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Inherited retinal degenerations: clinical characterization on the road to therapy

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STELLINGEN

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Inherited Retinal Degenerations: Clinical Characterization on the Road to Therapy

1. When including patients in an experimental therapy trial, other vision-affecting gene-specific parameters than photoreceptor degeneration, need to be taken into account (this thesis).
2. For female carriers of *RPGR* mutations, the term “carrier” may be misleading in many cases (this thesis).
3. Defining endpoints for clinical treatment trials, an important translational step towards therapy, is challenged by the inter-patient variability in disease expression and severity, despite the same monogenetic basis (this thesis).
4. *CRB1*-associated phenotypes are amenable to treatment in many patients (this thesis).
5. In experimental treatment trials for inherited retinal degenerations, the most viable control is as of yet the non-treated fellow eye (this thesis).
6. With the emergence of gene-based therapies, detecting the underlying genetic cause of a retinal dystrophy has become crucial and should be a continued pursuit.
7. Halting disease progression in inherited retinal degenerations should be feasible not only in early disease stages, but also in later disease stages.
8. A retrospective study design is not necessarily a limitation, as large retrospective studies in inherited retinal dystrophies can provide robust insights that cannot always be attained with a prospective design.
9. He who does not know his history, is not so much doomed to repeat it, as much as he is doomed to not know who he is (Arnon Grunberg, 2020, translated from Dutch). *Personal interpretation: understanding your past is essential to understanding your identity, and it gives you an indication of things to come and of how (not) to mold your future.*
10. A book is smarter than its author.