Probe compositions and relevant animal models used to evaluate the pathogenesis of retinitis pigmentosa and potentially new therapeutic modalities

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Technology description

Summary

MARKETS ADDRESSED:
Characterizing retinitis pigmentosa (RP) mutations has the potential to provide important insights relevant to genetic counseling and future applications to better characterize RP in conjunction with pipeline RP therapeutics. Furthermore, proprietary and investigative animal models at the Berman-Gund Laboratory may enable value-added drug validation and therapeutic targeting studies.
It is estimated that RP affects approximately 1 in 3,500 births in the United States. A U.S. disease prevalence of nearly 100,000 individuals is estimated. As a whole, continued growth in the genetic testing market is expected in conjunction with more targeted therapeutics and expansion in the prenatal screening market.
Retinitis pigmentosa is a devastating hereditary disease involving the gradual loss of vision due to deterioration of the retina's photoreceptor cells. Given the remarkable genetic heterogeneity of retinitis pigmentosa (RP), the characterization of genetic mutations underlying RP may provide valuable disease insights relevant to genetic testing, clinical care and future RP therapeutics.
Professor Berson and colleagues have discovered a series of RP mutations focused on the phototransduction cascade in rod photoreceptors. The patent estate covers most prevalent RP mutations including rhodopsin (dominant and recessive RP), the ?-subunit of retinal rod cGMP phosphodiesterase (recessive RP) and retinal degeneration slow protein (dominant RP, digenic RP and other retinal degenerations). The rhodopsin mutation is a major defect in RP, estimated to account for 11% of all RP cases.
In addition, animal models of photoreceptor degeneration available at the Berman-Gund Laboratory may be used to evaluate the pathogenesis of RP and potentially new therapeutic modalities. The Laboratory has created and/or investigated animal models broadly covering defects in phototransduction, the retinoid cycle, and outer segment maintenance.

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Advantages

-A series of genetic muta Advantages include: tions for characterizing RP are presented, including rhodopsin, which alone accounts for a significant percentage of RP (11%)
-The Berman-Gund Laboratory is one of the world's leading research centers on retinal degenerative disease. In conjunction with strong IP, commercialization efforts may leverage the lab's significant expertise in RP as well as a variety of proprietary research materials.

Institution

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