Supplementary Material

for

PSEUDODIASTROPHIC DYSPLASIA EXPANDS THE KNOWN PHENOTYPIC SPECTRUM
OF DEFECTS IN PROTEOGLYCAN BIOSYNTHESIS

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SUPPLEMENTARY FIGURE

Figure S1. Western blot analysis of recombinant B3GAT3 and CANT1. (A) Expression level of recombinant B3GAT3 in HEK293T cell lysates. (B) Expression level of recombinant CANT1 in conditioned media from HEK293T cells.
**SUPPLEMENTARY TABLE**

Table S1. Coverage statistics for the exome sequencing results

<table>
<thead>
<tr>
<th>Coverage statistics WES</th>
<th>1_Mother</th>
<th>1_Father</th>
<th>Patient 1A</th>
<th>Patient 1B</th>
<th>2_Mother</th>
<th>2_Father</th>
<th>Patient 2B</th>
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<tbody>
<tr>
<td>Average coverage (X)</td>
<td>109.9</td>
<td>84.9</td>
<td>65.8</td>
<td>69.7</td>
<td>147.6</td>
<td>148.3</td>
<td>124.2</td>
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<tr>
<td>% of targets covered &gt;20X</td>
<td>89.1</td>
<td>88.6</td>
<td>83.2</td>
<td>85.3</td>
<td>94.0</td>
<td>93.9</td>
<td>93.5</td>
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