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Genomic and Clinical Assessment of Norrie Disease/ND

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**Genomic and Clinical Assessment of Norrie Disease/ND**

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**Abstract**

*Purpose:* Discuss the clinical features of Norrie Disease, assess the pattern of genetic inheritance, highlight the genetic diagnosis and indicate treatment. The NDP gene with clinical diagnostic techniques are outlined.

*Methods:* Relevant comprehensive search using specific search terms used to look up appropriate literature in different databases. The next step was to assess the papers for further examination and the closely related articles were included in the poster. Genomic investigation was also performed in order to find updated data associated with NDP gene and Norrie disease.

**Norrie Disease**

*Norrie Disease* is a rare X-linked recessive disorder that affects males from birth. The primary feature is congenital blindness.

**Alternative names**
- Atrophia bulborum hereditaria
- Episkopi blindness

*Norrie Disease Pseudoglioma Gene* located at Xp11.3

**NDP Gene Mutation Map**

**Clinical Diagnosis**

Diagnosis of ND includes a three generation pedigree, a thorough physical examination, B ultrasound scan, CT Brain and molecular genetic testing of NDP gene.

**Genetic Testing**

*Sequence analysis* of the entire coding region by GENETIX
- XLID NGS Panel by fulgent
- Viteroretinopathy & Wagner Syndrome NGS Panel and eye disorder panel by fulgent
- Norrie disease (sequence analysis of NDP gene) by CGC Genetics

*Deletion and Duplication Analysis*
- Detects deletions and duplications that are not detected by sequence analysis.
- Performed by qPCR, real-time PCR, MLPA, or array CGH.

**Gene Interaction**

<table>
<thead>
<tr>
<th>Gene</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>TSPAN12</td>
<td>Regulator of cell surface receptor signal transduction</td>
</tr>
<tr>
<td>LRP5</td>
<td>Involved in the Wnt/beta catenin signaling pathway</td>
</tr>
<tr>
<td>LGALS8</td>
<td>Lectin, galactoside-binding, soluble, 8</td>
</tr>
<tr>
<td>FZD4</td>
<td>Receptor for Wnt proteins</td>
</tr>
<tr>
<td>PPP1CA</td>
<td>Protein phosphatase 1 is essential for cell division</td>
</tr>
<tr>
<td>FZD8</td>
<td>Receptor for Wnt proteins</td>
</tr>
<tr>
<td>NDP</td>
<td>Activates the canonical Wnt signaling pathway</td>
</tr>
</tbody>
</table>

**References available upon request**

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**Treatment**

*Incomplete retinal detachment and/or intraocular pressure may be treated with laser surgery.*

*Hearing loss is treated with hearing aids or cochlear implants.*

*Treatment for behavioral issues and cognitive impairment involves supportive interventions.*

*Genetic counseling and prenatal diagnosis offered to carrier females considering pregnancy.*

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