Chapter 2.6.2

Testicular microlithiasis in boys with pseudoxanthoma elasticum or chromosomal abnormalities

Testicular microlithiasis in 2 boys with a chromosomal abnormality

J Goede
WWM Hack
K Sijstermans
FH Pierik

*Indian J Urol; submitted after revision*
Introduction

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

Case presentations

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 (see Figure I). DNA-analysis confirmed the diagnosis of 18q- syndrome (46,XY,del(18)(q23)). Both parents and his twin sister had normal chromosomal patterns. 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 9, 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 thirteen-year-old boy was diagnosed with subtelomeric 11q- deletion based on typical facial deformities. Both parents and his eleven-year old brother had normal chromosomal patterns. The patient had been referred to the outpatient Department of Paediatrics 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 (see Figure II) 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

Subtelomeric 11q- deletion is a rare chromosomal abnormality and specific facial
deformities may include: down-slanting palpebral fissures, ptosis and hypertelorism. Furthermore, mild to moderate mental retardation is usually present.²

Testicular microlithiasis (TM) might be associated with impaired spermatogenesis and increased risk of malignant testicular germ cell tumour (TGCT). It is characterised 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 may be associated with cryptorchidism and varicocele.⁵ 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.

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.
References


