PE-15

UNEXPECTED ISOLATED HYPOPHOSPHATEMIC RICKETS ASSOCIATED WITH ELEMENTAL FORMULA FEEDING

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Sze Teik Teoh,[1,2] Annie Leong,[1,2] Arini Nuran Idris,[1] Poi Goik Lim[1]
[1]*Department of Paediatrics, Tunku Azizah Hospital, Kuala Lumpur, Malaysia*
[2]*Department of Paediatrics, Hospital Putrajaya, Malaysia*

INTRODUCTION
Phosphate deficiency is common in rickets but is accompanied by calcium or vitamin D deficiency (also named nutritional rickets). Isolated hypophosphatemic rickets (HR) without hypovitaminosis or defective renal phosphate handling is uncommon. Phosphate is abundant in diet and its GI absorption even though primarily mediated by vitamin D, is less tightly regulated with about 30% being passive. Recently, there has been an increase in such HR cases reported associated with the use of elemental formula (EF).

RESULTS
The patient was born prematurely at 31 weeks and diagnosed having Tetralogy-of-Fallot (TOF) with severe pulmonary stenosis requiring Blalock-Taussig (BT) shunt at 3-month-old. Postoperatively, he had heart failure and gastroesophageal-reflux-disease (GERD), for which he was treated with heart failure drugs (furosemide, spironolactone, captopril) and antireflux (omeprazole, domperidone), respectively. At 2-months-old, he was diagnosed with Cow's Milk Protein Allergy (CMPA) and EF was commenced. Both Neocate® and Comidagen® were used interchangeably. He developed rickets at 8-month-old with initial serum phosphate 0.5mmol/L, calcium 2.46mmol/L, ALP 1432 IU/L, and 25(OH)-Vitamin D 80 nmol/L (sufficient). His urine TRP was normal and radiological findings were consistent with rickets. Notably, he developed acute severe hypocalcemia with hyperphosphatemia, immediately following oral Sandoz phosphate, despite calcium supplementation. His subsequent response, however, was partial and the hypophosphatemia persisted. He was switched to soy-based formula at 10-months-old, with ensuing improvement in serum phosphate. He achieved biochemical and radiological healing of rickets within 3 months of follow-up.

CONCLUSION
HR in certain infants relating to the prolonged and sole use of EF had been reported elsewhere with its etiology not fully understood but could relate to the reduced bioavailability of phosphate in EF. Replacement with an alternative phosphate form could cause transient acute severe hypocalcemia and hyperphosphatemia possibly due to sudden upregulation of Na-Pi2b cotransporter in the gut after phosphate starvation. The cessation of EF reverses the pathology.

PE-16

PARTIAL ECTOPIC POSTERIOR PITUITARY GLAND IN A CHILD: A VARIANT OF AN ECTOPIC NEUROPHYPOPHYSIS SYNDROME

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LA Lim[1] and Subashini Rajoo[2]
[1]*Department of Medicine and Endocrinology, Penang General Hospital, Penang, Malaysia*
[2]*Department of Medicine and Endocrinology, Kuala Lumpur Hospital, Wilayah Persekutuan, Malaysia*

INTRODUCTION
Developmental abnormalitiy of the posterior pituitary can lead to an ectopic posterior pituitary at the median eminence or along the pituitary stalk with partial or complete pituitary stalk agenesis. An ectopic posterior pituitary gland is associated with isolated growth hormone or multiple anterior pituitary deficiencies but with normal posterior pituitary function. A partial ectopic pituitary gland is a less common entity described whereby there is presence of both an orthotopic (normally located) and ectopic neurohypophysis.

RESULTS
The patient first presented at 2 months old with prolonged jaundice. Thyroid function screening showed central hypothyroidism and she was started on L-thyroxine. She presented again at 2 years 10 months old with a hypoglycaemic seizure. Subsequently she was referred for further paediatric endocrine evaluation. Her IGF-1 was < 20mcg/L and glucagon stimulation test confirmed severe GH deficiency (peak GH 0.54ug/L) with an optimal cortisol peak of 698 nmol/L. Pituitary/brain MRI shows a hypoplastic pituitary gland and absence of pituitary stalk. There was a bright spot at the normal expected site of the neurohypophysis in the posterior sella with an additional ectopic focus of high signal intensity on T1-weighted imaging at the infundibulum measuring 3mm x 3mm which was most likely an additional and ectopic focus of the posterior pituitary gland. The pituitary stalk was not visualized. She was started on recombinant GH therapy and remains on L-thyroxine. Regular monitoring of her 8 am cortisol remains normal and she did not have symptoms or biochemistry suggestive of diabetes insipidus on follow-up.

