenogonadal fusion, limb deformities, and the characteristic face. Preoperative diagnosis was not established in our patient, but associated malformations and "spleen-like" tissue attached to the

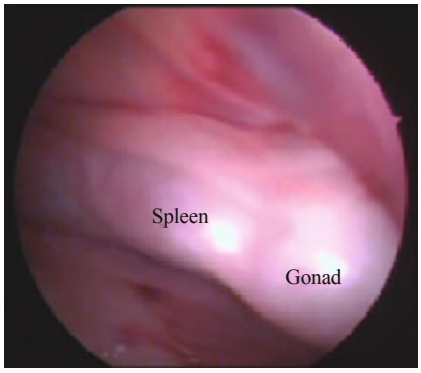


**Fig. 1.** Laparoscopic view of continuous type splenogonadal fusion.

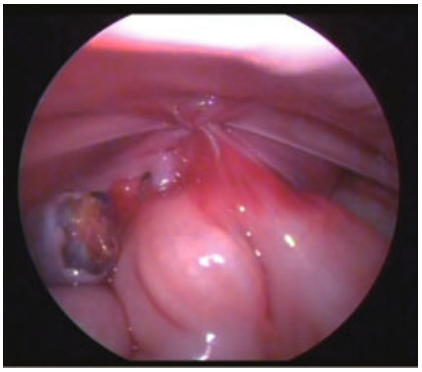

**Fig. 2.** After dissection of the spleen and gonad and high ligation of the tunica vaginalis, laparoscopy was performed again. This is the view of the internal inguinal ring and spleen after dissection.

gonad constituted the idea of splenogonadal fusion. Finally the diagnosis was confirmed by laparoscopy. Laparoscopy confirmed the diagnosis without the need of frozen section, complete evaluation of the site, but uncomplicated surgery. We consider that preoperative sonography is required to confirm the diagnosis of patients with abnormalities such as facial or limb defects.[10]

Malignancy in splenogonadal fusion was seen in 4 cases of undescended testis.[11-14] The four cases had primary malignancies of the testis but normal spleen tissue. Therefore malignancy was considered to be associated with cryptorchidism, not with splenogonadal fusion. No macroscopic abnormality in the testis was identified in our patient, therefore testis sparing surgery was performed.

In conclusion, we believe that splenogonadal fusion anomaly must be kept in mind in evaluation of undescended testis, especially in patients with facial and limb deformities. Laparoscopy may be useful in the diagnosis and management of the disease.

**Funding:** None.

**Ethical approval:** Written informed consent was obtained from the patient for publication of this case report and any accompanying images. A copy of the written consent is available for review.

**Competing interest:** There is no competing interest.

**Contributors:** TS wrote the main body of the article under the supervision of CA, DS and EO. CA and EO provided advice on medical aspects. CA is the guarantor.

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*Received March 26, 2014*
*Accepted after revision July 11, 2014*