Paediatrics E-Poster

EP_P034

A HEAVY DIAGNOSIS: CUSHING'S SYNDROME SECONDARY TO ADRENAL CORTICAL ADENOMA

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INTRODUCTION
Cushing's syndrome (CS) is very rare in childhood and adolescence. It may present as a diagnostic dilemma among clinicians.

CASE
We report an 11-year-old male with hypertensive emergency, congestive heart failure, pulmonary oedema and acute kidney injury. He had a two-year history of rapid weight gain and symptoms of obstructive sleep apnoea. He was obese with a body-mass-index (BMI) of 47 kg/m^2 (weight 109 kg, >95th centile; height 152 cm, 90th centile). He appeared depressed with severe acanthosis nigricans, truncal obesity, dorsocervical fat pad, striae and virilized.

He required intravenous labetalol and four antihypertensives on admission. Echocardiography revealed left ventricular impaired function (ejection fraction of 45%). Abdominal ultrasonography showed a left suprarenal lesion without renal artery stenosis. Abdominal computed tomography confirmed a lesion at the left suprarenal region (4.9 x 6.3 x 4.7 cm) and a right simple renal cyst. Urine biogenic amines and metanephrines were normal. He had elevated urinary cortisol at 3,574 nmol/24 hour (160-1,112) with loss of diurnal variation on salivary cortisol [midnight: 13.4 nmol/L (<11.3) and morning: 0.9 nmol/L (<24.1)] and on serum cortisol (midnight: 341.4 nmol/L and morning: 360.8 nmol/L). Further tests revealed suppressed ACTH: <0.33 pmol/L, and elevated serum dehydroepiandrosterone sulphate (DHEAS): 7.210 umol/L (0.660-6.700). There was no suppression on low- and high-dose dexamethasone suppression tests, consistent with ACTH-independent CS.

He underwent laparoscopic adrenalectomy, revealing an 8 x 5 cm, well-encapsulated left adrenal tumour. Preliminary histopathological analysis suggests adrenal adenoma. Perioperatively, he required stress-dose hydrocortisone (100 mg/m^2/day) and tapered to a physiological dose (7 mg/m^2/day) upon discharge. At follow-up, he was on two antihypertensive medications, demonstrated improved cardiac function (EF: 55%) and weight reduction (97 kg).

CONCLUSION
Although CS is rare in children, high levels of suspicion should be applied to those presenting with rapid onset obesity, hypertension and/or virilisation. Diagnosis of CS involves multiple investigative steps to guide treatment.

EP_P035

OCCULT MOSAICISM OF KARYOTYPING IN 45,X / 46,XY DSD

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INTRODUCTION
Disorder of sex development (DSD) with 45, X/46, XY mosaicism is a rare disorder. The prevalence is estimated to be less than 1:20,000.

CASE
A term baby, born with a good Apgar score at Hospital Sultan Abdul Halim. Genital examination showed atypical appearance with genital tubercle measuring 1.8 cm, bilateral labio-scrotal folds partially fused with single opening at perineum and no palpable gonads. External genitalia score (EGS) was 5/12. Ultrasound assessment revealed right inguinal lesion, equivocal for testis or inguinal hernia and small fluid-filled tubular structure posterior to the bladder which could represent either vagina or urogenital sinus. Genitogram report was consistent with vagina and complementary visualization of the uterus. Salt wasting was not observed during NICU stay. A 17-OHP screen on day-7-of-life reported 41.7 nmol/L. Urgent karyotyping initially reported 46, XY (cells analyzed 18, counted 10). On further assessment at the Hospital Sultanah Bahiyah, hormonal profile showed elevated gonadotrophins (FSH: 112.8I U/L, LH: 2.74I U/L and testosterone: 8.07 nmol/L). Follow-up 17-OHP was 82.8 nmol/L, and a short Synacthen test showed peak cortisol measuring 863.6 nmol/L. Serum AMH was 19.2 nmol/L (NV: 235.5-1125.9 for males and ≤31.2 for females). HCG stimulation showed increased testosterone: 2.00 nmol/L (Day 1) and 8.4 nmol/L (Day 3). Secondary analysis of initial chromosome samples with 48 cells analyzed and 15 counted [total 63], revealed mosaicism 45,X [14], 46,XY [49]. Surgical evaluation by 1-year-old reported presence of uterus with left streak gonad (removed) and right fimbriae-like-structure (biopsy) and suspected ovotestes (2 x 2 cm). HPE results are still pending by the time of report.

