

## REFERENCES

1. den Hollander, A. I., Roepman, R., Koenekoop, R. K., & Cremers, F. P. *Leber congenital amaurosis: genes, proteins and disease mechanisms.* Prog Retin Eye Res, 2008. **27**(4): p. 391-419.
2. Sitorus, R.S., B. Lorenz, and M.N. Preising, *Analysis of three genes in Leber congenital amaurosis in Indonesian patients.* Vision Res, 2003. **43**(28): p. 3087-93.
3. Sitorus, R., M. Preising, and B. Lorenz, *Causes of blindness at the "Wiyata Guna" School for the Blind, Indonesia.* Br J Ophthalmol, 2003. **87**(9): p. 1065-8.
4. Siemiatkowska, Anna M., L. Ingeborgh van den Born, Maria M. van Genderen, Mette Bertelsen, Ditta Zobor, Klaus Rohrschneider, Ramon AC van Huet, et al. *Novel compound heterozygous NMNAT1 variants associated with Leber congenital amaurosis.* Mol Vis, 2014. **20**: p. 753-9.
5. Testa, F., Maguire, A. M., Rossi, S., Pierce, E. A., Melillo, P., Marshall, K, et al, *Three-year follow-up after unilateral subretinal delivery of adeno-associated virus in patients with Leber congenital Amaurosis type 2.* Ophthalmology, 2013. **120**(6): p. 1283-91.
6. Richard G Weleber, P.J.F., Karmen M Trzupek, and Catie Beattie., *Leber Congenital Amaurosis*, in *GeneReviews® [Internet]*. A.M. Pagon RA, Arlinger HH, et al, Editor. 2004 Jul 7 [Updated 2013 May 2], University of Washington: Seattle (WA).
7. Burnight, E. R., Wiley, L. A., Drack, A. V., Braun, T. A., Anfinson, K. R., Kaalberg, E. E. et al, *CEP290 gene transfer rescues Leber congenital amaurosis cellular phenotype.* Gene Ther, 2014. **8**(10): p. 39.
8. Collin, R. W., den Hollander, A. I., van der Velde-Visser, S. D., Bennicelli, J., Bennett, J., & Cremers, F. P., *Antisense Oligonucleotide (AON)-based Therapy for Leber Congenital Amaurosis Caused by a Frequent Mutation in CEP290.* Mol Ther Nucleic Acids, 2012. **27**(1): p. 3.
9. Maguire, A. M., Simonelli, F., Pierce, E. A., Pugh Jr, E. N., Mingozi, F., Bennicelli, J, et al., *Safety and efficacy of gene transfer for Leber's congenital amaurosis.* N Engl J Med, 2008. **358**(21): p. 2240-8.
10. den Hollander, A. I., Black, A., Bennett, J., & Cremers, F. P., *Lighting a candle in the dark: advances in genetics and gene therapy of recessive retinal dystrophies.* J Clin Invest, 2010. **120**(9): p. 3042-53.
11. Leber, T., *Ueber Retinitis pigmentosa und angeborene Amaurose.* Archiv für Ophthalmologie, 1869. **15**(3): p. 1-25.
12. Pinckers, A.J., *Leber's congenital amaurosis as conceived by Leber.* Ophthalmologica, 1979. **179**(1): p. 48-51.

13. Koenekoop, R.K., *An overview of Leber congenital amaurosis: a model to understand human retinal development*. Surv Ophthalmol, 2004. **49**(4): p. 379-98.
14. Stone, E.M., *Leber congenital amaurosis - a model for efficient genetic testing of heterogeneous disorders: LXIV Edward Jackson Memorial Lecture*. Am J Ophthalmol. 2007 Dec;144(6):791-811. Epub 2007 Oct 26.
15. Franceschetti, A. and P. Dieterle, *[Diagnostic and prognostic importance of the electroretinogram in tapetoretinal degeneration with reduction of the visual field and hemeralopia]*. Confin Neurol, 1954. **14**(2-3): p. 184-6.
16. Cremers, F.P.M., J.A.J.M. van den Hurk, and A.I. den Hollander, *Molecular genetics of Leber congenital amaurosis*. Human Molecular Genetics, 2002. **11**(10): p. 1169-1176.
