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

very important. Early diagnosis is crucial for appropriate management and genetic counselling.

EP_P016

WHEN GENITAL AMBIGUITY LEADS TO GENETIC DISCOVERY: A CASE OF NR5A1-RELATED DISORDERS OF SEXUAL DEVELOPMENT

https://doi.org/10.15605/jafes.040.S1.246

Ameerah Ali and Suhaimi Hussain
Department of Paediatrics, School of Medical Sciences, Universiti Sains Malaysia, Kelantan, Malaysia

INTRODUCTION
Disorders of sexual development (DSD) encompass a broad, heterogeneous groups of congenital conditions characterized by atypical development of genetic, gonadal, or phenotypic sex accompanied by abnormal development of internal and/or external genitalia. Early diagnosis is crucial to preserve fertility, ensure normal sexual function and support appropriate sex assignment, which significantly impact psychosocial well-being.

CASE
A child assigned female at birth was referred to a Paediatric Endocrinologist at 6 weeks old for evaluation of ambiguous genitalia. Clinical examination revealed penoscrotal hypospadias, rugated labioscrotal folds, palpable gonads with phallus size of 2 cm. The child is the youngest of 2 siblings, with no family history of consanguinity. Notably, the father had hypospadias, which was surgically corrected in childhood. Pelvic ultrasound revealed bilateral oval echogenic structure within labial fold, suggestive of testes, with no visible uterine structure. Hormonal investigations revealed a high testosterone level (13.1 nmol/L) and an antimullerian hormone level of 103 pmol/L, indicating normal Sertoli cell function. Karyotyping confirmed 46,XY genotypes. Further genetic testing identified a heterozygous variant of uncertain significance in the NR5A1 gene. The child was treated with monthly intramuscular testosterone for three months, resulting in phallus growth to 3 cm.

Thorough genital examination during newborn assessment is essential to prevent missed diagnoses of DSD. This patient was diagnosed with an undervirilized male phenotype associated with an NR5A1 mutation – a principal genetic alteration implicated in DSD. The NR5A1 gene plays a crucial role in early gonadal development, testis determination and steroidogenesis.

CONCLUSION
This case highlights the importance of early recognition and management of DSD. Genetic testing for NR5A1 mutation should be considered in cases of 46,XY DSD with ambiguous genitalia, particularly when accompanied by a family history of hypospadias.

EP_P017

LATE DIAGNOSIS OF OVO-TESTICULAR DISORDER

https://doi.org/10.15605/jafes.040.S1.247

Tan Wu Pin and Suhaimi Hussain
Department of Pediatrics, School of Medical Sciences, Universiti Sains Malaysia, Kelantan, Malaysia

INTRODUCTION
Ovo-testicular disorder of sex development (OT-DSD), formerly known as true hermaphroditism is a rare condition characterized by the presence of both ovarian and testicular tissue in an individual.

CASE
A 16-year-old Malay, female, was initially referred at the age of 9 years for further evaluation of ambiguous genitalia. She was born with ambiguous genitalia and was raised as a female. However, the family defaulted follow-up due to logistic issues. She had no history to suggest adrenal crises or progressive skin hyperpigmentation. Clinically, she was short and underweight for age (<3rd percentile), not dysmorphic, with normal hydration. Detailed genital examination revealed penoscrotal hypospadias with no palpable gonads. Biochemically, 17-OHP was normal, testosterone was elevated with evidence of germ cell failure having elevated LH and FSH. Her chromosomal analysis revealed 2 populations of cells: 46,XX (27) -77% and 46,XY (8)-23%. PCR-based molecular analysis for the SRY gene confirmed the absence of SRY gene. Genitogram at 10 years old showed no demonstrable urogenital fistula. She underwent diagnostic laparoscopy and HPE. The right gonads showed features consistent with ovotestis (true hermaphrodites) and left gonad features compatible with streak gonads. Her serial hormonal workups showed primary gonadal failure with elevated FSH (51.44) and LH(14.86) with low testosterone (<0.087) and estradiol (<18.35). She was started on estradiol valerate while waiting for her vaginal construction operation.

OT-DSD is rare and most reported cases occurred in individuals with 46,XX karyotype. However, 46,XY and mosaic karyotypes(46,XX/46 XY) have also been observed.