### Table S1. Epidemiologic data of LHON diagnosis in the involved centers

<table>
<thead>
<tr>
<th></th>
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</thead>
<tbody>
<tr>
<td>Optic atrophy diagnosis*</td>
<td>371</td>
<td>1085</td>
<td>1140</td>
</tr>
<tr>
<td>LHON positive probands</td>
<td>104 (28%)</td>
<td>215 (19.81%)</td>
<td>280 (24.56%)</td>
</tr>
<tr>
<td>np11778 (%)</td>
<td>61.5</td>
<td>75.0</td>
<td>73.2</td>
</tr>
<tr>
<td>np3460 (%)</td>
<td>17.0</td>
<td>13.0</td>
<td>11.78</td>
</tr>
<tr>
<td>np14484 (%)</td>
<td>13.5</td>
<td>9.0</td>
<td>11.07</td>
</tr>
<tr>
<td>Number of complete mtDNA sequences</td>
<td>38</td>
<td>100</td>
<td>36</td>
</tr>
<tr>
<td>np14568 (%)</td>
<td>1.0</td>
<td>1.4</td>
<td>0.7</td>
</tr>
<tr>
<td>np14459 (%)</td>
<td>2.0</td>
<td>-</td>
<td>-</td>
</tr>
<tr>
<td>np14495 (%)</td>
<td>1.0</td>
<td>-</td>
<td>-</td>
</tr>
<tr>
<td>np14482 (%)</td>
<td>1.0</td>
<td>0.8</td>
<td>0.35</td>
</tr>
<tr>
<td>np3700 (%)</td>
<td>1.0</td>
<td>-</td>
<td>0.35</td>
</tr>
<tr>
<td>np3733 (%)</td>
<td>2.0</td>
<td>-</td>
<td>0.35</td>
</tr>
<tr>
<td>np4171 (%)</td>
<td>-</td>
<td>0.4</td>
<td>-</td>
</tr>
<tr>
<td>np10663 (%)</td>
<td>-</td>
<td>0.4</td>
<td>-</td>
</tr>
</tbody>
</table>

*Number of index patients tested for LHON mutations (does not include Dominant Optic Atrophy)