PP-81

Profound Endocrinopathies in a Delayed Presentation of Transfusion-dependent Thalassaemia Intermedia

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INTRODUCTION

Two-thirds of Malaysian children with transfusion-dependent thalassaemia experiences at least one endocrine dysfunction. Endocrine and metabolic complications in these individuals are partly attributed to chronic iron overload and high cellular turnover for erythropoiesis. This is a case report of endocrinopathies in a delayed presentation of transfusion-dependent thalassaemia intermedia.

CASE

A 17-year-old-lady with no known comorbid presented with bilateral lower limb swelling, orthopnoea and reduced effort tolerance. She has short stature with primary amenorrhoea. One of her siblings has transfusion-dependent thalassaemia. On examination, her height was 124 cm (<5th centile), weight 24.9 kg (<5th centile) BMI 16.1 with absent secondary sexual characteristics (Tanner Stage I). Midparental height was 159.5 cm. She was jaundiced, pale with frontal bossing, maxillary expansion, saddle nose, depressed cranial vault suggestive of extramedullary haematopoesis, exhibited genu valgum and hepatosplenomegaly.

Investigations showed microcytic hypochromic anaemia with Hb of 1.7 g/dL (12.0-15.0), MCV 72.1fL (83.0-110.0), MCH 18.6 pg (27.0-32.0), iron 42.5 umol/L (5-31), ferritin 204 ug/L (10-291). Hb analysis confirmed beta thalassemia intermedia. Endocrine investigations revealed FBS 5.3 mmol/L, TSH 2.78 mIU/L(0.48-4.17), FT4 12.36 pmol/L (10.70-18.40), serum cortisol 457 nmol/L, IGF1 122 ug/L(246-533), prolactin 169.2 mU/L (89.9-489.7) corrected Ca 2.2 mmol/L (2.16-2.60) iPTH 6.4 pmol/L(1.95-8.49), FSH 0.5IU/L (2.2-10.1), LH <0.1IU/L (1.0-52.2), oestradiol 66.7 pmol/L, 25-hydroxyvitamin D 34.0 nmol/L(60-160.0). MRI pituitary was normal. Glucagon stimulation test and insulin tolerance test demonstrated inadequate growth hormone response with good cortisol peak. Radiographs showed ricket-like porous lesion with delayed bone age of 7 years old and platyspondyly of the vertebra. Bone mineral densitometry awaiting. She was initiated on regular blood hyper transfusion every 3 weeks, cholecalciferol 1000 iu/day, calcium carbonate 500 mg BD, folic acid 5 mg OD, growth hormone therapy with a view to inducing puberty subsequently.

CONCLUSION

Optimal quality care for these patients is the basic prerequisite to achieve good quality of life. Early intervention, screening and regular monitoring are imperative towards prevention of significant morbidities. Coordinated multidisciplinary approach is paramount to achieve this aim.