Severe Hyponatremia and Repeated Intestinal Resections for Intestinal Dysmotility Mimicking Congenital Aganglionic Megacolon Due to Delay in the Diagnosis of Congenital Hypothyroidism

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BACKGROUND
Congenital hypothyroidism (CH) may present with non-specific signs and symptoms though majority of infants can be asymptomatic. Therefore, underestimation and delay in diagnosis may result in severe complications. Herein, we report delay in the diagnosis of CH in a female infant, who developed severe neurodevelopmental delay, severe hyponatremia and abdominal distention mimicking congenital aganglionic megacolon which required repeated surgery and relieved complications.

CASE REPORT

Born after 40 weeks uneventful gestation
• Birth weight was 4000 gr
• Developed prolonged neonatal jaundice
• Treated with phototherapy and phenobarbital

1 month old
• Vomiting, abdominal distention and poor feeding
• Diagnosis of intestinal obstruction due to congenital aganglionic megacolon was considered
• Surgical resection and reanastomosis performed

1-5 months
• Complaints had not been resolved thereby required subsequent six different operations
• 5 months old referred our hospital

5 months old
• Weight: 3800 gr (<3 pc)
• Anterior fontanelle: 3”3 cm
• Dried and crumped skin
• Abdominal distention
• Neurodevelopmental delay.

2nd day of hospitalization
• Developed mild hyponatremia. (Na :125 mEq/L, K:4.6 mEq/L)
• No signs of volume expansion or depletion, vomiting, diarrhea or gastrostomy tube loss
• Corrected with saline replacement.
• Histopathologic evaluation of previously resected intestinal specimens revealed a normal ganglion cell including colon samples.

3rd day of hospitalization
• Free T4 (FT4): 0.4 pmol/L (7.8-14.4) and TSH: >100 µIU/ml (0.34-5.6)
• L-thyroxin therapy  a daily dose of 50 µg (14.2 µg/kg/day) was started which than tapered to a daily dose of 37.5 µg (10.7 µg/kg/day) on day 4
• Thyroid imaging using 99mTc-pertechnetate thyroid scan revealed thyroid agenesis (Figure 1)

11th day of hospitalization
• Poor feeding, vomiting, abdominal distention, and respiratory distress
• Laboratory investigations:
  • Severe hyponatremia with normal potassium level (Na :106 mEq/L, K:4.3 mEq/L)
  • FT4: 2.73 pmol/L (7.8-14.4) and TSH >100 µIU/ml (n: 0.34- 5.6)
  • L-thyroxin dose was increased to 50 µg per day
  • Infusion of hypertonic saline and subsequent replacement of sodium deficit was commenced

13th day of hospitalization
• Plasma sodium level was 120 mEq/L
• Normal renin and aldosterone levels in course of hyponatremia (Table 1)
• FT4 were still at hypothryoid level with elevated TSH >100 µIU/ml
• To achieve euthyroid state L-thyroxin dose was increased up to 100 µg/day which was tapered after attaining a normal FT4 level.
• After achievement of euthyroid state plasma sodium level rised to normal ranges and remained stable with no requirement of sodium replacement

Table 1. Presenting and follow up biochemical and hormonal characteristics of patient

<table>
<thead>
<tr>
<th></th>
<th>Day 1</th>
<th>Day 2</th>
<th>Day 3</th>
<th>Day 6</th>
<th>Day 11</th>
<th>Day 12</th>
<th>Day 13</th>
<th>Day 16</th>
</tr>
</thead>
<tbody>
<tr>
<td>Plasma Na (mEq/L)</td>
<td>132</td>
<td>125</td>
<td>126</td>
<td>134</td>
<td>106</td>
<td>117</td>
<td>120</td>
<td>136</td>
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<tr>
<td>K (mEq/L)</td>
<td>3.74</td>
<td>4.6</td>
<td>4.4</td>
<td>4.9</td>
<td>8.0</td>
<td>93</td>
<td>95</td>
<td>110</td>
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<tr>
<td>Cl (mEq/L)</td>
<td>103</td>
<td>96</td>
<td>91</td>
<td>108</td>
<td>80</td>
<td>93</td>
<td>95</td>
<td>110</td>
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<tr>
<td>Urine (Na mEq/L)</td>
<td>119</td>
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<tr>
<td>Uric acid (mg/dL)</td>
<td>1.57</td>
<td>2.2</td>
<td>0.43</td>
<td>0.82</td>
<td>0.94</td>
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<tr>
<td>Osmolality</td>
<td></td>
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<td>245</td>
<td></td>
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<tr>
<td>TSH (µIU/ml)</td>
<td>&gt;100</td>
<td>&gt;100</td>
<td>&gt;100</td>
<td></td>
<td></td>
<td></td>
<td>11</td>
<td>33.4</td>
</tr>
<tr>
<td>FT4 (pmol/L) (N:7.86-14.5)</td>
<td>0.4</td>
<td>2.73</td>
<td></td>
<td></td>
<td></td>
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<td>5.56</td>
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<td>Renin (pg/mL) (N: 2.7-16.5)</td>
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<td></td>
<td></td>
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<tr>
<td>Aldosterone (pg/mL) (N:10-160)</td>
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<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>142</td>
</tr>
</tbody>
</table>

CONCLUSION
Since presenting symptoms are variable and non-specific, for prompt diagnosis and immediate treatment, congenital hypothyroidism should be kept in mind in the differential diagnosis of neonates with persistent abdominal distention mimicking aganglionic megacolon and severe hyponatremia of unknown origin.