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A CASE SERIES OF SUSPECTED CONGENITAL ADRENAL HYPERPLASIA IN ONE WEEK IN A REGIONAL ENDOCRINE UNIT

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Congenital adrenal hyperplasia (CAH) is the most common cause of salt wasting during infancy. During workup other rarer causes should also be considered. We report three cases of suspected CAH, aiming to highlight that CAH is not always the cause of adrenal insufficiency.

Case 1 A two-week-old term female born to non-consanguineous parents, presented with failure to thrive, hyponatraemia, Na 118 mmol/L (132–144 mmol/L), hyperkalaemia, K 7.2 mmol/L (3.5–5.1 mmol/L), plasma (am) cortisol 134 nmol/L (101–536 nmol/L). A diagnosis of CAH was considered and Hydrocortisone (HC), Fludrocortisone and Sodium Chloride (NaCl) (30%) were commenced. Urinary steroid profile (USP) was sent to Kings College, London diagnosing Aldosterone Synthase Deficiency (ASD) type I, prompting to stop HC and to continue Fludrocortisone. Further testing showed high renin >300 pg/ml/hour (normal <61 pg/ml/hour), low aldosterone 50 pg/ml (normal >300–1900 pg/ml). 17-OH Progesterone and Adrenocorticotrophic-hormone (ACTH) were normal.

Case 2 A term 3.7 kg female, antenatally diagnosed bilateral cleft lip & palate, delivered by caesarean-section, to 44-year-old primi mother was admitted to NICU on day-3 with low Sodium 122 mmol/L (132–144 mmol/L), high Potassium 6.2 mmol/L (3.5–5.1 mmol/L) normal Cortisol (am) 377nmol/L (101–536nmol/L), serum/urine osmolality. CAH was suspected and HC, Fludrocortisone and NaCl (30%) were commenced. MRI brain showed partial agenesis of corpus callosum, with normal pituitary. Further investigations: am cortisol: 85 nmol/L (101–536nmol/L), ACTH (am): 23.1 pmol/L (1.1–13.2pmol/L), aldosterone, renin, 17 OH progesterone, USP, urine-biochemistry, LH, FSH, B-HCG, AMH, TFT's & Blood glucose were normal. Microarray-CGH showed 16p13.11 microdeletion (Father same deletion), karyotype 46XX. Trial of stopping Fludrocortisone & NaCl (30%) was unsuccessful and NaCl (30%) was recommenced. She failed a synacthen test and remains on HC and NaCl (30%). Current diagnosis is adrenal insufficiency related to 16p13.11 microdeletion.

Case 3 A term 23 day old male was transferred to Cork University Hospital (CUH) from a peripheral hospital with 12 hours of projectile, non-bilious vomiting. On examination he was dehydrated, unwell, with hyper-pigmented scrotum (normal descended testes) and normal phallus. Initial tests revealed hyponatremia, Na 108 mmol/L (132–144 mmol/L), & hyperkalemia, K 6.9 mmol/L (3.5–5.1 mmol/L). CAH was suspected and HC (8–10 mg/m²/day), Fludrocortisone 200mcg & NaCl (30%) were commenced. 17-OH progesterone was >400 nmol/L (<4nmol/L). Genetic tests confirmed a diagnosis of CAH secondary to 21-hydroxylase deficiency.

Conclusion All three cases presented within a week in CUH with acute adrenal insufficiency with salt wasting, thought to

be CAH. However; only one was confirmed CAH. During the workup, USP and genetic testing were key to distinguish these rarer diagnoses.

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OHVIRA SYNDROME- AN UNUSUAL VARIANT IN AN INFANT

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Obstructed hemivagina and ipsilateral renal anomaly (OHVIRA) syndrome is a rare Mullerian duct anomaly. The classical description is that of a uterus didelphys, unilateral obstructed hemivagina caused by a longitudinal *sagittal* vaginal septum and ipsilateral renal *agenesis*. An updated classification allows for inclusion of a wide variety of renal anomalies such as dysplastic kidneys, pelvic kidneys, or ectopic ureters.

We report a rare case of OHVIRA syndrome in a fourteen month old girl, with a *single normal uterus*, obstructed hemivagina with a longitudinal *coronal* vaginal septum and unilateral dysplastic kidney. Antenatal ultrasound scan at twenty weeks of gestation revealed a suspected hydrocolpos and dysplastic kidney. The infant was born at term via elective caesarean section (maternal request for elective repeat caesarean). Of note mum had a personal history of uterus didelphys which was diagnosed at her first delivery (emergency caesarean section). Mum had no known associated renal anomaly.

The patient was reviewed at five weeks old in the paediatric and adolescent gynaecology (PAG) clinic. She was thriving, but had a small vaginal bulge on examination. Repeat abdominal ultrasound scan revealed a single uterus, a normal right kidney and dysplastic left kidney, and a small hydrocolpos. As the patient was asymptomatic this was managed conservatively, and mum was advised to contact the PAG service if any concerns. At twelve months of age mum reported copious grey non-odorous discharge into the patient's nappy. This was thought to be the hydrocolpos draining spontaneously and the patient was called in for review in the PAG clinic, however on clinical examination a large mass was palpable to the umbilicus. Repeat ultrasound suggested a persistent large hydrocolpos (10 × 6 × 4 cm).

Examination under anaesthesia was performed. This demonstrated an elongated vagina with no cervix palpable at the superior aspect. A bulging septum in the *coronal* plane was then noted and a small punctum identified just inferior to the urethral orifice. This was opened draining 300mls of fluid. A single cervix was located in this obstructed anterior hemivagina.

Clinical and sonographic review at six weeks post-operative confirmed a single uterus, unilateral dysplastic kidney and no recurrence of hydrocolpos or discharge and the infant was thriving.

Cases of OHVIRA syndrome with variations in the classically described anatomical anomalies found in OHVIRA have been reported, however our patients variant with a *single normal uterus*, and a *coronal septum* may be a new variant not previously identified.