hypopituitarism such as PROP1 gene. Even with similar mutation, individuals can have different levels of hormone deficiencies and be affected differently.

CONCLUSION

Careful evaluation of a dysmorphic child with features of midfacial hypoplasia is crucial to avoid missing congenital hypopituitarism. Early identification with comprehensive hormonal work-up is important to initiate hormonal therapy.

EP_P014

IT IS NOT WHAT IT SEEMS

https://doi.org/10.15605/jafes.040.S1.244

Chee Enn Han,[1] Raja Aimee Raja Abdullah,[1] Hui Lynn Khoo,[2] Phaik Khee Chong[3]

[1]Paediatric Endocrine Unit, Hospital Pulau Pinang, Malaysia
[2]Paediatric Department, Hospital Bukit Mertajam, Pulau Pinang, Malaysia
[3]Paediatric Department, Sunway Medical Center, Selangor, Malaysia

INTRODUCTION

Elevated thyrotropin-releasing hormone (TRH) in response to very low thyroxine (T4) level can lead to pituitary gland hyperplasia. This condition can mimic a pituitary adenoma, making it radiographically difficult to differentiate from hyperplasia.

CASE

A 10-year-8-month-old male presented to medical attention due to poor growth and learning difficulties. He was noted to be small since he was 5 years old. There were no significant medical or dietary issues. He was not dysmorphic, but growth parameters corresponded to a 4-year old male. He was prepubertal and there was no goiter noted. Investigations initially showed a normal thyroid function, low IGF-1 and a bone age which corresponds to a 6-month-old. An MRI of the brain was arranged several months later which revealed a pituitary macroadenoma compressing / indenting the optic chiasm. A repeat pituitary panel showed elevated prolactin which did not change post PEG. There was also new evidence of primary hypothyroidism. As the biochemical results exclude a macroadenoma, it was postulated that high TRH as a response to a low FT4 leads to the stimulation of the pituitary thyrotroph and lactotroph cells resulting in pituitary gland enlargement. This is thought to be rare in children but documented to occur in those with severe primary hypothyroidism with a TSH >50 mIU/L. This could be misdiagnosed as a macro-adenoma especially when thyroid function test was not performed prior to an MRI of the pituitary gland.

CONCLUSION

This case illustrates the importance of differentiating a macroadenoma from pituitary hyperplasia. The treatment differs with invasive surgery for macroadenoma and thyroxine replacement in pituitary hyperplasia.

EP_P015

A CASE OF FAMILIAL GLUCOCORTICOID RESISTANCE SYNDROME PRESENTING WITH HYPOKALEMIC PARALYSIS AND HYPERTENSION

https://doi.org/10.15605/jafes.040.S1.245

Muhammad Farisham and Suhaimi Hussain

Department of Pediatric, School of Medical Sciences, Universiti Sains Malaysia, Kelantan, Malaysia

INTRODUCTION

Familial glucocorticoid resistance syndrome (FGRS) is a rare condition leading to compensatory ACTH hypersecretion and excess adrenal steroid production. Patients often present with mineralocorticoid and androgen excess but without features of Cushing's syndrome.

CASE

A 17-year-old, male, Malay was referred for recurrent episodes of acute paralysis secondary to hypokalaemia and concomitant hypertension since the age of 7 years. Investigations showed persistent hypokalaemia with metabolic alkalosis. Clinically, he was not dysmorphic, taller for his age, increased skin pigmentation and was in pubertal with testicular volume of 6 ml and stretched penile length of 7 cm. Laboratory investigations showed a very marked increase in random serum cortisol of more than 2000 with elevated ACTH level. Luteinizing hormone-releasing hormone (LHRH) test confirmed a diagnosis of peripheral precocious puberty. Adrenal ultrasound did not show any suspicion of malignancy. He was started on oral dexamethasone and anti-hypertensive. He showed some improvement clinically and biochemically with no further history of paralysis and improvement in serum cortisol and potassium levels.

This patient presentation is consistent with FGRS where impaired cortisol signaling leads to compensatory increase in ACTH causing excess mineralocorticoid and sex hormones. Management focuses on reducing ACTH stimulation using high-dose dexamethasone and addressing complications such as hypertension and electrolyte imbalances.

CONCLUSION

Careful evaluation of a child presenting with unexplained hypertension, hypokalaemia and hyperpigmentation is