Recurrent severe hypernatremia in a young man with hydrocephalus and normal osmoregulatory function

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Context
Chronic hypernatremia is a rare clinical entity, encountered predominantly in the elderly population.
In the younger population, chronic hypernatremia is often as consequence of failure to generate thirst in response to osmotic stimuli.
We report the first case of a patient with a disconnect between normal osmoregulated thirst and abnormal drinking behaviour.

Case report
A 24 year old man presented with gait instability, myalgia, and cognitive decline. He was dehydrated and had marked facial dysmorphism. No focal neurological signs were noted on examination. The patient’s biochemical picture was consistent with hypernatremic dehydration (Table 1).

Table 1. Results of laboratory investigations during admission

<table>
<thead>
<tr>
<th>Parameter</th>
<th>Day 1</th>
<th>Day 2</th>
<th>Day 3</th>
<th>Day 4</th>
<th>Day 5</th>
</tr>
</thead>
<tbody>
<tr>
<td>Na (ref 133-146 mmol/L)</td>
<td>175</td>
<td>170</td>
<td>161</td>
<td>150</td>
<td>140</td>
</tr>
<tr>
<td>K (ref 3.5-5.3 mmol/L)</td>
<td>2.2</td>
<td>2.3</td>
<td>4.1</td>
<td>4.2</td>
<td>3.9</td>
</tr>
<tr>
<td>Urea (ref 2.7-7.8 mmol/L)</td>
<td>16.9</td>
<td>15.6</td>
<td>12.7</td>
<td>11.3</td>
<td>8</td>
</tr>
<tr>
<td>Creatinine (ref 64-104 mmol/L)</td>
<td>135</td>
<td>128</td>
<td>108</td>
<td>90</td>
<td>76</td>
</tr>
<tr>
<td>Posm (ref 275-295 mOsm/kg)</td>
<td>374</td>
<td>-</td>
<td>344</td>
<td>-</td>
<td>296</td>
</tr>
<tr>
<td>Uosm</td>
<td>894</td>
<td>963</td>
<td>-</td>
<td>-</td>
<td>496</td>
</tr>
<tr>
<td>CK (ref 0-170 U/L)</td>
<td>15540</td>
<td>41297</td>
<td>37260</td>
<td>5288</td>
<td>2320</td>
</tr>
</tbody>
</table>

MRI brain showed marked hydrocephalus (Image 1).

The patient was treated with IV dextrose and recovered without neurologic sequelae.

A reset osmostat for thirst and AVP release was suspected and following discharge, the patient underwent 5% saline infusion.1 Plasma AVP rose from 1.4 to 7.3 pmol/l and linear regression analysis defined a normal osmotic threshold for AVP release of 283 mOsm/kg; pAVP = 0.27 (pOsm-283), r = 0.88, p = 0.002. Thirst (visual analogue scale) rose appropriately, with a normal osmotic threshold: thirst = 0.31 (pOsm – 283), r = 0.98, p< 0.0001. The patient therefore had normal osmoregulatory function (Fig 1).

However, in the 30 mins following infusion, the patient only drank 400 mls water, despite normal thirst dynamics (normal water intake 700-1200 mls).³

Therefore, there was a disconnect between normal osmoregulated thirst and his abnormal drinking behaviour.

The patient was recommended to have a fixed fluid intake of 2-3 litres daily. He remained eunatremic until ten years later, when during febrile illness, his fluid intake fell and he again presented with hypernatremic dehydration.

The patient has had no progression in his neuroimaging and no development of neurological features after 10 years of follow-up.

Key points
• Our patient has an unique disconnect between the osmoregulatory function and fluid intake.
• The mechanism of this abnormality is unknown.
• It is difficult to dissociate this patient’s abnormal drinking behaviour from his marked hydrocephalus, as distortion of the complex anatomic structures that translate thirst appreciation into the central drive to drink seems inevitable.³
• Learned behaviour with fixed fluid intake has prevented day today hypernatremia, but the patient remains vulnerable to severe life threatening hypernatremia during intercurrent febrile illnesses.

References