Case report

Splenogonadal fusion and sex reversal

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Abstract

Splenogonadal fusion is a rare congenital malformation where an abnormal union occurs between the spleen and gonad or mesonephric derivatives. Although it occurs in females it is much less prevalent than in males (male:female ratio, 16:1), but this may partly be because of the inaccessibility of the female gonads leading to under-diagnosis. To our knowledge this is the first case of splenogonadal fusion associated with sex reversal reported in the literature.

Keywords: associated abnormalities; sex reversal; splenogonadal fusion.

Introduction

Splenogonadal fusion is a rare congenital malformation where an abnormal union occurs between the spleen and gonad or mesonephric derivatives (1). Although it occurs in females it is much less prevalent than in males (male:female ratio, 16:1), but this may partly be because of the inaccessibility of the female gonads leading to under-diagnosis. To our knowledge this is the first case of splenogonadal fusion associated with sex reversal reported in the literature.

Case report

The baby was born by normal vaginal delivery at 39 weeks’ gestation weighing 3.845 kg. Antenatal scans had shown a hypoplastic left heart and the infant received corrective surgery on day 1 of life. Routine chromosome analysis demonstrated a 46,XY genotype despite a female phenotype.

Further investigations revealed no functioning testicular tissue, with a negative β human chorionic gonadotrophin (βhCG) stimulation test and undetectable anti-Müllerian hormone levels, despite the positive detection of the SRY (sex-determining region of the Y-chromosome) by in situ hybridization using a DNA probe. Abdominopelvic ultrasound scan revealed two small areas of probable testicular tissue and no uterus.

Laparoscopy performed prior to gonadectomy demonstrated a chain of four small nodules of tissue on the left, extending laterally from near the bladder up towards the kidney. These were removed and sent for histology. No gonadal tissue was seen on the right, just a very thin cord-like structure in the midline above the bladder that was not retrieved for histological analysis. No female internal structures were present.

Histological analysis of these nodules showed the presence of ectopic spleniculae without gonadal tissue in two specimens. One nodule contained spermatic cord and vessels. The fourth tissue nodule was formed of splenic tissue only. Despite the absence of definitive gonadal tissue, this was thought to represent splenogonadal fusion (2) (Figure 1).

Discussion

Splenogonadal fusion is a rare abnormality that has been classified into two types (1). Firstly, in the continuous form there is a direct anatomical connection between the spleen and gonad, usually as a cord of splenic or fibrous tissue, or rarely as beads of splenic tissue in a fibrous cord. This type is associated with major congenital abnormalities, in particular limb defects and micrognathia (1–3). The second type is discontinuous: ectopic splenic tissue attached to the gonad, but with no direct splenic connection. The accepted classification is contentious, with others arguing that the discontinuous type is actually an accessory spleen and therefore has a different etiology (4). Only one case has been reported where there was intermingling of gonadal and splenic tissue; normally they are separated by a distinct capsule (1, 2, 4).

The underlying cause of splenogonadal fusion is not known, but is thought that some insult during the fifth to eighth week of gestation, where the developing spleen and left urogenital fold containing the gonadal mesoderm are in close proximity, causes fusion of these structures (4). Splenogonadal fusion occurs almost exclusively on the left side (1, 2).
The condition is known to be associated with other congenital anomalies, commonly with lower limb defects, although it has also been reported in association with cardiac defects (but not previously a hypoplastic left heart) (2).

Splenogonadal fusion interferes with the normal descent of the testes and the closure of the processus vaginalis (4). It also manifests as a testicular mass, so diagnosis is commonly made incidentally during testicular or inguinal surgery (2, 4). It can also present acutely causing bowel obstruction in the continuous form or as an acute scrotum as a result of scrotal splenic enlargement (5). The vast majority of cases present during childhood or early adult life (1, 2).

Although splenogonadal fusion is occasionally seen in phenotypical females and has been described in children with abnormal genitalia (3), there are no previous reports in the literature where it has occurred in a case of complete phenotypic sex reversal.

In normal sex development the presence of testis-determining factor causes the differentiation of sex cord cells into Sertoli cells. These secrete anti-Müllerian hormone inducing the degeneration of the paramesonephric ducts. Leydig cells also differentiate from mesenchymal cells within the genital ridges and produce testosterone that causes the previously bi-potential external genitalia to develop into a male phenotype (6).

In this particular case, Sertoli cells must have differentiated earlier in development, resulting in the absence of female internal organs, but a lack of virilization suggests that Leydig cells did not develop properly. Alternatively, it is possible that the gonad underwent regression, the so-called ‘vanishing testes syndrome’.

The spleen is a mesodermal derivative and during early development, is in close proximity to the urogenital ridge. At a similar gestation the outflow tract of the heart undergoes separation into aorta and pulmonary trunk and ventricular septation is completed. An embryonic event occurring at this time (7–8 weeks) may have affected both cardiac and gonadal development and caused the hypoplastic left heart and fusion of the developing testis and spleen (6).

References


Figure 1  (A) Microphotography depicting splenic tissue, with presence of white and red pulp (hematoxylin and eosin ×20). (B) Spermatic cord was identified adjacent to the splenic tissue.