PE-25

FULL BLOWN HYPERFUNCTIONING ENDOCRINOPATHIES IN MCCUNE ALBRIGHT SYNDROME: CHALLENGES IN MANAGEMENT

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INTRODUCTION

McCune Albright syndrome is a rare, wide spectrum of disease caused by post zygomatic GNAS mutation leading to activation of functions. It is characterized by typical skin hyperpigmentation and distribution, skeletal dysplasia and hyperfunctioning endocrinopathies.

RESULTS

NS, a 6-year-old Malay girl, presented at 5 months old with PV bleed. She was referred to us at 13 months old. She has extensive hyperpigmented lesions following line of Blaschko on nape, trunk and limbs with rickets changes. LH:0.1 mU/L, FSH:0.1 mU/L, Estradiol: 190 pmol/L. ALP:768 IU/L, Ca:2.68 mmol/L, PO4:1.28 mmol/L, TRP:86%, Mg:0.9 mmol/L, Vitamin D: 68 nmol/L. NS was started on oral cholecalciferol, calcitriol and Sandoz phosphate. At 3 years old, she was symptomatic for hyperthyroidism, TSH:<0.02, fT4:33 pmol/L needing carbimazole. Subsequently she continues to develop multiple long bone fractures associated with trivial falls. Intravenous Pamidronate was started for worsening bone pain. At 4 years 6 months, her PV bleed recurred every 2-3 monthly. LH: <0.2 mU/L, FSH: <0.2 mU/L, Estradiol:107 pmol/L. Pelvic ultrasound – showed bilateral ovarian follicular cyst . Bone age was advanced. Unfortunately she was unable to tolerate aromatase inhibitor (Letrozole). Simultaneously, she has height acceleration with frontal bossing. IGF-1: 168-249 ug/L (22-200), random GH: 4-12 ug/L (0.077-5.00). Her prolonged OGTT was not supressed. MRI brain and pituitary was normal. Long acting octreotide was started for GH excess. NS is currently under close surveillance of her endocrinopathies which has affected her mobility making her- dependent on parents for ADL. Otherwise she is asymptomatic.

CONCLUSION

This case illustrates the severe spectrum of McCune Albright syndrome. NS manifested with full blown hyperfunctioning endocrinopathies which proved to be challenging in management. She has gonadotropin-independent estrogen production, hyperthyroidism and GH excess. Her FGF-23 induced renal phosphate wasting is an important evidence of her fracture risk and complications.

PE-26

PSEUDOHYPOALDOSTERONISM — A CASE REPORT

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INTRODUCTION

Pseudohypoaldosteronism is a heterogeneous group of disorders manifested by end-organ resistance to mineralocorticoids. We describe a case of recurrent salt wasting, hyperkalaemia and metabolic acidosis from this condition.

RESULTS

A 2.5-year-old girl was first presented to us at day 8 of life with poor feeding, lethargy and in circulatory collapse requiring cardiopulmonary resuscitation. She was not dysmorphic, had unremarkable systemic findings with normal female genitalia. Laboratory parameters showed persistent hyponatraemia, hyperkalaemia and hyperchloraemic metabolic acidosis. She was initially treated with steroid therapy, but had shown no clinical improvement. The only significant hormonal work-ups were significantly elevated Renin and Aldosterone levels. Genetic confirmation test unfortunately was not available. Other secondary causes of adrenal resistance were also excluded. She subsequently had a few more episodes of salt-losing crisis, with improvement in terms of severity and the frequency of the event. These episodes were successfully treated with high dose sodium supplements and potassium-binding resin. She is now growing along a low growth percentile curve with reasonable age-corresponding developmental milestones.

CONCLUSION

Salt wasting crisis carries significant mortality and morbidity in children. Despite being rare, condition such as mineralocorticoid disorder must be considered when response to treatment does not follow common pattern.