SUPPLEMENTARY DATA

Table S1: Variant prioritization and filtering strategy used to analyze exome sequencing data.

<table>
<thead>
<tr>
<th>Total called variants</th>
<th>39,431</th>
</tr>
</thead>
<tbody>
<tr>
<td>Rare variants (&lt;1%)*</td>
<td>4170</td>
</tr>
<tr>
<td>Read number (&gt;20)</td>
<td>3300</td>
</tr>
<tr>
<td>Exonic and splicing variants</td>
<td>1189</td>
</tr>
<tr>
<td>Excluding synonymous variants</td>
<td>859</td>
</tr>
<tr>
<td>Homozygous/ Heterozygous variants (one or more) in genes known to cause human disease</td>
<td>217</td>
</tr>
<tr>
<td>Variant in concordance with observed phenotype</td>
<td>1</td>
</tr>
</tbody>
</table>

*Variant frequency equal to or less than 1% in population database gnomAD and our in-house data of 693 exomes

Web Resources:

1. OMIM, https://www.omim.org/