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)

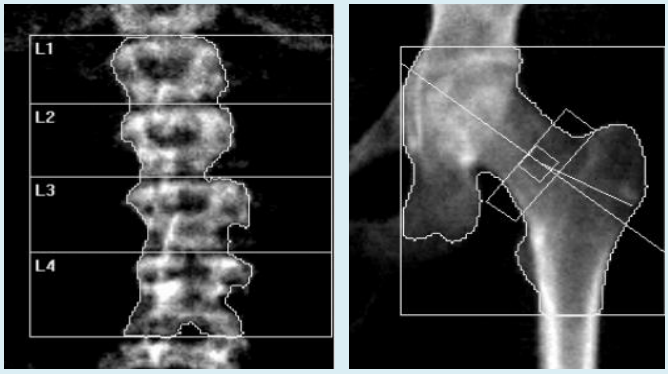


Figure 1. DEXA bone scan of lumbar spine and left femoral neck

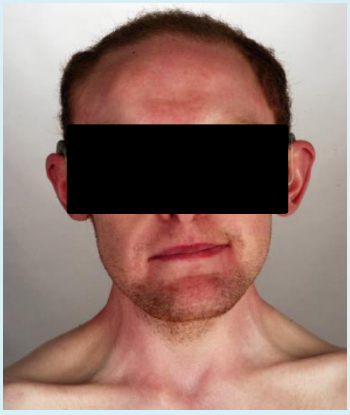
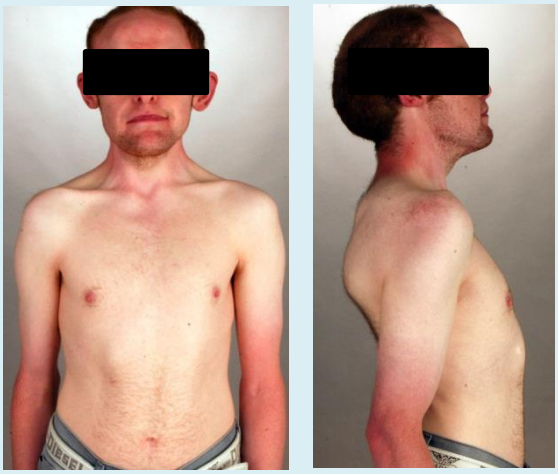


Figure 2. Head and neck abnormalities in Noonan syndrome including triangular face, low set ears, thick helix and webbed neck.



Figures 3 & 4. Thoracic abnormalities of Noonan syndrome including shield chest

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.