CONCLUSION
Chromosomal analysis with adequate cells is crucial in identifying subtypes of DSD. When mosaicism is suspected, a larger number of cells (at least 30) should be analyzed to accurately detect and characterize these conditions.

EP_P036

WHEN WATER BECOMES A FRENEMY: A CASE SERIES ON THIRSTY CHILDREN AND LITERATURE REVIEW

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INTRODUCTION
Polydipsia is defined as excessive thirst causing the consumption of large amounts of fluids, more than 2 liters/m^2/day in children, with consequential polyuria. It is of paramount importance to distinguish between diabetes insipidus (DI) and primary polydipsia as treatment differs, and inappropriate use of desmopressin can be detrimental in patients with primary polydipsia.

CASE
We present 3 children referred to the Paediatric Endocrine Clinic who exhibited a long history of excessive drinking.

Case 1. A 9-year-old male presented with an unquenchable thirst, drinking 6 to 8 L per day that required him to wake up 3-4 times nightly to drink water. A water deprivation test was performed, yielding inconclusive results, hence needed further investigation.

Case 2. A 9-year-old male's excessive drinking during school hours concerned his teachers, prompting an investigation. A subsequent water deprivation test confirmed primary polydipsia.

Case 3. A 2-year-old toddler presented with a progressive history of excessive drinking. Although his water deprivation test showed equivocal findings, his cranial MRI confirmed the diagnosis of central DI.

Fortunately, our patients did not demonstrate any red flags, such as dehydration, visual field loss, recurrent vomiting, headache or altered consciousness. Our school-going patients denied a history of school bullying or truancy. None of the children were on medication and there was no family history of similar symptoms.

CONCLUSION
These cases underscore the importance and limitations of a water deprivation test in diagnosing polydipsia and polyuria in children. Inconclusive results must be interpreted with caution and necessitate further investigation, as baseline clinical and biochemical variables cannot substitute for the water deprivation test.

EP_P037

THYROID CHANGES IN INFANTS OF MOTHERS WITH GRAVES' DISEASE: A CASE SERIES

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INTRODUCTION
Maternal Graves' disease (GD) can affect neonatal thyroid function. Maternal factors such as timing of diagnosis, TSH-receptor Ab (TRAb) titre, anti-thyroid medications and prior radioiodine therapy will affect outcome.

CASE
We describe six infants born to mothers with GD (2 mothers diagnosed before pregnancy and 4 mothers during pregnancy) in Hospital Sultanah Bahiyah in 2023-2024. All mothers had elevated TRAb, from 3.34 IU/L to >40 IU/L, taken at 16-35 weeks of gestation. Five were treated with carbimazole (10-40 mg daily). Four started treatment during pregnancy and one prior to pregnancy. One mother had RAI before pregnancy and her infant had negative TRAb. Two (2/6) neonates had low birth weight and four (4/6) were premature. One neonate had fetal goiter and required elective LSCS via EXIT procedure by paediatric ORL. This neonate's goitre resolved following L-thyroxine initiation and was extubated within 3 days. Four neonates had elevated TRAb ranging 11.2I U/L to 39.5I U/L. Within 1st week, five had hyperthyroidism, of whom, one was symptomatic for moderate tachycardia. Two required low dose carbimazole for 4-6 weeks. The highest fT4 was 61.24 pmol/L. One patient with no thyrotoxicosis initially developed central hypothyroidism by 1-month-old. Of those with initial transient hyperthyroidism, three (3/5) developed central hypothyroidism thereafter requiring L-thyroxine. Two of them (2/3) had transient central hypothyroidism that resolved between 2-month-old and 1-year-7-month-old. By the time of report, three (3/6) infants still require L-thyroxine of whom two (2/3) had central hypothyroidism with prior hyperthyroidism. All these infants have appropriate growth and development during follow-up.