17. Foxman, S. G., Heckenlively, J. R., Bateman, J. B., & Wirtschafter, J. D., *Classification of congenital and early onset retinitis pigmentosa*. Arch Ophthalmol, 1985. **103**(10): p. 1502-6.
18. Heher, K.L., E.I. Traboulsi, and I.H. Maumenee, *The Natural History of Leber's Congenital Amoaurosis: Age-related Findings in 35 Patients*. Ophthalmology, 1992. **99**(2): p. 241-245.
19. Fulton, A.B., R.M. Hansen, and D. Mayer, *VIision in leber congenital amaurosis*. Archives of Ophthalmology, 1996. **114**(6): p. 698-703.
20. Breclj, J. and B. Stirn-Kranjc, *ERG and VEP follow-up study in children with Leber's congenital amaurosis*. Eye, 1999. **13**(Pt 1): p. 47-54.
21. Koenekoop, R. K., Lopez, I., Den Hollander, A. I., Allikmets, R., & Cremers, F. P., *Genetic testing for retinal dystrophies and dysfunctions: benefits, dilemmas and solutions*. Clin Experiment Ophthalmol, 2007. **35**(5): p. 473-85.
22. Hufnagel, R. B., Ahmed, Z. M., Corrêa, Z. M., & Sisk, R. A., *Gene therapy for Leber congenital amaurosis: advances and future directions*. Graefes Arch Clin Exp Ophthalmol, 2012. **250**(8): p. 1117-28.
23. Wang, H., Chen, X., Dudinsky, L., Patenia, C., Chen, Y., Li, Y., et al., *Exome capture sequencing identifies a novel mutation in BBS4*. Mol Vis, 2011. **17**: p. 3529-40.
24. Estrada-Cuzcano, A., Roepman, R., Cremers, F. P., den Hollander, A. I., & Mans, D. A., *Non-syndromic retinal ciliopathies: translating gene discovery into therapy*. Human Molecular Genetics, 2012. **21**(R1): p. R111-R124.
25. Saudou, F., *A "so cilia" network: cilia proteins start "social" networking*. J Clin Invest, 2012. **122**(4): p. 1198-202.
26. Coppieters, F., Lefever, S., Leroy, B. P., & De Baere, E., *CEP290, a gene with many faces: mutation overview and presentation of CEP290base*. Human Mutation, 2010. **31**(10): p. 1097-1108.

27. Revere, K.E. and D.C. Chung, *Recent breakthroughs in gene therapy for inherited retinal degeneration*. Discov Med, 2009. **8**(42): p. 125-9.
28. Hammond, S.M. and M.J.A. Wood, *Genetic therapies for RNA mis-splicing diseases*. Trends in Genetics, 2011. **27**(5): p. 196-205.
29. Rowe-Rendleman, C. L., Durazo, S. A., Kompella, U. B., Rittenhouse, K. D., Di Polo, A., Weiner, A. L., et al., *Drug and gene delivery to the back of the eye: from bench to bedside*. Invest Ophthalmol Vis Sci, 2014. **55**(4): p. 2714-30.
30. Simonelli, F., Maguire, A. M., Testa, F., Pierce, E. A., Mingozzi, F., Bennicelli, J. L., et al., *Gene therapy for Leber's congenital amaurosis is safe and effective through 1.5 years after vector administration*. Mol Ther, 2010. **18**(3): p. 643-50.
31. Trapani, I., A. Puppo, and A. Auricchio, *Vector platforms for gene therapy of inherited retinopathies*. Progress in Retinal and Eye Research, 2014. **43**(0): p. 108-128.
32. Bartholomae, C. C., Arens, A., Balaggan, K. S., Yáñez-Muñoz, R. J., Montini, E., Howe, S. J., et al., *Lentiviral vector integration profiles differ in rodent postmitotic tissues*. Mol Ther, 2011. **19**(4): p. 703-10.
33. Balaggan, K. S., Binley, K., Esapa, M., Iqball, S., Askham, Z., Kan, O., et al., *Stable and efficient intraocular gene transfer using pseudotyped EIAV lentiviral vectors*. The Journal of Gene Medicine, 2006. **8**(3): p. 275-285.
