Triorchidism: genetic and imaging evaluation in an adult male

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CASE REPORT

A 54 years old man presented to our outpatients Department with complaints of erectile dysfunction. His past medical history revealed diabetes type 1 since the age of 28 years and 10-year history of a left-sided scrotal swelling associated with some discomfort. He was married and fathered two daughters.

Abdominal examination was normal with no palpable mass or groin herniae. Scrotal examination revealed a normal right testis and scrotal content, but on the left side there were two similar size lumps. Laboratory studies, including hormonal and oncological markers, were within normal limits.

A scrotal ultrasonography revealed the presence of 2 testes within the left hemiscrotum with complete septation and echotexture and vascular flow pattern similar to the vascular flow of the normal right testis. There was no focal abnormal echogenicity suggesting malignancy (Figure 1). Scrotal MRI confirmed 2 soft-tissue structures in the left hemiscrotum with normal signal intensity at T1w and T2w images. Both testes had a tunica albuginea with low-signal intensity (Figure 2).

Chromosomal preparations for the karyotype analysis were obtained according to standard techniques. Cytogenetic analysis at a resolution of 400 bands resulted in normal male karyotype 46XY.

Patient DNA was analysed by Array-CGH analysis using a commercially available oligonucleotide microarrays containing about 44.000 60-mer probes (Human Genome CGH Microarray 44B Kit, Agilent Technologies, Santa Clara, California) according to the manufacturer's instructions.

Array-CGH analysis detected the presence of two interstitial rearrangements: a ~120 Kb deletion of chromosome 1(arr1q31.1(79,356,819-79,476,571)x1) and a ~140 Kb deletion of chromosome 16 (arr 16q22.1(70,052,164-70,193,889)x1). Parents were not available for testing. The microdeletion of chromosome 1 includes the ELTD1 (latrophilin and seven transmembrane domain contain-

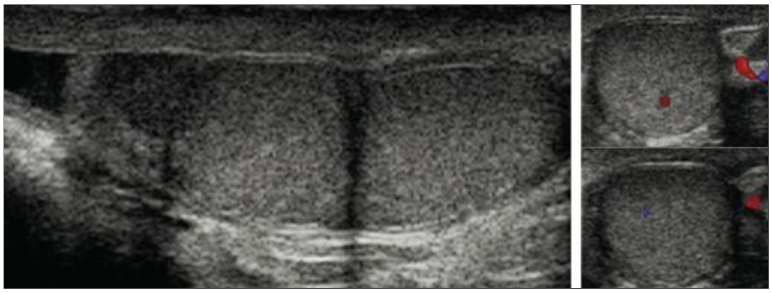
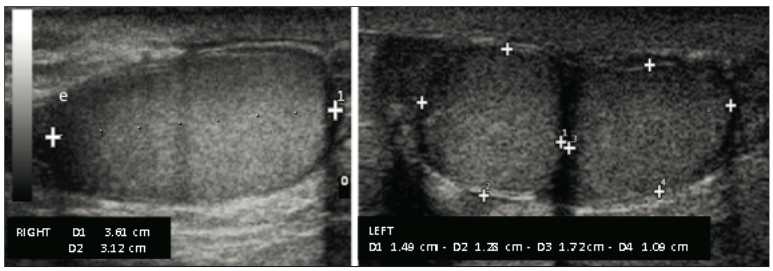


Figure 1.

A. *Sagittal sonographic image of the left scrotum showing two testicles completely separated. The supernumerary testis is smaller and superior to the more normal-sized and it appears as an oval, isoechoic mass with a homogeneously echogenic pattern identical to that of the other testicle; they share the epididymis and the vas deferens.*



B. *Color Doppler image showing the same vascular pattern of the normal and accessory testicle.*

C. *Both testicles have approximately the same size.*

No conflict of interest declared

ing1) gene, while the microdeletion of chromosome 16 includes the PDPR (*Pyruvate Dehydrogenase Phosphatase Regulatory Subunit*) gene. Currently there are little details on the functions of both genes. The protein encoded by ELTD1 could be involved in cardiac development. The protein encoded by PDPR is a regulatory subunit of human mitochondrial pyruvate dehydrogenase phosphatase. It decreases the sensitivity of PDP1 to magnesium ions, and this inhibition is reversed by the polyamine spermine. Both these proteins are expressed also in testis. A diagnosis of triorchidism was made and the patient was placed in sonographic follow up.

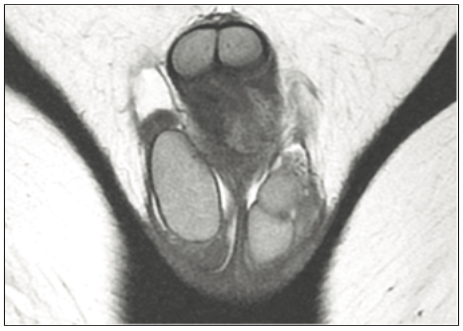
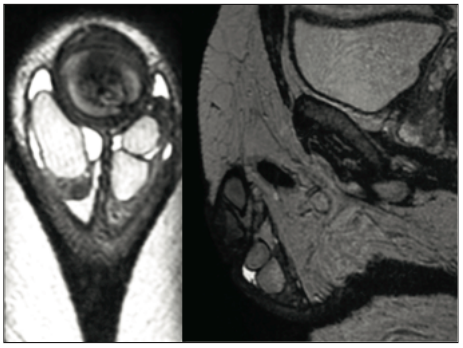


Figure 2.

Coronal T2-weighted.

A. *Image and 3D-FSE-Cube sequence reformatted in multiple imaging planes*



B. *Showing two testes within the left hemiscrotum.*
They are identical in signal intensity to the normal right testis.
All three testes are surrounded by a low-signal-intensity tunica albuginea.