BC_P001

MEDICAL MANAGEMENT OF PAEDIATRIC CUSHING SYNDROME PRESENTING WITH SEVERE HYPERCORTISOLISM

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INTRODUCTION

Endogenous Cushing syndrome is a rare manifestation of chronic cortisol excess. It may present with severe hypercortisolism complicated by life-threatening opportunistic infections. Surgical management of a cortisol-secreting tumor is the mainstay of therapy. However, when the source is not found or when surgery is not feasible, medical therapies may be employed for rapid control of hypercortisolism.

CASE

A 13-year-old male presented with breathlessness for one day. He also had a cough associated with weight loss, weakness, and hallucinations for 1 month. On presentation, he was tachypneic and had moon facies, acne, fine moustache, limb wasting, and pigmented nail beds. He was also hypertensive and developed an episode of seizure during admission. Investigation results showed severe hypokalemia and lymphopenia. Radiological examination revealed pneumothorax with multiple lung cavitations and brain tuberculomas. Bronchoalveolar lavage (BAL) was positive for *Aspergillus fumigatus*. However, both BAL and CSF PCR and culture for TB were negative. Serum ACTH was 379 pg/mL (0–46), and morning serum cortisol was 2,948 nmol/L with loss of diurnal rhythm. His 24-hour urine cortisol was 14,252 nmol/24 hour (31.7–282). This is consistent with ACTH-dependent Cushing syndrome. He was started on anti-TB and anti-fungal treatment. Serum cortisol levels were persistently high while on high-dose intravenous dexamethasone treatment for TB meningitis. An MRI pituitary and a CT scan thorax, abdomen, and pelvis could not reveal an ACTH-secreting tumor source. Metyrapone was started and titrated upwards to control the hypercortisolism. Serum cortisol reduced to 200–300 nmol/L after 1 month, and 24-hour urinary cortisol was down to 22.4 nmol/24 hour after 4 months, requiring weaning of metyrapone doses.

CONCLUSION

Metyrapone is effective in the rapid control of severe hypercortisolism with life-threatening complications, as illustrated in this case. However, it warrants careful monitoring and titration.