34. Surace, E.M. and A. Auricchio, *Adeno-associated viral vectors for retinal gene transfer*. Prog Retin Eye Res, 2003. **22**(6): p. 705-19.
35. Surace, E.M. and A. Auricchio, *Versatility of AAV vectors for retinal gene transfer*. Vision Res, 2008. **48**(3): p. 353-9.
36. Duan, D., Sharma, P., Yang, J., Yue, Y., Dudus, L., Zhang, Y., et al., *Circular intermediates of recombinant adeno-associated virus have defined structural characteristics responsible for long-term episomal persistence in muscle tissue*. J Virol, 1998. **72**(11): p. 8568-77.
37. Read, S.P., S.M. Cashman, and R. Kumar-Singh, *A poly(ethylene) glycolylated peptide for ocular delivery compacts DNA into nanoparticles for gene delivery to post-mitotic tissues in vivo*. J Gene Med, 2010. **12**(1): p. 86-96.
38. Han, Z., Conley, S. M., Makkia, R., Guo, J., Cooper, M. J., & Naash, M. I, *Comparative analysis of DNA nanoparticles and AAVs for ocular gene delivery*. PLoS One, 2012. **7**(12): p. 18.
39. Veltrop, M. and A. Aartsma-Rus, *Antisense-mediated exon skipping: Taking advantage of a trick from Mother Nature to treat rare genetic diseases*. Experimental Cell Research, 2014. **325**(1): p. 50-55.

40. Dias, N. and C.A. Stein, *Antisense oligonucleotides: basic concepts and mechanisms*. Mol Cancer Ther, 2002. **1**(5): p. 347-55.
41. Urban, E. and C.R. Noe, *Structural modifications of antisense oligonucleotides*. II Farmaco, 2003. **58**(3): p. 243-258.
42. Miller, S.A., D.D. Dykes, and H.F. Polesky, *A simple salting out procedure for extracting DNA from human nucleated cells*. Nucleic Acids Research, 1988. **16**(3): p. 1215.
43. Don, R. H., Cox, P. T., Wainwright, B. J., Baker, K., & Mattick, J. S., et al., *'Touchdown' PCR to circumvent spurious priming during gene amplification*. Nucleic Acids Research, 1991. **19**(14): p. 4008.
44. Siemiatkowska, A. M., Arimadyo, K., Moruz, L. M., Astuti, G. D., de Castro-Miro, M., Zonneveld, M. N., et al., *Molecular genetic analysis of retinitis pigmentosa in Indonesia using genome-wide homozygosity mapping*. Mol Vis, 2011. **17**: p. 3013-24.
45. Sohocki, M. M., Perrault, I., Leroy, B. P., Payne, A. M., Dharmaraj, S., Bhattacharya, S. S., et al., *Prevalence of AIPL1 mutations in inherited retinal degenerative disease*. Mol Genet Metab, 2000. **70**(2): p. 142-50.
46. Akey, D. T., Zhu, X., Dyer, M., Li, A., Sorensen, A., Blackshaw, S., et al., *The inherited blindness associated protein AIPL1 interacts with the cell cycle regulator protein NUB1*. Human Molecular Genetics, 2002. **11**(22): p. 2723-2733.
47. Ramamurthy, V., Niemi, G. A., Reh, T. A., & Hurley, J. B., *Leber congenital amaurosis linked to AIPL1: a mouse model reveals destabilization of cGMP phosphodiesterase*. Proc Natl Acad Sci U S A, 2004. **101**(38): p. 13897-902.
48. den Hollander, A. I., Koenekoop, R. K., Yzer, S., Lopez, I., Arends, M. L., Voesenek, K. E., et al., *Mutations in the CEP290 (NPHP6) gene are a frequent cause of Leber congenital amaurosis*. Am J Hum Genet, 2006. **79**(3): p. 556-61.
49. Kinali, M., Arechavala-Gomeza, V., Feng, L., Cirak, S., Hunt, D., Adkin, C., et al., *Local restoration of dystrophin expression with the morpholino oligomer AVI-4658 in Duchenne muscular dystrophy: a single-blind, placebo-controlled, dose-escalation, proof-of-concept study*. Lancet Neurol, 2009. **8**(10): p. 918-28.
