Corrigendum: The Spectrum of SPTA1-Associated Hereditary Spherocytosis

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A Corrigendum on

The Spectrum of SPTA1-Associated Hereditary Spherocytosis


In the original article, there was a mistake in Table 1 as published. The SPTA1 mutation of Allele 2 in Patient 1, is stated as “c.4294T>A (p.L1432∗).” The correct mutation should read “c.4295del (p.L1432∗).” The corrected Table 1 appears below.

The authors apologize for this error and state that this does not change the scientific conclusions of the article in any way. The original article has been updated.

REFERENCES


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### TABLE 1 | Genetic mutations and associated phenotype in HS due to SPTA1 mutations.

<table>
<thead>
<tr>
<th>Phenotype</th>
<th>Patient</th>
<th>Allele 1</th>
<th>Allele 2</th>
<th>Age at time of report and comments</th>
<th>Ektacytometry</th>
<th>α-spectrin in RBC ghosts (%) of control</th>
</tr>
</thead>
<tbody>
<tr>
<td>GROUP I (patients 1–4) Severe, recessive HS (transfusion-dependent, responding to splenectomy)</td>
<td>1</td>
<td>c.4339-99C &gt; T c.4295del (p.L1432*)</td>
<td></td>
<td>11 year-old, chronic transfusion requirement with partial response to partial splenectomy, resolved after total splenectomy</td>
<td>54%</td>
<td></td>
</tr>
<tr>
<td></td>
<td>2</td>
<td>c.4339-99C &gt; T c.5102A &gt; T (p.L1701*)</td>
<td></td>
<td>7 year-old, chronic transfusion requirement, improved with partial splenectomy</td>
<td>64%</td>
<td></td>
</tr>
<tr>
<td></td>
<td>3</td>
<td>c.4339-99C &gt; T c.3267A &gt; T (p.L1089*)</td>
<td></td>
<td>11 year-old, not splenectomized due to family preference, continues to require frequent transfusions</td>
<td>Not evaluable in a transfused sample</td>
<td></td>
</tr>
<tr>
<td></td>
<td>4</td>
<td>Mutation not identified Gross deletion of SPTA1</td>
<td></td>
<td>3.5 year-old, RT-PCR demonstrated significantly decreased α-spectrin expression; hemoglobin has normalized after recent splenectomy</td>
<td>Not evaluable in a transfused sample</td>
<td></td>
</tr>
<tr>
<td>GROUP II (patients 5–8) Severe to moderately severe, recessive HS</td>
<td>5</td>
<td>c.4339-99C &gt; T c.1120C &gt; T (p.R374*)</td>
<td></td>
<td>4 year-old, chronic transfusion requirement for first three years with improved pattern since.</td>
<td>Sample not provided after age 3, when transfusion-independent</td>
<td></td>
</tr>
<tr>
<td></td>
<td>6</td>
<td>c.4339-99C &gt; T c.1351-1G &gt; T</td>
<td></td>
<td>7 year-old, occasional transfusion requirement, resolved after splenectomy at 5 years of age</td>
<td>59%</td>
<td></td>
</tr>
<tr>
<td></td>
<td>7</td>
<td>c.4339-99C &gt; T c.2671C &gt; T (p.R891*)</td>
<td></td>
<td>4 year-old, has not been transfused so far, Hgb 7.1-8.9 g/dL, ARC 420-572 x 10^3/µl.</td>
<td>61%</td>
<td></td>
</tr>
<tr>
<td></td>
<td>8</td>
<td>c.4339-99C &gt; T c.3257delT</td>
<td></td>
<td>8 year-old, transfused once as neonate, Hgb 10.6–11.9 g/dL, ARC 354–635 x 10^3/µL, now Hgb 15–16 g/dL with normal ARC after splenectomy at 6 years of age (splenectomy performed because of chronic abdominal pain due to co-morbidities)</td>
<td>Not performed.</td>
<td></td>
</tr>
<tr>
<td>GROUP III (patients 9–11) Life-threatening anemia in utero leading to fatal hydrops fetalis if untreated (transfusion-dependent, not responding to splenectomy)</td>
<td>9</td>
<td>c.4206delG (fs) c.4180delT (fs) in haplotype with c.6631C &gt; T (p.R2211C)</td>
<td></td>
<td>Died at birth. Post-mortem diagnosis from parental studies and DNA extracted from liver tissue saved in paraffin block</td>
<td>N/A</td>
<td></td>
</tr>
<tr>
<td></td>
<td>10</td>
<td>c.6788+11C &gt; T c.6788+11C &gt; T</td>
<td></td>
<td>11 year-old, born prematurely at EGA of 33 weeks with hydrops fetalis, remained transfusion-dependent even after splenectomy; now doing well after matched sibling transplant</td>
<td>Not evaluable in a transfused sample (required chronic transfusions up until bone marrow transplant) 26% (performed in CD71+ cells)</td>
<td></td>
</tr>
<tr>
<td></td>
<td>11</td>
<td>c.6154del (p.Ala2052fs) c.6154del (p.Ala2052fs)</td>
<td></td>
<td>2 year-old, severe in-utero anemia requiring five in-utero transfusions. Born with severe neonatal hyperbilirubinemia requiring exchange transfusion. Remains transfusion-dependent</td>
<td>Not evaluable in a transfused sample</td>
<td></td>
</tr>
</tbody>
</table>

Of note, all the SPTA1 variants reported here except c.4339-99C > T (αLEPRA) and c.2671C > T; p.R891* (Bogardus et al., 2014) have not been previously described.