CASE REPORT

TESTICULAR ADRENAL RESTS IN A CHILD WITH CONGENITAL ADRENAL HYPERPLASIA

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ABSTRACT

Males with congenital adrenal hyperplasia (CAH) may lead to development of testicular adrenal rests as a result of hyper-stimulation of aberrant adrenal cells in the testes. Congenital adrenal hyperplasia is a broad spectrum of autosomal recessive disorders caused by a deficiency of an enzyme involved in the production of glucocorticoids, which leads to decrease in cortisol and/or aldosterone production from the adrenal cortex. This causes an increase in production of adrenocorticotropic hormone (ACTH), therefore resulting in adrenal hyperplasia.

We present a case of a 7 year old boy with congenital adrenal hyperplasia and precocious puberty who was found to have bilateral testicular adrenal rests on ultrasonography. Diagnosis of this condition is important when evaluating young males with CAH because it can be often misinterpreted as a primary testicular germ cell tumor or any other common benign testicular lesion. These benign tumors generally present as multiple, bilateral hypoechoic masses on ultrasound, which serves as the primary imaging modality of choice.

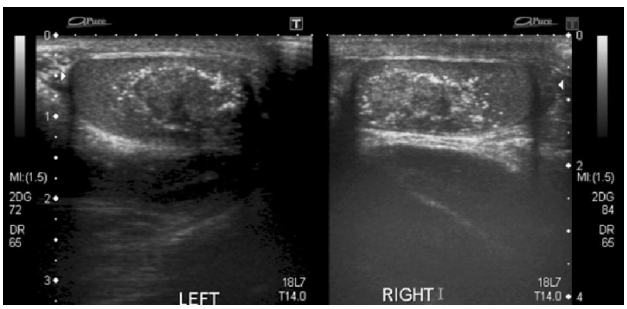
Key words: Congenital adrenal hyperplasia, testicular adrenal rest tumors, testicular ultrasonography

Introduction

Congenital adrenal hyperplasia (CAH) is an autosomal recessive genetic disorder that is caused by a deficient 21-hydroxylase (CYP21) enzyme in majority of cases. The incidence is approximately 1 in 15,000 births.[1] Testicular lesions commonly known as testicular adrenal rest tumors may occur in such patients. It has been postulated that these tumors arise from aberrant adrenal cells that migrate with the testes during the prenatal period. These lesions are usually benign, may be multiple and commonly occur bilaterally. They are mostly found in younger males and can occur in up to 94% of patients with congenital adrenal hyperplasia.[2]

Case Report

A 7 year old male child underwent evaluation for precocious puberty. The patient was diagnosed with congenital adrenal hyperplasia in infancy and had been intermittently receiving steroid supplementation over the course of his life. A testicular ultrasound was then performed which showed multiple sharply marginated, predominantly hypoechoic masses ranging from sizes 2-20 mm in both testes which occupied approximately 50% of the testicular parenchyma (Fig. 1). The lesions were predominantly round and partially coalescent with the testicular paren-



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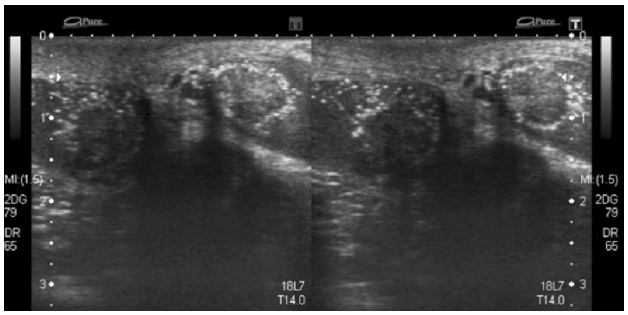

Figure 1: Sonographic images showing multiple hypoechoic rounded masses with peripheral hyperechoic rim in the testes of a patient with congenital adrenal hyperplasia. These were suggestive of adrenal rest tumors.

chyma. They showed a peripheral hyperechoic rim with scattered areas of microcalcifications in the testicular parenchyma. Doppler examination did not reveal any significantly increased color flow within the lesions.

Discussion

Ultrasound is vital in being the first line modality used in the diagnosis and monitoring of testicular adrenal rest tumors. Advantages of sonography include low cost and widespread availability. However, a well-known limitation of ultrasound is operator-dependence. Sonographic features of these lesions include the fact that they are frequently bilateral and can significantly vary in size.[2] Smaller lesions are usually sharply marginated and hypoechoic and mostly located near the mediastinum testis, although lesions larger than 2 cm may have central areas of increased echogenicity. Lesions that appear heterogeneous can have calcifications, a hyperechoic rim or internal spoke-like areas of echogenicity. This pattern was also observed in our patient. The lesions are usually hypovascular but may show variable amounts of vascularity on colour Doppler examination.[3] MRI may aid in correctly identifying exact extent of disease and can therefore play an adjunctive role when planning for testicular surgery.[4]

Imaging findings of testicular adrenal rest tumors can sometimes overlap with other testicular lesions such as primary germ cell tumors or sex cord tumors. Primary testicular tumors are however frequently unilateral.[5] Leydig's cell hyperplasia is another condition that presents with multiple discrete, hypoechoic, usually bilateral lesions, therefore closely mimicking testicular adrenal rest tumors. However, endocrine dysfunction such as raised serum testosterone, luteinizing hormone or hCG are often found, which helps in reaching a diagnosis.[5] Other similar testicular lesions include lymphoma and metastases, although both conditions occur in older males.

Adrenal rest tumors usually show regression when glucocorticoids are given in amounts that suppress endogenous ACTH levels, but can also considerably grow when supplementary hormone therapy is insufficient. Untreated lesions may compress the adjacent normal testicular parenchyma which can eventually lead to low testosterone production and subsequent subfertility. Surgical options may be warranted in those patients with chronic testicular pain or discomfort that is not amenable to hormone replacement therapy.[4]

Conclusion

Testicular sonography is important in the evaluation of males with congenital adrenal hyperplasia. The presence of testicular adrenal rest tumors in these patients is indicative of suboptimal hormone replacement therapy. In addition, these adrenal rest tumors may enlarge and lead to sterility. They may be mistaken for germ cell tumors such as Leydig cell tumors. Ultrasound, which serves as the primary imaging modality, along with appropriate hormonal laboratory investigations can help secure the diagnosis. Physicians and radiologists both need to be aware of this condition to avoid needless intervention. Treatment with glucocorticoid replacement therapy may result in regression of these lesions.

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