50. Van Deutkom, J. C., Janson, A. A., Ginjaar, I. B., Frankhuizen, W. S., Aartsma-Rus, A., Bremmer-Bout, M., et al., *Local dystrophin restoration with antisense oligonucleotide PRO051*. N Engl J Med, 2007. **357**(26): p. 2677-86.
51. Hammond, S.M. and M.J. Wood, *Genetic therapies for RNA mis-splicing diseases*. Trends Genet, 2011. **27**(5): p. 196-205.
52. Phillips, M.I. and Y.C. Zhang, *Basic principles of using antisense oligonucleotides in vivo*. Methods Enzymol, 2000. **313**: p. 46-56.

53. Garanto, A., van Beersum, S. E., Peters, T. A., Roepman, R., Cremers, F. P., & Collin, R. W. (2013). Unexpected CEP290 mRNA splicing in a humanized knock-in mouse model for Leber congenital amaurosis. *PLoS one*, 8(11), e79369.
54. Plotnikova, O.V., E.N. Pugacheva, and E.A. Golemis, *Primary cilia and the cell cycle*. Methods in cell biology, 2009. **94**: p. 137-160.
55. Tucker, B. A., Mullins, R. F., Streb, L. M., Anfinson, K., Eyestone, M. E., Kaalberg, E., et al., *Patient-specific iPSC-derived photoreceptor precursor cells as a means to investigate retinitis pigmentosa*. eLife, 2013. **2**: p. e00824.
56. MacLaren, R. E., Groppe, M., Barnard, A. R., Cottrall, C. L., Tolmachova, T., Seymour, L., et al., *Retinal gene therapy in patients with choroideremia: initial findings from a phase 1/2 clinical trial*. Lancet, 2014. **383**(9923): p. 1129-37.
57. Cideciyan, A. V., Aleman, T. S., Jacobson, S. G., Khanna, H., Sumaroka, A., Aguirre, G. K., et al., *Centrosomal-ciliary gene CEP290/NPHP6 mutations result in blindness with unexpected sparing of photoreceptors and visual brain: implications for therapy of Leber congenital amaurosis*. Hum Mutat, 2007. **28**(11): p. 1074-83.
58. Bainbridge, J.W., M.H. Tan, and R.R. Ali, *Gene therapy progress and prospects: the eye*. Gene Ther, 2006. **13**(16): p. 1191-7.
59. Maguire, A. M., High, K. A., Auricchio, A., Wright, J. F., Pierce, E. A., Testa, F., et al., *Age-dependent effects of RPE65 gene therapy for Leber's congenital amaurosis: a phase 1 dose-escalation trial*. Lancet, 2009. **374**(9701): p. 1597-605.
60. Ashtari, M., Cyckowski, L. L., Monroe, J. F., Marshall, K. A., Chung, D. C., Auricchio, A., et al., *The human visual cortex responds to gene therapy-mediated recovery of retinal function*. J Clin Invest, 2011. **121**(6): p. 2160-8.
61. Bennett, J., Ashtari, M., Wellman, J., Marshall, K. A., Cyckowski, L. L., Chung, D. C., et al., *AAV2 gene therapy readministration in three adults with congenital blindness*. Sci Transl Med, 2012. **4**(120): p. 3002865.
62. Vasireddy, V., Mills, J. A., Gaddameedi, R., Basner-Tschakarjan, E., Kohnke, M., Black, A. D., et al., *AAV-mediated gene therapy for choroideremia: preclinical studies in personalized models*. PLoS One, 2013. **8**(5).
63. Baye, L. M., Patrinostro, X., Swaminathan, S., Beck, J. S., Zhang, Y., Stone, E. M., et al., *The N-terminal region of centrosomal protein 290 (CEP290) restores vision in a zebrafish model of human blindness*. Hum Mol Genet, 2011. **20**(8): p. 1467-77.
64. Gerard, X., Perrault, I., Hanein, S., Silva, E., Bigot, K., Defoort-Delhemmes, S., et al., *AON-mediated Exon Skipping Restores Ciliation in Fibroblasts Harboring the Common Leber Congenital Amaurosis CEP290 Mutation*. Mol Ther Nucleic Acids, 2012. **1**: p. e29.