Supplementary Material

for

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

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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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</thead>
<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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