Case Report: Testicular microlithiasis and epidermoid cysts – a common pathway?

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Abstract:

We present the case of a boy with antenatally diagnosed bilateral testicular microlithiasis. Following annual surveillance the child developed metachronous bilateral epidermoid cysts. These were subsequently excised with testicular sparing surgery. Currently there are only three cases of bilateral epidermoid cysts in the paediatric population, and none have been associated with testicular microlithiasis. This reignites the discussion regarding the histogenesis of epidermoid cysts and how its pathophysiology might relate to testicular microlithiasis.

KEY WORDS: testicular microlithiasis; epidermoid cyst; tubular degeneration; germ cell tumours.

Simple epidermoid cysts in pre-pubertal boys are rare benign tumours that account for 3-10% of all primary testicular tumours^{1, 2}. They are defined as an intra-parenchymal testicular cyst, filled with keratinized material and lined with squamous epithelium without any teratomatous elements or cutaneous adnexal structures³. Since the first case was reported in 1942, there have been over 22 cases in pre-pubertal children of which testicular microlithiasis has not been detected⁴.

The histogenesis of epidermoid cysts is contentious. However the prevailing theory maintains that they are a monophasic teratoma with adherence to a single cell line⁵. This is supported by similarities in age distribution and racial clustering when compared to other germ cell tumours^{6, 7}. Also, an identical neoplasm arising in the ovary is universally accepted as being of germ cell origin. However, the absence of testicular intra-epithelial neoplasia in some 'simple' cysts suggests there might be a separate group of epidermal cysts not related to monodermal teratomas⁸. An alternative theory states epidermoid cysts are a consequence of squamous metaplasia of the seminiferous epithelium or rete testis^{9, 10}.

CASE

A term boy of part aboriginal descent was born with prenatally diagnosed testicular microlithiasis and both testes in the scrotum. Diffuse calcification, both fine and coarse, was noted bilaterally. This prompted annual ultrasound surveillance which demonstrated appropriate testicular growth. There was no personal or family history of testicular pathology.

At age 7, he developed a right-sided testicular mass. On clinical examination the right testis was comparably larger than the left, with a palpable hard nodule in the upper half. Ultrasound imaging revealed an avascular hypoechoic lesion within the right testis with associated coarse calcifications. Intra-operatively a

well-encapsulated cystic mass measuring 20mm was excised through a midline scrotal incision with preservation of normal testicular tissue. Histological examination confirmed the diagnosis of epidermoid cyst. There was no evidence of intratubular germ cell neoplasia in the surrounding testicular tissue.

One year later he developed a sub-clinical lesion within his left testis. This was well circumscribed and hyperechoic measuring 12mm within the left testis. A cystic mass was subsequently enucleated from within the mid-testis through the existing scrotal incision. Histological examination revealed a keratin filled cyst lined by thin stratified squamous epithelium without atypia. The peripheries of the specimen revealed fibrotic testicular tissue with recognisable seminiferous tubules. Small foci of dystrophic calcification were also present in adjacent stroma. Two or three smaller cyst locules were seen in the section, also containing keratin. It is now 12 months since his last surgery, and he continues to be followed with annual ultrasound surveillance.

Discussion:

The sonographic findings consistent with an epidermoid cyst are varied. They can be systematically classified into four groups: a whorled or onion-ring appearance (type I), a hyperechoic mass with posterior acoustic shadowing (type II), a hypoechoic mass with a well-defined rim (type III) and a heterogeneous lesion with poorly defined margins and a few foci of calcification (type IV). Types I and III are highly suggestive of an epidermoid cyst^{11, 12}. The current case had a type III lesion and developed a type II lesion in the contralateral testis.

Testicular-sparing excision of the cyst is safe and effective as shown in large adult series with unilateral disease¹³. This is particularly important in children because of the lifetime risk of subsequent contralateral testis loss. Furthermore the most likely alternative diagnosis, teratoma, is universally benign in prepubertal boys¹⁴.

Bilateral epidermoid cysts are rare. Presently only twelve cases have been reported in the English literature¹¹. Only three of these cases have been in the paediatric population¹⁵⁻¹⁷.

Testicular microlithiasis was first observed in 1970 and has since become a well recognized diagnosis on high resolution ultrasound examination¹⁸. The prevalence of 'classic' testicular microlithiasis in asymptomatic boys is only 2%¹⁹. This has been associated with both benign and malignant tumours in adults, including intratesticular cysts²⁰. The association appears equally valid in the paediatric age group. As such long-term clinical and radiological follow-up is recommended²¹⁻²⁴.

Testicular microlithiasis occurs secondary to deposits within the seminiferous tubules consisting of calcified central cores and surrounding layers of cellular debris²⁵. This is due to a possible defect in the basement membrane of the seminiferous tubule allowing the sloughing of degenerated cells within the tubule, with resulting obstruction and degeneration. However, a competing

hypothesis is that the microliths are actually located in the testicular stroma, and are present from the early stages of gonadal embryogenesis²⁶.

The current case represents the first report of antenatally diagnosed testicular microlithiasis. It is consistent with the hypothesis that both testicular microlithiasis and germ cell tumours share a common pathway; tubular degeneration²⁷. This adds circumstantial evidence to those researchers developing histogenic theories that relate to epidermoid cysts and testicular microlithiasis.

Figure I:

Type II appearance of an epidermoid cyst within the left testis: a 9mm hyperechoic area within the mid-testis with posterior acoustic shadowing.

Figure II:

Histology of the epidermoid cyst showing stratified squamous epithelium with keratin and seminiferous tubules at the periphery.

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