Increasing testicular size due to bilateral Large Cell Calcifying Sertoli Cell tumours (LCCSTs) in a peri-pubertal child with Carney Complex.

Geoghegan S1, Morrissey R1, Moore M2, O’Sullivan M2, Murphy M3, Irvine A2, Quinn F2, O’Connell SM1
1. Department of Paediatrics and Child Health, Cork University Hospital, Ireland
2. Department of Dermatology, South Infirmary Victoria University Hospital, Cork, Ireland
3. Department of Histopathology, Our Lady’s Hospital Crumlin, Dublin, Ireland
4. Department of Paediatric Dermatology, Our Lady’s Hospital Crumlin, Dublin, Ireland
5. Department of Paediatric Surgery, Our Lady’s Hospital Crumlin, Dublin, Ireland

The authors have no disclosures

Case Report:

Background: 11 year old boy. Diagnosis: Carney Complex; Heterozygous for a known nonsense mutation of the PPARG/1A gene (p.R42)

Presentation: Referred for Endocrine follow-up. Short stature; height 131.6. SDS -1.64; height velocity 5.63 SDS + 0.86

Pubertal Assessment: Testicular volume increased in 6 months from 4 to 8 cc and appeared bulky. No axillary or pubic hair.


Results: Biochemistry was consistent with a pre pubertal status. Bone age equivalent to chronological age. Testicular ultrasound showed bilateral multiple small echogenic foci not typical for microlithiasis irregularly spread throughout testes (FIG 1) Testicular Biopsy : Consistent with Large cell calcifying sertoli cell tumour. Also showed pubertal spermatogenesis (FIG 2)

Management: Conservative with close clinical and radiological follow up.

Discussion:

- Carney Complex (CNC) is a rare multi endocrine neoplasia syndrome associated with endocrine and non-endocrine tumours. 1
- Three types of testicular tumour have been described; Large cell calcifying Sertoli Tumours (LCCST), Leydig cell tumours and testicular tumours of adrenal origin.
- LCCST is a rare benign stromal tumour. That had been observed in 33-41% of males affected with Carney Complex, usually appearing in the first decade of life.
- It can be hormonally active, presenting with gynaecomastia or gonadotropin-independent precocious puberty. 2,3
- It is generally benign although malignant transformation has been described.
- In pre-pubertal patients conservative management is preferred, with anti sex steroid therapy as needed, to manage secondary sexual characteristics. 3
- LCCST can cause replacement obstruction of seminiferous tubules leading to reduce fertility. CNC patients have morphologically reduced sperm and abnormal sperm number. 4
- Testicular sparing surgery is often not suitable due to the multifocal nature of the tumour.

Conclusion:

Assessment of boys with CNC in the peri-pubertal age group can be complex. The clinical evaluation of growth and puberty must be balanced with known complications of this multi-system condition, with a high index of suspicion for the associated endocrine features. This case illustrates the challenges in monitoring pubertal progress and growth in adolescent boys with this condition.

References:

2. Esperanza Berenstein, Alicia Belgorosky, María T.G. de Díaz, and Marco A. Rivas. Testicular steroid biosynthesis in a boy with a large cell calcifying Sertoli cell tumor producing prepubertal gynaecomastia; Steroids 1995 60 Feb.