CONCLUSION

Osteopaenia of prematurity with nutritional deficiency is commonly observed in preterm infant. Nonetheless, the presence of severe rickets with inconsistent bone profile warrants further work-up for other alternative diagnoses, including VDDR1.

EP_P029

EXPLORING THE SPECTRUM OF HORMONAL DEFICIENCY IN PITUITARY STALK INTERRUPTION SYNDROME AND ITS OUTCOME WITH GROWTH HORMONE THERAPY: CASE SERIES FROM A TERTIARY PEDIATRIC ENDOCRINOLOGY CENTER IN MALAYSIA

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INTRODUCTION

Pituitary stalk interruption syndrome (PSIS) is a rare congenital condition characterized by either isolated or combined pituitary hormone deficiency. This paper presents 6 cases of pituitary stalk interruption syndrome diagnosed and managed in a tertiary Pediatric Endocrinology Center.

CASE

Analytical review of the medical records of patients followed up in Putrajaya Hospital, Malaysia from year 2017–2024 revealed 6 male patients with confirmed diagnosis of PSIS.

Among the cohort, 50% of them had significant perinatal events including severe neonatal jaundice, prolonged non-invasive ventilation support, sepsis or hypoxic events. A total of 17% were delivered via emergency caesarean section and the rest were born via unremarkable spontaneous vaginal delivery. Clinical presentation varied with 50% of patients presenting at birth with ambiguity of genitalia, another 50% of patients presented in adolescents with short stature and delayed puberty. Features of soft dysmorphism were observed in 67% of them. All patients have growth hormone deficiency, with 83% of them having additional pituitary hormone deficiency. Half of them have multiple pituitary hormone deficiencies. None of the patients in the cohort had clinical manifestations of diabetes insipidus. MRI imaging revealed absence of pituitary stalk on all patients. All patients who have been treated with growth hormone therapy showed improvement in height velocity with a mean of 10 (± 2.5) cm per year.

CONCLUSION

Children with PSIS often have a very broad spectrum of clinical and biochemical presentations. Screening and evaluation of the pituitary-hypothalamic hormone axis is critical to guide management. This clinical entity often presents with growth retardation and thus early diagnosis is critical to allow for timely management of these patients with growth hormone therapy.

EP_P030

A CASE SERIES OF POTENTIAL CONSEQUENCES FOLLOWING INTRAMUSCULAR INJECTIONS IN CHILDREN

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INTRODUCTION

Apart from regular vaccinations, there are limited instances where deep intramuscular (IM) injections are given to children. I.M gonadotropin agonist (GnRHa) is used for the treatment of central precocious puberty (CPP). It is given deep IM at the upper outer quadrant of the buttock. We describe three cases to demonstrate complications of this procedure.

CASE

Case 1. An 8-year-old female was diagnosed with CPP at the age of 5 when she presented with breast development. She has been receiving 3-monthly IM GnRHa since then. In the clinic, parents informed that she had a 1-week history of upper respiratory tract infection. She was afebrile and was given an injection as usual. Patient came back a week later complaining of pain and swelling at injection site. After inspection, a diagnosis of sterile abscess was made. She was treated with local and oral antibiotics.

Case 2. An 8-year-3-month-old female was diagnosed with CPP at the age of 7 years and 5 months. She was due for her 3rd i.m GnRHa. She has been anxious about injection pain and needed comfort from parents/nurses at each visit. She insisted on more pain relief prophylaxis. She came back after 1 week with circular erythematous rash and blister formation around injection site. Diagnosis of superficial burn secondary to ethylchloride spray was made.

Case 3. An 8-year-old female with underlying mild autism was started on GnRHa injections for CPP. At every clinic visit, she will cry, shout and throw tantrums which were attributed to injection anxiety. Parents and nurses had a lot of difficulty getting her ready for injections. On her last visit, the nurse who gave her the injection reported bite marks and bruises on her arm because the patient bit her.

CONCLUSION

Although rare, one should always take extra precautions when dealing with IM injections in children. Repeated procedures carry higher risk as mentioned in this case series.

EP_P031

PITUITARY HYPERPLASIA SECONDARY TO PRIMARY HYPOTHYROIDISM – A CASE REPORT

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INTRODUCTION

Primary hypothyroidism in children can present insidiously and mimic other systemic conditions, including neurological symptoms. In rare cases, it may lead to pituitary hyperplasia due to lack of negative feedback on thyrotrophs. Timely recognition is essential to prevent complications and avoid unnecessary neurosurgical intervention.

CASE

We report a case of an 8-year-old female who presented with chronic headaches, cold intolerance, constipation and frontal scalp hair thinning. Symptoms were insidious, with persistent headaches noted since the age of six. Despite multiple outpatient visits, including private pediatric and ophthalmology consultations, no clear diagnosis was made. Ophthalmological evaluations were also normal.

Due to persistent symptoms, neuroimaging was done to exclude intracranial mass or raised intracranial pressure. MRI of the brain and pituitary revealed enlarged pituitary gland measuring 7.7 mm (AP) x 12.4 mm (width) x 9.7 mm (height), with normal posterior pituitary bright spot and pituitary stalk. Other surrounding structures were normal. Thyroid function test (TFT) performed revealed an elevated TSH of 150 mIU/L with low fT4 at 8.7 pmol/L, consistent with primary hypothyroidism. Anti-thyroid peroxidase (TPO) and thyroglobulin antibodies were positive, confirming Hashimoto's thyroiditis. Other pituitary hormones were normal. She was initiated on levothyroxine, and serial TFTs demonstrated gradual improvement. MRI features were consistent with pituitary hyperplasia secondary to long-standing hypothyroidism (PHPH), and no neurosurgical intervention was warranted. A repeat MRI scan performed 10 months after commencement of treatment showed normal study with a pituitary gland measuring 6.7 mm (AP) x 12.2 mm (width) x 5.7mm (height). Her latest TFT has normalised with TSH of 3.76 mIU/L and fT4 of 18.5pmol/L on levothyroxine 37.5 mcg *qd* Monday to Friday, and 50 mcg *qd* on weekends.

CONCLUSION

PHPH is an uncommon cause of pituitary enlargement in children. This case highlights the importance of comprehensive endocrine assessment in children with chronic headaches. Early diagnosis and thyroid hormone replacement can lead to complete resolution of symptoms and regression of pituitary enlargement, avoiding misdiagnosis and overtreatment.

EP_P032

AN UNCOMMON CAUSE OF PERSISTENT HYPERCALCAEMIA WITH NEPHROCALCINOSIS IN INFANCY

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INTRODUCTION

Hypercalcaemia with nephrocalcinosis in infants is commonly caused by excessive calcium or vitamin D supplementation, neonatal primary hyperparathyroidism, subcutaneous fat necrosis or various genetic disorders.

CASE

We present a 6-month-old Indian male infant who was born preterm at 33 weeks, via elective LSCS for poly-hydramnios with weight of 1.33 kg, length of 46 cm and head circumference of 27 cm. His mother had severe polyhydramnios, requiring amnioreduction thrice. Both parents were consanguineous. During his 3-month-stay at NICU, he had persistent hypercalcaemia with inter-mittent polyuria. Serum calcium ranged: 2.5-2.9 mmol/L,