Paediatrics E-Poster

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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.

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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.