

A Unique Case of a Child with Two Inherited Salt-Losing Conditions



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Introduction

Salt losing conditions can be challenging to manage well during infancy and childhood. Prolonged sodium depletion is known to be associated with growth failure and other problems in young animals¹. We describe a unique case of a child with both Salt-Wasting 21-Hydroxylase Deficiency (SW21OHD) and Cystic Fibrosis (CF).

Presentation & Diagnosis

A male infant, AB, birth weight 3500gm (-0.2SDS), presented with salt-wasting crisis on D9 of life with weight loss (12%), hyponatraemia (120mmol/L) and hyperkalemia (8.4mmol/L).

Serum 17-hydroxyprogesterone (17OHP) was >1000nmol/L. Genetic analysis indicated he was a compound heterozygote with 2 pathogenic mutations including a deletion/gene-to-pseudogene conversion and a c955C>T. A diagnosis of SW21OHD was made.

Neonatal screening confirmed the co-existence of CF with genotype ΔF508/A455E (some preservation of chloride transport). AB's sweat chloride was 73mmol/L and his stool elastase was normal (pancreatic sufficient).

Management

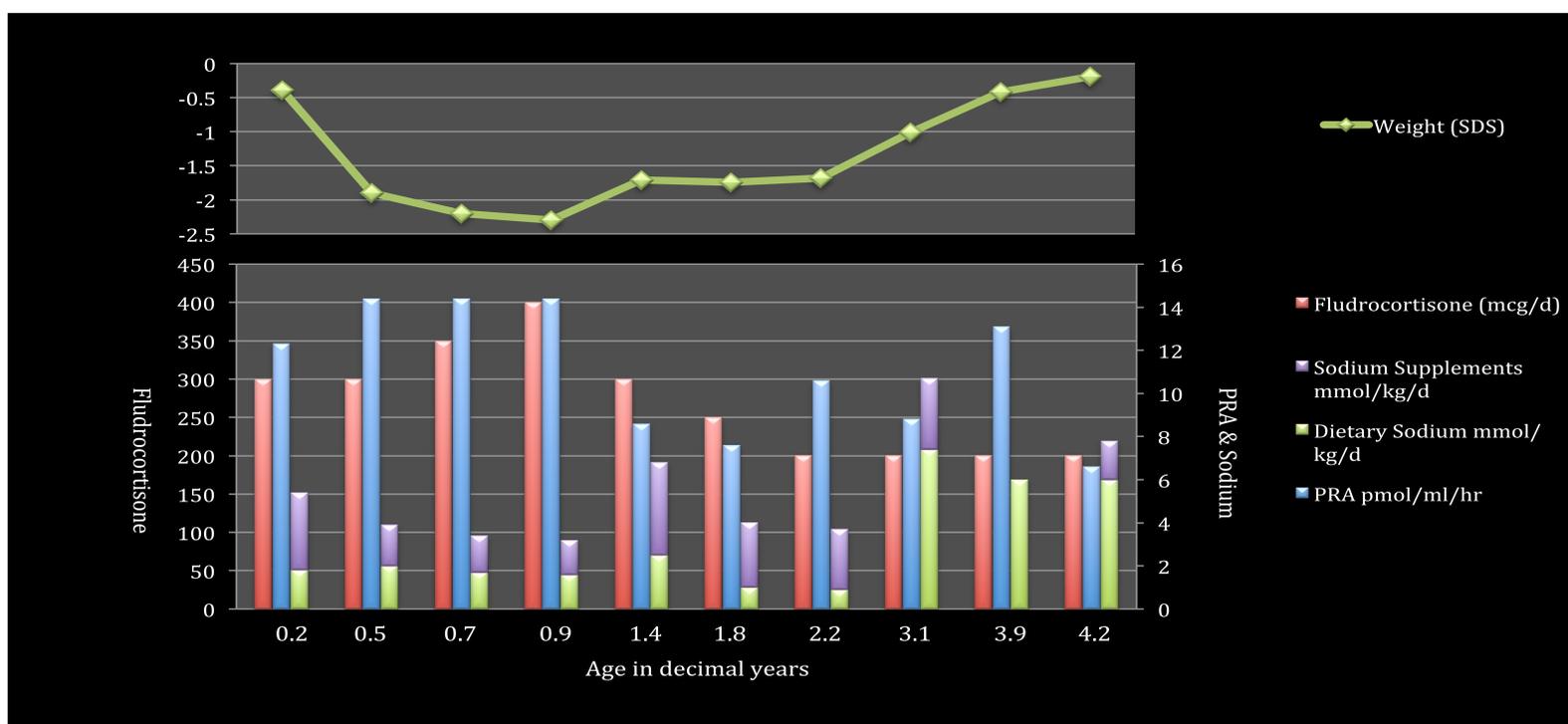
At diagnosis AB was established on treatment with Hydrocortisone (HC) 13mg/m²/day, Fludrocortisone (FC) 300micrograms/day together with salt (NaCl) supplements 4mmols/kg/day and Flucloxacillin.

Serial measurements of serum sodium (Na⁺), potassium (K⁺) and systolic blood pressure (BP) were within normal limits throughout infancy and early childhood.

However, plasma renin activity (PRA) was elevated, particularly following the introduction of proprietary low-salt weaning foods. This was associated with poor weight gain, reaching a nadir of 7.5kg (-2.3SDS) at 0.9yrs. FC dose was increased to 400mcg/d but with little beneficial effect.

Salt supplements were increased gradually, reaching a maximum daily salt intake of 11mmol/kg/day at around 2.2yrs. FC dose was decreased gradually to 200mcg/day in view of its potential growth suppressing effects.

At 4.1yrs, AB's weight had increased to 16.7kgs (-0.1SDS), height 100.7cms (-0.7SDS), height velocity 6.6cms/yr and PRA was within normal limits.



Discussion

In our tertiary paediatric centre, infants with SW21OHD or CF are commenced on NaCl supplements (2-4mmols/kg/d) at diagnosis and these are continued until the child is fully weaned.

Our child, AB, grew poorly during infancy and early childhood despite provision of fludrocortisone, hydrocortisone & NaCl supplements. The PRA was persistently elevated suggesting chronic sodium depletion despite normal values of Na⁺, K⁺ and systolic BP. Increasing the dose of fludrocortisone was not effective in reducing PRA and achieving weight gain. AB has required continuing NaCl supplementation, which is provided as commercially available salt sachets (each containing about 13mmols NaCl).



Conclusions

This case highlights a number of important issues:

1. The importance of monitoring salt intake and ensuring adequate salt intake in infants with salt-wasting conditions to allow optimal growth.
2. The relationship between salt intake, FC dose and PRA and their relative roles in the optimal management of SW21OHD.
3. The possible role of salt depletion in growth failure in infants and children with cystic fibrosis.

The endocrine & metabolic adaptation to salt depletion in CF patients has not been well studied and requires further evaluation.

References

1. SJ Wassner. *Pediatr. Nephrol.* 1991; 5:501-504