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CASE REPORT

Beta thalassemia major, a cause of testicular microlithiasis

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Abstract

Testicular microlithiasis is a rare condition characterized by deposition of calcium in the Lamina of semineferous tubules of testis. The ultrasound of testicular microlithiasis shows bright 1-3 mm echogenic foci in parenchyma. Herein, we report a first case of testicular microlithiasis in major beta thalassemia that was demonstrated in testicle's ultrasound.

Keywords: Testicular microlithiasis; Beta thalassemia; Malignant precursor Scrotal ultrasound

Introduction

Testicular microlithiasis (TM) is uncommon and incidental finding in scrotal ultrasound.[1] The prevalence of TM is reported to be 0.6% in patients referred for scrotal ultrasound, but has not been demonstrated in normal population.[1,2] It has been shown in various benign and malignant diseases and is usually bilateral.[2-4] There is evidence of an association between TM, intratesticular germ cell neoplasia and other testicular and extratesticular tumors,[5-7] a reason for considerable attention paid to testicular pathology.[8,9] The age of patients ranged from a few months to 80 years,[12-16] with the mean age varying from 22 to 37 years.[1,10,11]

To our knowledge, this is the first report of TM in thalassemia, which underlines the importance of TM and thalassemia in regard to relative risk of concurrent tumor and infertility. TM and calcium deposition may have a synergistic role in testicular dysfunction.

Case Report

A 10-year-old boy with beta thalassemia underwent ultrasonic evaluation of testis for scrotal pain. In scrotal ultrasound (G50-Siemens), tiny non-shadowing echogenic foci appeared in parenchyma that was compatible with pathognomonic configuration of TM. (Fig 1)

Testes were normal in size. Hydrocele, varied and congenital anomalies were not observed in ultrasound. Epididymis was normal in shape, size and echogenicity. There were no pathologic findings in regard to color, power and spectral Doppler. TM was not present in his family history. Erythema, swelling and retraction of scrotum were not found on clinical examination. He had no history of trauma, radiation and scrotal pathology.

Clinical diagnosis was neglected trauma with normal uric acid, calcium and phosphorous. The patient was followed for one month, receiving blood transfusion and was treated with deferuxamin and folate.

Discussion

TM was first reported by Priebe and Garrent in 1970 in a healthy 4-year-old boy.[12] Its Ultrasonic appearance was first described by Doherty in 1987.[13] Testicular microlithiasis Usually occurs bilaterally. Patient is generally asymptomatic. Previous reports have indicated a prevalence of testicular microlithiasis of 0.6% in a population referred for symptomatic scrotal ultrasound.[10] The mean age of patients presenting with testicular microlithiasis was reported 22.3 years.[1,6] However, reported age range for this condition has ranged from a few months to 80 years.[1,2] The

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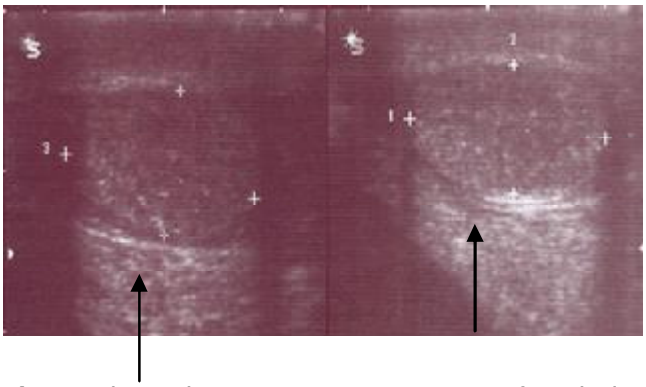

Fig 1: Ultrasonic snowstorm appearance of testicular microlithiasis

mean age of patients with concurrent testicular microlithiasis and tumor was defined 30 years.[1,4]

The exact cause of testicular microlithiasis remains unknown. In pathology of testicular microlithiasis, the microliths that are intratubular bodies with a central calcified core and surrounding concentric lamination composed of collagen fibers. Microcalcification is thought to be resulted from congenital defective phagocytosis of degenerated tubular cells by sertoli cells.[17]

Today, testicular microlithiasis is most commonly diagnosed by high frequency (5-10 MHZ) testicular ultrasound. On sonography, testicular microlithiasis exhibits pathognomonic extensive tiny (1-3 mm), nonshadowing, echogenic foci that randomly widespread throughout the testicular parenchyma.[2,5]

Several association were reported with testicular microlithiasis including pulmonary alveolar microlithiasis, male pseudohermaphrodism, cryptorchidism, Down syndrome, previous radiotherapy, Klinefelter syndrome, subfertility states, congenital urthroperineal fistula, familial tumoral calcinosis, Mccune–Albright syndrome, fragile x syndrome, ductal carcinoma of breast and Leiomyomatosis. Most important association was with testicular neoplasm's.[17,18]

Symptomatic patients with testicular microlithiasis exhibited an incidence of testicular neoplasm greater than of general population. However, only associations, and no cause-and-effect relationship have been established. Testicular microlithiasis may be only as a marker of an abnormal testis affected by a range of abnormal processes. But, there is close association between it and intratubular germ cell neoplasia. In autopsy, the prevalence of neoplasia in the general population is approximately 0.8%, and it has been demonstrated in 4.5% of biopsies of the contralateral testis in patients presenting with testicular tumor.[19,20] Intratubular germ cell neoplasia can progress to carcinoma in 50% of cases.[20] The importance of TM is due to its premalignant tendency. It is recommended to encourage self-examination and to perform regular evaluation of biohumoral (alphafetoprotein, and Betachorionic gonadotropim and ultrasound almost once a year. Biopsy is rarely suggested except for terato- spermia.[24-26]

A number of testicular tumors such as teratoma, seminoma, teratoseminoma and extratesticular tumor such as abdominal and thoracic germ cell tumor, mediastinal choriocarcinoma and ductal carcinoma of breast have been associated with TM.[21-23,] but none in relation to thalassemia. Thalassemia comprises a group of genetic disorders with reduced biosynthesis of one or more of the globin chains of hemoglobin. Iron loading (hemochromatosis) is the cause of complications in thalasemia. The iron has a particular prediction for the endocrine system, liver and heart with resulting lysosomal deposition, disruption and fibrosis. These phenomenons probably lead to tissue damage due to enzyme releasing.

Iron deposition does not have echogenic configuration in ultrasonography. We know the echogenic foci in testes are due to calcium deposition; perhaps iron particle deposition also may play a role in this pattern.

So, doing further studies that can evaluate probable correlation between testicular microlithiasis and thalassemia is recommended.

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