

**DISEASE:**  
**Norrie disease**

<b>NAME:</b>	Norrie disease
<b>DESCRIPTION:</b>	A rare developmental defect during embryogenesis characterized by abnormal retinal development with congenital blindness. Common associated manifestations include sensorineural hearing loss and developmental delay, intellectual disability and/or behavioral disorders.
<b>ORPHACODE:</b>	649
<b>SYNOMYS:</b>	Atrophia bulborum hereditaria Episkopi blindness Norrie-Warburg disease
<b>XREF(S):</b>	<a href="#">Orphanet</a> <a href="#">ICD-10</a> <a href="#">MeSH</a> <a href="#">OMIM</a> <a href="#">MedDRA</a>
<b>ANALYTE(S):</b>	<a href="#">NDP</a>
<b>CREATED:</b>	13 May 2019 - 01:02
<b>CHANGED:</b>	22 Jun 2023 - 16:14

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## RELATED CONTENT

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### Related Genetic Tests

- Norrie disease (NDP gene)

### Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB

### Related Analytes

- norrin cystine knot growth factor NDP

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