Pass the salt please!

Electrolyte Disturbance in an Infant with Cystic Fibrosis

Aneurin Young
ST4 Trainee in Paediatrics
Wessex Deanery

Tracey Farnon
Consultant Paediatrician
Salisbury District Hospital

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Background

Normal pregnancy and delivery. Had routine IM vitamin K.

- IRT blood spot screening diagnosis of cystic fibrosis in neonatal period with ΔF508 homozygous mutation.
- Diagnosis confirmed by sweat test (Cl 86mmol/l).
- Started routine therapy – physiotherapy, pancreatic lipase supplementation (Creon), multivitamin, Vit E.
- Exclusively breastfed with excellent initial weight gain.
- Atrial septal defect diagnosed.
- Mother noticed unusually sweaty baby.
- Advised to wean early but gagging on solids and unable to wean.
Admission

At five months of age:
- Persistent wet cough with near-continuous courses of oral antibiotics at home.
- Intermittent focal chest signs and borderline sats.
- Increased vomiting and not weaning.

Admitted for IV antibiotics
- Cough swab prior to admission - *Pseudomonas aeruginosa*.
- Suboptimal response to antibiotics.
- Parents struggling to cope with diagnosis and complications.

Twelve days into admission…
Widespread Bruising
Blood Tests

FBC: normal
INR: “very high”
APTT: “very high”
[Recommend repeat]

Na\(^+\): 123
K\(^+\): 2.3
Urea: 1.6
Creat: 15

ΔΔ: Registrar cock-up

Repeat bloods

INR: >10
APTT: 3.1
[both corrected with 50% normal plasma]

Na\(^+\): 124
K\(^+\): 2.2
Urea: 1.9
Creat: 19
Work-Up

d/w Haematologist, check factors, most likely vitamin K deficiency. Give IV Vit K.

What next?

Weird salts +

Very confusing

Pseudo-Bartter (lots more blood tests)

VBG

- pH: 7.61
- pCO₂: 6.6
- Gluc: 7.8
- Lact: 2.6
- BE: 24.5
- HCO₃: 48.1
Bartter Syndrome

= a rare genetic syndrome affecting the cells of the thick ascending limb of the loop of Henle causing impaired reabsorption of sodium, chloride and potassium.

Results in:
- Low serum Na\(^+\)
- Low serum Cl\(^-\)
- Low serum K\(^+\)
- Metabolic alkalosis
- Elevated plasma renin and aldosterone
- Inappropriately high urine Na\(^+\), K\(^+\) and Cl\(^-\)

VAN’T HOFF WG Molecular developments in renal tubulopathies
*Archives of Disease in Childhood* 2000;83:189-191.
Pseudo-Bartter Syndrome

= any cause of non-renal sodium, chloride and potassium loss which mimics the biochemical features of Bartter Syndrome

Causes of Pseudo-Bartter Syndrome:
• Diarrhoea or laxative abuse
• Vomiting or surreptitious purging (including pyloric stenosis)
• Loss of electrolytes in sweat e.g. in infants with cystic fibrosis
• Congenital chloride diarrhoea
• Mitochondrial cytopathies
• (Loop diuretic overuse/misuse)

Results in:
• Low serum Na⁺
• Low serum Cl⁻
• Low serum K⁺
• Metabolic alkalosis
• Elevated plasma renin and aldosterone
• Appropriately low urine Na⁺, K⁺ and Cl⁻

Cystic Fibrosis

= a genetic disorder caused by dysfunction of the cystic fibrosis transmembrane conductance regulator and characterised by production of thick mucus affecting the lungs and causing exocrine gland failure.
Electrolyte Loss in Sweat in CF

Why do babies with CF get Pseudo-Bartter Syndrome?
Our Patient

Vitamin K Deficiency Bleeding
- Exclusively breastfed and unable to wean
- Not tolerating Creon?
- Pancreatic insufficiency
- IV antibiotics disturbing gut flora
- Severe phenotype?

Pseudo-Bartter Syndrome
- Vomiting ?GORD
- Sweaty baby?
- Warm(ish) weather
- ASD
- Disturbing gut flora
Further Biochemistry

Serum osmolality: 247
Urine osmolality: 78

Urine Na: <10mmol/l

Renin: 3854pmol/l (16.6-145)
Aldosterone: 521pmol/l (140-2490)
Aldosterone/Renin Ratio: 0.1
Management

VKDB – initial IV Vit K 5mg completely resolved coagulopathy with no new bruises. Oral Vit K 2mg OD started.

Pseudo-Bartter

- NaCl 2mmol/kg/day PO (split QDS)
- KCl 0.5mmol/kg/day PO (split QDS)
- Titrated to bloods until stable at NaCl and KCl both 1.5mmol/kg/day
Progress

Serum Sodium and Potassium

<table>
<thead>
<tr>
<th>Day</th>
<th>Serum Na (mmol/l)</th>
<th>Serum K (mmol/l)</th>
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<tbody>
<tr>
<td>01</td>
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<tr>
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<td>09</td>
<td>135</td>
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</tbody>
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Day 11-27:

- Sodium: 135-140 mmol/l
- Potassium: 3-5 mmol/l
Progress

pH and pCO$_2$

Blood pH

pCO2 (kPa)

Day
Progress

BE and $\text{HCO}_3^-$

Base Excess vs. Day

Serum Bicarbonate (mEq/l)
Questions: Should we routinely supplement with NaCl?

Nutritional Management of Cystic Fibrosis

5.7.3 Good practice points
- Urinary sodium:creatinine ratio or urinary sodium should be routinely checked in all breast and formula fed infants.
- Infants with low urinary sodium:creatinine ratio or urinary sodium should be given additional sodium supplementation with sodium chloride solution to initially provide an additional 1–2 mmol/Kg/day. Urinary sodium should be checked and additional supplements given as necessary.

Should we routinely supplement with Vit K?

5.5.4 Good practice points
- Routine supplementation with vitamin K1 (phytomenadione) in people with CF and pancreatic insufficiency is recommended