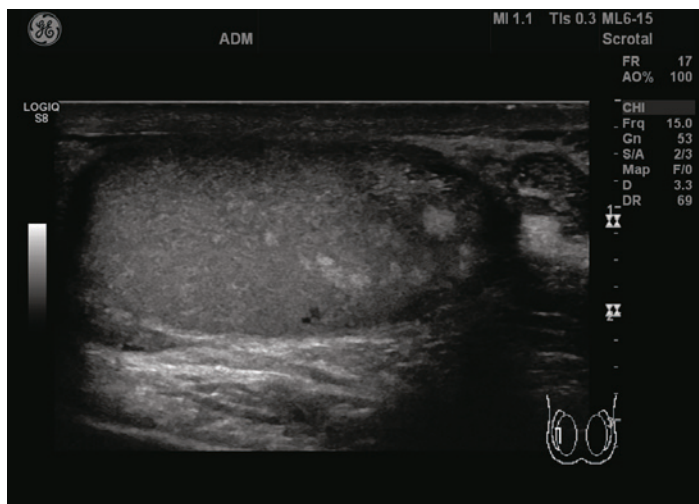


Fatty tissue in testicles



A healthy man in his 40s experienced spontaneous pain in both groins and testicles and was therefore referred by his primary doctor for an ultrasound scan. The scan revealed numerous foci of increased echogenicity spread through his testicles. They were indistinctly delimited, threw no shadow and no doppler signal was detected. The lesions varied in size up to about 5 mm. The testicles were otherwise symmetrical and of normal size, with homogeneous echogenicity and symmetric doppler signal. There was a 2 mm cyst on the left head of epididymis, otherwise the epididymis was normal. No additional findings. See video of the examination on the internet. The image shows lesions with the appearance of fatty tissue in the testicles. This must not be confused with microcalcifications, which are more common and as a rule harmless, but with an unclarified correlation with later testicular cancer (1).

Testicular lipomatosis is a pathognomonic finding that can be related to Cowden's syndrome, a rare, hereditary condition which entails an increased risk of the development of several types of cancer (2, 3). In light of this finding, the patient was therefore referred for assessment at the Section of Inherited Cancer, Oslo University Hospital. Other differential diagnoses associated with bilateral, multifocal intratesticular lesions include germ cell tumours, leukaemia, lymphoma, metastases, granulomatous disease (sarcoidosis, tuberculosis) and Leydig cell hyperplasia (3).

Cowden's syndrome (multiple hamartoma syndrome) is an autosomal dominant disorder caused by mutation of the gene *PTEN* on chromosome 10 (2). This tumour suppressor gene plays a central part in the

control of cell proliferation, differentiation and apoptosis, and a mutation promotes cellular proliferation in the ectoderm, mesoderm and endoderm. The syndrome is associated with numerous benign hamartomas on the skin and mucous membranes. Benign lesions can be seen in the thyroid and gastrointestinal tract. This patient group also has an elevated risk of cancer of the breast, thyroid, uterus and kidney. Almost all patients with this syndrome have a head circumference larger than the 97.5 percentile.

The patient in question has a large head and has had difficulty all his life in finding headgear to fit him. As a child he had a large «fatty lump» removed from under his arm. Subsequent to the ultrasound scan he has been found to have a defect in the *PTEN* gene c.675T>G, and because of the finding of multiple non-specific nodules in the thyroid, he has had a prophylactic total thyroidectomy.

A total of 37 persons with Cowden's syndrome are registered at the Section of Inherited Cancer at Oslo University Hospital, two of them with testicular lipomatosis. There are also some cases registered at other hospitals. Testicular lipomatosis is an unusual ultrasound finding that may be incorrectly interpreted as microcalcifications. Patients with this finding should be referred for genetic assessment with Cowden's syndrome in mind.

The patient has consented to the publication of this article.

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