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## **Genomic and Clinical Assessment of Norrie Disease**

### **The Purpose/Problem Statement**

Norrie Disease is a very rare x-linked recessive disorder that affects males from birth. The primary feature is congenital blindness. The purpose of this poster is to discuss the clinical features of Norrie Disease, assess the pattern of genetic inheritance with family history and pedigree data, highlight the genetic diagnosis via linkage analysis, deletion/duplication assay, and gene sequencing techniques, and indicate treatment of Norrie Disease. Mutation of the *NDP* gene, the subsequent disruption in the Norrin protein, the resulting phenotype in individuals affected by this mutation, as well as clinical diagnosis and treatment, are features of this presentation. The genetic component of Norrie Disease is a focus. Up to date clinical diagnostic techniques are outlined.

### **Methodology and Results**

Relevant comprehensive search using specific search terms such as “Norrie Disease”, “Clinical features of ND”, “Norrie Disease Pseudoglioma (*NDP*) Gene”, “*NDP* protein”, “PND diagnosis”, “ND clinical diagnosis”, “ND and linkage analysis”, “Xp11.4”, “gene ID 4693”, “ND treatment”, “ND prevention” used to look up appropriate literature. Different databases used to find the articles with broader search terms. The databases include PubMed, scholarly article search through Clemson library, OMIM, NCBI, Europe PMC, UCSC Genome Browser, Genetics Association Database, GeneCards, UniProt, EMBL-EBI, and Google scholar. The journal articles dated after 2009 were selected for more investigation (few seminal articles from older publications are also used). The next step was to read the abstracts and assess the papers for further examination. The results from each study closely related to the subject were included in the poster. Genomic database search were also performed in order to find updated data associated with *NDP* gene and Norrie disease.

### **Summary and Discussion**

ND is a rarely inherited condition that can be clinically diagnosed using a three-generational pedigree, a physical examination including a thorough ophthalmic examination, neurological examination, hearing tests, B ultrasound scan, Computed Tomography of Brain and molecular genetic testing of *NDP* gene. Genomic assessment includes sequence analysis of the entire coding region, deletion/duplication analysis, and linkage analysis. Genetic testing for *NDP* is available nationally at Emory Genetics Laboratory and Massachusetts General Hospital, and internationally in a few countries. Genetic counselling along with carrier testing for at-risk females, prenatal testing, and family planning are available when a disease-causing mutation has been identified in the family.

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