Testicular Microlithiasis related to McCune-Albright Syndrome
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History
12 year old with McCune-Albright syndrome.

Diagnosis
Testicular Microlithiasis related to McCune-Albright Syndrome

Additional Clinical
Recent partial thyroidectomy for "mass".

Discussion
McCune-Albright syndrome was initially described in females with precocious puberty, cafe-au-lait spots and fibrous dysplasia. This syndrome is linked to a mutation in the GNAS gene which codes for the alpha subunit of the stimulatory G protein which occurs in the post-zygomatic state. As such, this disease has a quite varied phenotypic expression in males and females.
Endocrinopathies are well recognized in McCune-Albright syndrome, including gonadotrophin independent precocious puberty, thyrotoxicosis, acromegaly and Cushing syndrome. Precocious puberty in males usually occurs later and slower than in females. Macro-orchidism in McCune-Albright is more common than true gonadotrophin independent sexual precocity and is likely related to Sertoli cell hyperactivity and elevated anti-mullerian hormone activity. Testicular enlargement and testicular microlithiasis are seen in 62% of males with McCune-Albright syndrome.
Fibrous dysplasia is related to replacement of the normal medullary space of bone with fibro-osseous tissue. Most often lesions are isolated to a single bone but multiple bones can be involved. Commonly involved bones include the craniofacial bones, ribs and long bones (the epiphysis is usually spared). McCune-Albright syndrome is found in 2-3% of patients with polyostotic fibrous dysplasia.

Findings
US-Extensive testicular microlithiasis on conventional and 3D rendered images.
CR-Multifocal osseous lesions involving bilateral ribs, left femur and left sphenoid bone. The medullary cavity is expanded with endosteal scalloping by a mixed sclerotic and lucent lesion in the proximal left femur. The right 5th rib is broadly expanded and the left 5th rib is focally expanded by lesions with a ground glass matrix; likewise the skull base is expanded by a lesion within the sphenoid bone.
CT-Axial images confirm lesion with ground glass matrix in the left sphenoid bone.

Reference

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