CONCLUSION
Partial ectopic posterior pituitary gland is a variant of ectopic posterior pituitary that is a rarely described imaging entity. Although there is a wide differential diagnosis for T1-hyperintensities (e.g., lipid-containing lesions, protein, metallic substances, methemoglobin and calcifications) on MRI, the diagnosis can be narrowed with the aid of additional MRI sequences and clinical manifestations.

PE-17

HYPOTHYROIDISM IN DOWN SYNDROME (DS) CHILDREN WITH CONGENITAL HEART DISEASE (CHD)

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Hanaa Zainuddin, Norazah Zahari, Azriyanti Anuar Zaini
Department of Paediatrics, University of Malaya, Malaysia

INTRODUCTION
Down syndrome is the most common chromosomal disorder in liveborn infants, and is associated with thyroid dysfunction, estimated to occur in 4-8% of children. The reported incidence of congenital hypothyroidism (CHT) in children with Down syndrome is estimated 20 - 30 times higher than the incidence of CHT in the general population. We aim to investigate the prevalence of CHT in Down syndrome children with congenital heart disease (CHD) seen in our centre, as well as the aetiology of hypothyroidism in this group of paediatric patients.

METHODOLOGY
We performed a retrospective analysis of paediatric Down syndrome patients born between 1 January 1990 and 31 December 2020 who attended the paediatric cardiology clinic for routine review in the 3.25-year period between 1 September 2017 and 31 December 2020.

RESULTS
Children with DS accounted for 12.9% (n=62) of patients attending the paediatric cardiology clinic, with cyanotic CHD (cCHD) diagnosed in 29% (n=18) and acyanotic CHD (aCHD) in 71% (n=44).

The prevalence of hypothyroidism in our population of DS children with all types of CHD was 19.4% (n=12). However, among DS children with cCHD, the prevalence of hypothyroidism was 22.2% (n=4/18), which was higher than the prevalence of hypothyroidism in aCHD, 18.2% (n=8/44).

The mean age at presentation was 11.8 weeks (2 weeks – 32 weeks), and the mean TSH at presentation was 15.6 mIU/L (7.63 – 27.51 mIU/L).

All patients with suspected CHT (n=12) underwent thyroid scintigraphy at age 3. Six patients (50%) were confirmed to have permanent CHT: 42% (n=5) had thyroid dyshormonogenesis, and 8% (n=1) had thyroid dysgenesis. None had ectopic thyroid. Autoimmune hypothyroidism occurred in 8% (n=1). Subclinical hypothyroidism occurred in 42% (n=5).

CONCLUSION
The prevalence of CHT in DS children with CHD is 50% in this studied population. Children with DS tend to present later with subclinical hypothyroidism rather than high cord TSH. Routine screening in the first year of life is mandatory to detect thyroid abnormalities.

PE-18

CASE REPORT OF COMPLETE ANDROGEN INSENSITIVITY SYNDROME

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Mastura I, Jeanne SLW, Nalini MS, Cheng GG, Janet YHH
Paediatric Endocrine Unit, Hospital Putrajaya, Ministry of Health, Malaysia

INTRODUCTION
Androgen insensitivity syndrome (AIS), also known as testicular feminization, encompasses a wide range of phenotypes that are caused by numerous different mutations in the androgen receptor gene. AIS is an X-linked recessive disorder that is classified as complete, partial, or mild based on the phenotypic presentation.

RESULTS
A 2 year and 10-month-old child, presented to private practice for inguinal hernia at 15 months age. Herniotomy was performed at 20 months age and there was intra operative finding of gonad in the inguinal canal. Histology confirmed male gonad. This child was raised as girl as external genitalia was phenotypically female, no clitoromegaly with 2 perineal opening. Karyotyping confirmed 46 XY with variant in AR gene:c.5A>G(p. Glu2GIy). Pelvic UTS showed presence of vagina, no uterus and ovaries visualised .FSH was 4.1 mU/mL, LH was 2.7 mU/mL, testerone was normal with value of <0.35, Antimüllerian hormone was elevated 164.2 pmol/L.