Genital abnormalities can be present but are not part of the diagnostic criteria. We describe one case of Silver-Russell Syndrome with atypical genitalia.

CASE

A 4-month-old male was referred to our clinic for atypical genitalia. He was born premature at 36 weeks 1 day, small for gestational age (SGA) with birth weight of 1.34 kg (<3rd centile), length of 41 cm (<3rd centile) and relative macrocephaly with head circumference of 32 cm (50th centile).

On further examination, he had dysmorphic features with prominent forehead, frontal bossing and triangular facies. His limbs were asymmetrical with hemihypertrophy of the left upper and lower limbs and bilateral 5th finger clinodactyly.

Examination of the genitalia revealed underdeveloped scrotum with no scrotal fusion, micropenis with stretched penile-length 0.5 cm (<10th percentile), penoscrotal hypospadias and non-palpable gonads. The EMS score was 0, in support of undervirilization.

Investigations revealed intact mini puberty with LH, FSH and testosterone of 1.6 U/L, 14.4 U/L and 2.95 nmol/L, respectively. Other anterior pituitary hormones were normal and 17-OHP was not elevated. Karyotype was normal with 46,XY. Short beta-hCG stimulation test revealed good testosterone level, with normal testosterone to androstenedione (T:A) ratio and testosterone to dihydrotestosterone (T:DHT) ratio, excluding both 17-hydroxysteroid deficiency and 5-alpha reductase deficiency respectively.

He was assessed by the genetic team and was noted to fulfill all the six NH-CSS criteria for clinical diagnosis of Silver-Russell syndrome. SRS methylation testing was sent to determine the molecular mechanism for future recurrence risk counselling.

CONCLUSION

Although SRS is primarily a growth disorder, it may present with atypical genitalia along with growth failure and dysmorphic features. Hence, it should be considered in the differential diagnosis of a newborn with dysmorphic features, SGA and atypical genitalia.

EP_P028

UNRAVELING THE MANIFESTATION OF VITAMIN D-DEPENDENT RICKETS TYPE 1 IN PREMATURE INFANT

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INTRODUCTION

Vitamin D-dependent rickets type 1 (VDDR1) is one of the genetic causes of calciopenic rickets. This rare autosomal recessive disorder is due to the defective 1-α hydroxylase which results in deficient active vitamin D or 1,25-dihydroxyvitamin D. It manifests as stunted growth, skeletal deformities and bone pain in young children. Diagnosing this uncommon disease requires a high index of clinical suspicion and is confirmed through genetic testing.

CASE

A seven-month-old female was born prematurely at 24 weeks of gestation with birth weight of 600 grams. Both parents were non-consanguineous. She had a stormy neonatal period with prolonged ventilation due to severe respiratory distress syndrome. In early neonatal phase, she had hypocalcaemia and hypophosphataemia, with subsequent gradual increment of alkaline phosphatase (ALP) – the overall picture initially pointing towards osteopaenia of prematurity. With time, she developed severe skeletal deformities which were bowing of the limbs, palpable widening of distal radius and double malleoli, Harrison's groove and long bones fracture. Apart from low calcium (1.88 mmol/L) and phosphate (1.32 mmol/L), other bone profiles showed: 25-hydroxyvitamin D levels were insufficient (47 nmol/L), both parathyroid hormone (PTH, 47.9 pmol/L) and serial ALP (1008 U/L) were elevated. A 1,25-dihydroxyvitamin D level was not investigated in view of financial limitations. Radiological imaging revealed rickets changes over the metaphyseal plate, including Looser's zone at the humerus and tibia. Considering the severe clinical manifestations of rickets, but inconsistent with insufficient level of stored 25-hydroxyvitamin D, this indicates deficient active vitamin D level that is consistent with clinical VDDR1. The whole exome sequencing was negative, but further workup for more expensive genetic study such as whole genome sequencing will incur additional costs.

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.