Status epilepticus – an rare cause of unusual electrolytes!
Dr. P Gallogly
Guys and St Thomas’ NHS Foundation Trust

Case Presentation
- A 29 year old Caucasian female presented with a two week history of diarrhoea and vertigo.
- This has been preceded by six months of leg cramps and fatigue, for which she had attended her GP once and A&E three times, with one previous 24hour hospital admission to the acute admissions ward.
- Examination was normal apart from a blood pressure of 100/64mmHg and bilateral nystagmus on lateral gaze
- Shortly after admission, the patient developed status epilepticus, requiring intubation and an ITU admission
- Investigations (below) showed severe hypomagnesaemia, and following Mg and K replacement, the patient made a full recovery

<table>
<thead>
<tr>
<th>FBC</th>
<th>Normal</th>
<th>CRP</th>
<th>Normal</th>
<th>VBG on admission (on air)</th>
<th>ABG (post seizure on 15L O2)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Na</td>
<td>149</td>
<td>K</td>
<td>2.7 (L)</td>
<td>pH 7.51</td>
<td>7.12</td>
</tr>
<tr>
<td>Chloride</td>
<td>90 (L)</td>
<td>Calcium</td>
<td>2.30</td>
<td>pO2 5.0kPa</td>
<td>31.5 kPa</td>
</tr>
<tr>
<td>Mg</td>
<td>0.13 (L)</td>
<td>Phosphate</td>
<td>0.90</td>
<td>PC02 7.21kPa</td>
<td>6.2 kPa</td>
</tr>
<tr>
<td>LFTs</td>
<td>Normal</td>
<td>Coag</td>
<td>Normal</td>
<td>HCO3 36</td>
<td>34</td>
</tr>
<tr>
<td>Renin</td>
<td>3.9pmol/ml/hr (H)</td>
<td>Aldosterone</td>
<td>980pmol/L (H)</td>
<td>Lactate 1.0</td>
<td>15.6</td>
</tr>
<tr>
<td>Cortisol</td>
<td>320</td>
<td>24hr urinary Ca</td>
<td>1.6mmol (L)</td>
<td>BE +14</td>
<td>+6.3</td>
</tr>
</tbody>
</table>

Plain CT Head
MRI Head | Normal
ECG | QTc 483. Otherwise normal.

Diagnosis
- After exclusion of the common causes of low magnesium, full workup revealed metabolic alkalosis, hypochloraemia, hypokalaemia, secondary hyperaldosteronism and hypocalcuria, in keeping with a diagnosis of **Gitelman syndrome**

Gitelman Syndrome
- Rare (1 in 40,000 prevalence) autosomal recessive condition affecting SLC12A3 gene encoding the thiazide sensitive Na-Cl co-transporter in distal convoluted tubule (DCT), leading to a characteristic profile of electrolyte abnormalities [1]

<table>
<thead>
<tr>
<th>Impaired Na reabsorption in DCT leads to mild salt wasting and either normotension/hypotension</th>
<th>Volume reduction causes a secondary hyperaldosteronism</th>
<th>Raised aldosterone increases Na absorption to maintain sodium haemostasis, at the expense of loss of K and H+</th>
<th>This results in a metabolic alkalosis with hypokalaemia and normal sodium</th>
</tr>
</thead>
</table>

Symptoms
- Most patients are asymptomatic, resulting in the condition often remaining undiagnosed until adult life
- Classically patients have periods of good health with symptomatic periods, often when Mg/K deficits are exacerbated by fever/diarrhoea/vomiting (as in our case study)

<table>
<thead>
<tr>
<th>Seizures, irritability</th>
<th>Arrhythmias, prolonged QTc</th>
<th>Polyuria</th>
<th>Chondrocalcinosis</th>
<th>Muscle cramps, weakness, fatigue</th>
</tr>
</thead>
</table>

Diagnosis
- Relies on clinical suspicion and demonstration of classical electrolyte abnormalities +/- genetic tests

Management
- Asymptomatic – monitoring only
- Symptomatic – potassium/magnesium replacement (oral/IV depending on severity)
- Amiloride/spironolactone for patients where potassium cannot be maintained with oral supplements

Discussion
- Evaluation of the patient’s 3 A&E attendances (including one medical admission) showed metabolic alkalosis and mild hypokalaemia which were noted on each episode but were not investigated further. Magnesium was not checked on any occasion, and no diagnosis/differential for the cause of the patients symptoms was formulated
- Three key hospital opportunities to make a diagnosis were missed, leading to a potentially preventable ITU admission with status epilepticus
- Reasons discussed for possible missed diagnosis included: Not believing a mild alkalosis was an acute medical issue, hypomagnesaemia not being considered in the differential and the non-specific nature of symptoms/recurring hospital presentation being interpreted as a functional illness

Case Learning Points
- Electrolyte disturbance commonly cause vague symptoms. Always check full electrolyte profile in cases of diagnostic uncertainty
- An unexplained alkalosis should be taken just as seriously as an acidosis
- Whilst mildly abnormal bloods may not require an acute admission, follow-up should be arranged to ensure resolution or further investigation if needed
- Listen to the patient. Repeat presentation patients that have not had a full specialist workup or confirmed diagnosis (even a functional diagnosis) should warrant investigation
- Common things are common, but once excluded rarer diagnosis should be considered in a patient with unexplained symptoms