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 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 Hydrocortisone. 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 Adrenocorticotropic-hormone (ACTH) were normal.

Case 2 A term 3.7 kg female, antenatally diagnosed bilateral clef 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) and mum had no known associated renal anomaly. Hydrocolpos (10 × 6 × 4 cm).

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.