Case Report

Testicular microlithiasis in two boys with a chromosomal abnormality

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ABSTRACT

A nine and 13-year-old boy, previously diagnosed with 18q syndrome and an 11q deletion, respectively were diagnosed with testicular microlithiasis (TM). Both cases demonstrate that TM occurs in patients with various chromosomal abnormalities

Key words: Testicular microlithiasis, 18q syndrome, 11q deletion

INTRODUCTION

We report testicular microlithiasis (TM) in a nine and 13-year-old boy who had been previously diagnosed with 18q syndrome and 11q deletion respectively. To the best of our knowledge, this is the first documentation of patients suffering from these chromosomal abnormalities together with TM.

CASE REPORTS

Case 1

A nine-year-old boy was diagnosed with 18q syndrome at the age of three based on characteristic dysmorphic features, mainly comprising hypertelorism, maxilla hypoplasia, a small nose and aural deformities [Figure 1]. DNA analysis confirmed the diagnosis of 18q syndrome (46,XY,del(18)(q23)). Both parents and his twin sister had normal chromosomal patterns. Family history was negative on testicular cancer, genital abnormalities and subfertiltiy. The boy was also diagnosed with bilateral congenital undescended testis and hypospadia. Orchidopexy was performed at the age of one. He was also known to suffer from asthma and used inhalation steroids (salmeterol/fluticason) and β-sympaticomimeticum (salbutamol).

At the age of nine, the left-sided testis appeared to be undescended again and a diagnosis of secondary acquired undescended testis was made. Ultrasound of both testicles revealed classical TM, Grade 2 (10-20 diffuse scattered foci per transducer) in both testes.

Case 2

A 13-year-old boy was diagnosed with partial monosomy of 11q as well as partial trisomy of Chromosome 4q (46,XY,11,+der(11)t(4;11)(q31.3;q25)) based on specific deformities. Both parents and his 11-year-old brother had normal chromosomal patterns. Family history was negative on testicular cancer, genital abnormalities and subfertility. The patient had been referred to the outpatient Department of Pediatrics of the Medical Centre Alkmaar because of bilateral acquired undescended testes. Ultrasound revealed classical TM Grade 2 (10-20 diffuse scattered foci per transducer) in the right testis [Figure 2] and Grade 1 (5-10 foci per transducer) in the left testis.

DISCUSSION

Patients with a deletion on the long arm of Chromosome 18 display a wide variety of phenotypic traits. The most common manifestations are short stature, midface hypoplasia, hypertelorism, congenital aural atresia, foot deformities, mental retardation and hypotonia.[1]

Monosomy of 11q as well as partial trisomy of Chromosome 4q are a rare chromosomal abnormality and may include the specific deformities of two distinct chromosomal aberration

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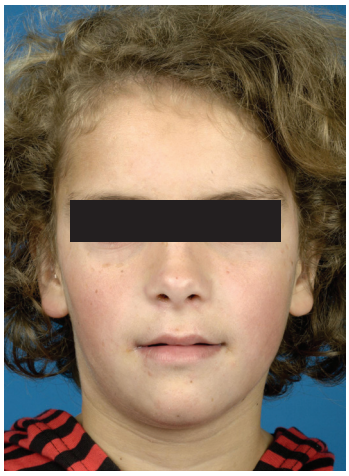

Figure 1: Facial appearance of Case 1 showing hypertelorism, maxilla hypoplasia, prognathia and a small nose (published with written permission of the parents)

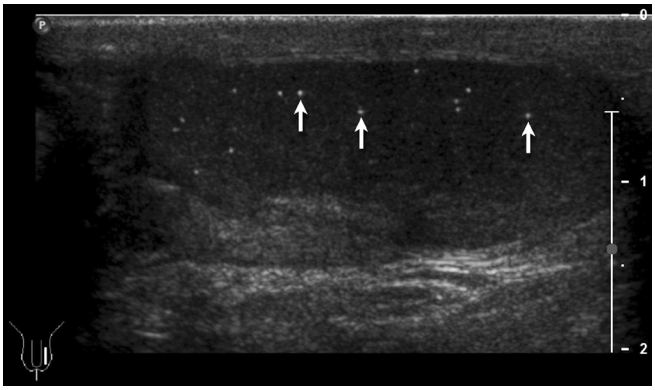

Figure 2: Transverse ultrasound image of the left testis of Case 2 showing 10-20 microliths per sonographic plane (see arrows). The microliths are without acoustic shadowing and are scattered diffusely throughout the testicular parenchyma

syndromes: protruding metopic suture, undescended testis and hypertelorism. Furthermore, mild to moderate mental retardation is present.[2]

Testicular microlithiasis (TM) might be associated with impaired spermatogenesis and increased risk of malignant testicular germ cell tumor (TGCT). It is characterized by multiple echogenic foci (< 3 mm) without acoustic shadowing within the testis parenchyma. The presence of five or more foci is defined as classical TM and less than five microliths as limited TM.[3] Suggested sources of TM are the tubuli Seminiferi, as well as the surrounding tissue layers.[4] The pathogenesis is unclear and the age of onset is largely unknown. Only a small number of cases in infancy have been reported. TM might be one of the components of male reproductive problems, together with TGCT, genital abnormalities, reduced semen quality and subfertility. This phenotype has previously been described as the testicular dysgenesis syndrome.[5]

TM may be associated with cryptorchidism and varicocele.[6] It has also been found to occur in boys with various chromosomal abnormalities including Down syndrome, Fragile X syndrome and Klinefelter syndrome. The patients reported presented with additional chromosomal abnormalities. In boys with chromosomal abnormalities, TM might be a signal of degenerative changes in the testis resulting in impaired fertility which is commonly associated with these conditions. It might be worthwhile screening all boys with a chromosomal abnormality for TM and concomitant testicular abnormalities, especially if additional risk factors for TGCT are present.

REFERENCES

1. Wertelecki W, Gerald PS. Clinical and chromosomal studies of the 18q-syndrome. J Paediatr 1971;78:44-52.
2. Lewanda AF, Morsey S, Reid CS, Jabs EW. Two craniosynostotic patients with 11q deletions, and review of 48 cases. Am J Med Genet 1995;59:193-8.
3. Backus ML, Mack LA, Middleton WD, King BF, Winter TC, True LD. Testicular microlithiasis: Imaging appearances and pathologic correlation. Radiology 1994;192:781-5.
4. Bercovitch RS, Januario JA, Terry SF, Boekelheide K, Podis AD, Dupuy DE, *et al*. Testicular microlithiasis in association with pseudoxanthoma elasticum. Radiology 2005;237:550-4.
5. Skakkebaek NE, Rajpert-De Meyts E, Main KM. Testicular dysgenesis syndrome: An increasingly common development disorder with environmental aspects. Hum Reprod 2001;16:972-8.
6. Domínguez Hinarejos C, Bonillo García MA, Sangüesa C, Serrano Durbá A, García Ibarra F. Intratesticular calcifications in childhood. Actas Urol Esp 2007;31:33-7.

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