

Congenital Adrenal Hyperplasia – Subtle presentations with critical electrolyte imbalances & cardiac arrhythmias

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Background

Congenital adrenal hyperplasia (CAH) can present with salt loss, hyperkalemia and arrhythmias in the neonatal period.

If CAH is not diagnosed and treated early, neonates are susceptible to sudden death in the first few weeks of life.

This problem is particularly critical in boys who have no genital ambiguity to alert physicians before the onset of dehydration and shock.

Method / Case Report

History

A 10 day old boy presented to the emergency department with a history of:

- Poor feeding
- Weight loss (5% of body weight)
- Lethargy
- Parents were consanguineous.

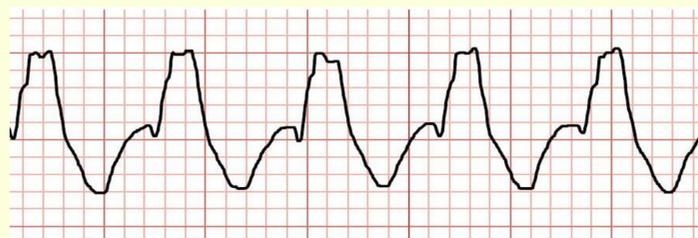
Examination

- SpO2 99% on air
- Normal breath sounds
- Bradycardic, heart rate of 65bpm and systolic murmur
- Normal male genitalia with scrotal hyperpigmentation

Investigations

ECG: broad complex bradycardia

Blood Gas Analysis



pH 7.3

Na+ 112mEq/L

K+ 9.7mEq/L

Diagnosis

A presumptive diagnosis of CAH was made and treatment was commenced

Treatment

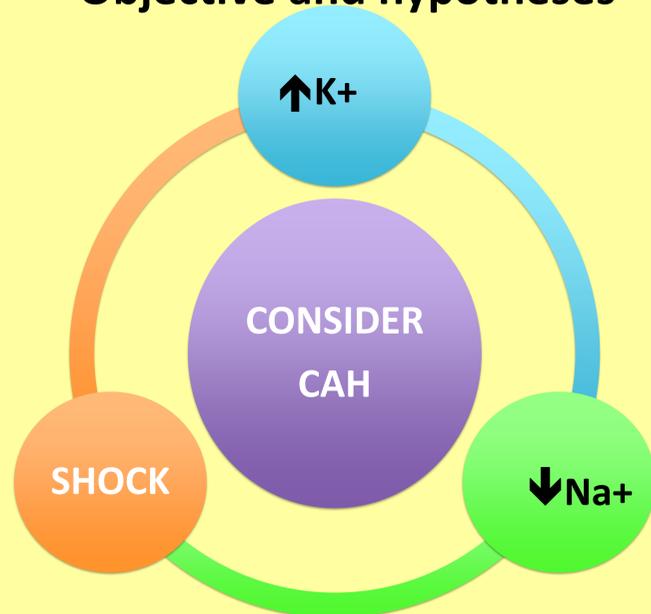
- Saline Bolus
- Nebulised Salbutamol
- Calcium Gluconate
- IV hydrocortisone

Following administration of calcium gluconate, his heart rate increased to 150/minute with regular sinus rhythm.



He was later confirmed to have 21-hydroxylase deficiency with a 17-OHP of 280nmol/L. His heart murmur was confirmed to be innocent.

Objective and hypotheses



We recommend that a diagnosis of CAH be considered in neonates presenting with hyponatraemia, hyperkalaemia and shock.

Results and Conclusion

- In the index case, CAH presented with a life-threatening arrhythmia. The presence of a cardiac murmur was a confounding factor.
- Timing and appropriate medical management in the emergency unit can prevent fatality.
- Finally, a combination of hyperkalaemia and hyponatraemia with metabolic acidosis is suggestive of adrenal insufficiency.
- Treatment with hydrocortisone gives excellent response.

