Hereditary primary hypomagnesemia (generic term)

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Abstract

Several syndromes are known in which hypomagnesemia is the primary defect. So far, one recessive form caused by a defect of intestinal Mg²⁺ absorption, and two recessive forms and one dominant form all resulting from defective renal reabsorption have been described. In order to differentiate primary renal loss of Mg²⁺ from intestinal loss, a 24-h urinary Mg²⁺ excretion or the fractional excretion of Mg²⁺ in a random urine specimen should be obtained. Daily excretion of more than 10-30 mg Mg²⁺ or a fractional excretion of Mg²⁺ exceeding 2% in an individual with normal renal function points to renal Mg²⁺ wasting.

Keywords

hypomagnesemia with hypocalcemia, hypomagnesemia hypercalciurianephrocalcinosis, dominant/recessive renal hypomagnesemia, gitelman syndrome

Several syndromes are known in which hypomagnesemia is the primary defect (Table I). So far, one recessive form caused by a defect of intestinal Mg²⁺ absorption, and two recessive forms and one dominant form all resulting from defective renal reabsorption have been described. In order to differentiate primary renal loss of Mg²⁺ from intestinal loss, a 24-h urinary Mg²⁺ excretion or the fractional excretion of Mg²⁺ in a random urine specimen should be obtained. Daily excretion of more than 10-30 mg Mg²⁺ or a fractional excretion of Mg²⁺ exceeding 2% in an individual with normal renal function points to renal Mg²⁺ wasting (Agus et al., 1999).

Table I: Genetic loci for hereditary primary hypomagnesemia disorders

<table>
<thead>
<tr>
<th>Disease</th>
<th>Locus</th>
<th>Gene</th>
<th>Reference</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hypomagnesemia with Hypocalcemia (HSH)</td>
<td>Secondary 9q12-9q22.2</td>
<td>?</td>
<td>Walder et al, 1997</td>
</tr>
<tr>
<td>Hypomagnesemia</td>
<td>3q28-3q29</td>
<td>CLDN16</td>
<td>Simon et al, 1999</td>
</tr>
<tr>
<td>Hypercalciuria</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Nephrocalcinosis (HN)</td>
<td>11q23</td>
<td>FXYD2</td>
<td>Meij et al, 1999, 2000</td>
</tr>
<tr>
<td>Dominant renal hypomagnesemia/hypercalciuria</td>
<td>16q13</td>
<td>SLC12A3</td>
<td>Simon et al, 1996</td>
</tr>
</tbody>
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References
