Abstract

Background

Short stature is mostly due to constitutional growth delay, idiopathic or familial causes. Other causes include malnutrition, chronic illness and hormonal deficiencies.

Neurofibromatosis is an autosomal dominant disease due to mutation in the NF1 gene causing a wide spectrum of clinical findings. It is also a rare cause of short stature.

Case presentation – A 15-year-old previously healthy school boy presented with short stature. He was found to be below 3 standard deviation of height for age with evidence of delayed puberty. Upon examination he had multiple large café au lait patches multiple lisch nodules and inguinal freckling suggestive of neurofibromatosis. He had growth hormone deficiency confirmed by insulin tolerance test with evidence of hypogonadotropic hypogonadism. Both of which though less emphasised are considered to be documented associations of neurofibromatosis. Imaging did not reveal tumours of pituitary origin.

Conclusion

Although short stature in neurofibromatosis could be due to varying causes such as suprasellar lesions, skeletal abnormalities and irradiation in treating various complications, it could also be exclusively due to hormonal deficiency leading to short stature.