Hypogonadism in Noonan Syndrome: a case report

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INTRODUCTION

- Delayed puberty is multifactorial; it is important to recognise hypogonadism early & treat appropriately in order to prevent long term complications.

CASE HISTORY

- Noonan syndrome, diagnosed at 2 months age
- Clinical features: typical facial features (hypertelorism, ptosis, low set ears, & small pointed chin), heart murmur and bilateral cryptorchidism
- 22 months: patent ductus arteriosus closure
- 24 months: left testis was excised for testicular torsion
- Learning difficulties, attended special needs school
- 13 yrs: noted to be pre-pubertal, no pubertal growth spurt
- 18 yrs: no secondary sexual characteristics & bone age, 13-14 yrs.
- 18 yrs: Initiated on IM testosterone (sustanon)
- Did not receive testosterone regularly (non compliance)
- 22 years: 165 cm tall, reviewed in the adult endocrine clinic with absent secondary sexual features, low libido, micropenis, chest wall deformity & reduced muscle bulk.
- Commenced: testosterone undecanoate (1000 mg, 12 weekly, IM)
- Testosterone levels normalised (10 – 22 nmol/L) and this contributed to wellbeing, increased body hair and musculature.
- Associated vitamin D deficiency was treated.

INVESTIGATIONS (when seen in the Adult Endocrine Clinic)

- Testosterone 1.1 (nr 9 – 32 nmol/L)
- FSH 0.3 (nr 2-5 IU), LH 0.1 ( nr 2-6 IU)
- Prolactin, IGF1, TFTs: normal
- MRI pituitary: normal
- US Scrotum: right testis atrophic
- DEXA bone scan: osteoporosis (T score < -3.0 LS; -2.7 LFN)

DISCUSSION & CONCLUSION

- Noonan syndrome is an autosomal-dominant disorder
- Incidence: 1 in 1000 - 1 in 2500
- Common clinical features: facial dysmorphisms, congenital heart disease, chest deformity & short stature
- Cryptorchidism occurs in 60–69% of cases
- This case report highlights the difficulties associated with recognition of delayed puberty in conditions like Noonan syndrome (complicated by cryptorchidism).
- Timely initiation of testosterone is vital to prevent osteoporosis.
- These patients need regular follow up throughout childhood, puberty and into adulthood, in specialist multidisciplinary clinics.