ABSTRACT

Thesis title: Study on the molecular characterization of Retinoschisis and Norrie disease in children from Northern Vietnam

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Inherited retinal disorders (IRDs) are a heterogeneous group of hereditary disorders that directly affect human retinal structure and function directly and often lead to progressive vision loss, in which X-linked retinoschisis and Norrie disease are especially notable because they mainly affect male children. X-linked retinoschisis (XLRS) is a monogenic disorder mainly caused by pathogenic variants in the *RS1* gene, resulting in the retinal layers splitting and affecting retinal structural integrity. While Norrie disease (ND) is another severe retinal disorder due to the mutations in the *NDP* gene, resulting in retinal detachment, blindness, and possible ocular complications. This study aims to identify the underlying genetic causes for pediatric patients from Northern Vietnam diagnosed with or suspected of having XLRS or ND by physicians at the National Eye Hospital from 2020 to 2025. The sequencing technology, such as whole-exome sequencing (WES) or Sanger sequencing, was employed to detect causative variants in patients, along with segregation analysis for the familial members of the patients. Totally, fourteen XLRS and two ND male patients were enrolled in this study, and they were successfully identified the pathogenic or likely pathogenic variants related to their conditions. Besides, thirteen females of patient's families were identified to be asymptomatic carriers. These findings not only showed the genetic etiologies of these two diseases in Vietnam but also broadened the mutational data of RS1 and NDP gene in the global. By introducing new variants to the national genetic database, the study enhances diagnostic ability, supports genetic counseling, a foundation for future research and potential gene-based therapies.

Keywords: X-linked retinoschisis, Norrie disease, *RS1* gene, *NDP* gene, WES, Vietnam.