

## BIOGRAPHICAL SKETCH

**Name:**  
den Hollander, Anneke I.

**Position and title:**  
Workgroupleader Multifactorial Eye Diseases; Associate professor

**Education & training:**

Institution and Location	Degree	Year	Field of Study
Leiden University Department of Human Genetics, Radboud University Nijmegen	M.Sc. Ph.D.	1991 1996	Biomed. Sciences Molecular Genetics

### A. Positions and Honors.

#### Positions and Employment

1996-2001	PhD student, Department of Human Genetics, Radboud University Nijmegen Medical Centre
2001-2002	Postdoctoral researcher, Department of Human Genetics, Radboud University Nijmegen Medical Centre
2002	Guest researcher, Téléthon Institute of Genetics and Medicine (TIGEM), Naples, Italy
2002-2007	Senior research scientist, Department of Human Genetics, Radboud University Nijmegen Medical Centre
2007-2008	Research fellow, The Berman-Gund Laboratory, Massachusetts Eye and Ear Infirmary, Harvard Medical School, Boston, MA, USA
2008-2009	Assistant professor, Department of Ophthalmology, Radboud University Nijmegen Medical Centre
2009-current	Associate professor, Department of Ophthalmology, Radboud University Nijmegen Medical Centre

#### Other Experience and Professional Memberships

2002-2004	Establishment of a postdoc platform within the Nijmegen Center for Molecular Life Sciences.
2004-2005	Member of the Interaction Committee, Genomics Network for Young Scientists (GeNeYouS).
2006-2008	Member of the Medical Advisory Committee, Retina Netherlands
2007-present	Aspiring Principal Investigator, Nijmegen Centre for Molecular Life Sciences
2008-present	Junior Principal Investigator, Radboud University Nijmegen Medical Centre
2009-2012	Annual Meeting Program Committee, The Association for Research in Vision and Ophthalmology (ARVO).

#### Honors and Awards

- Award for the best 3<sup>rd</sup>-year traineeship, Biomedical Sciences, University of Leiden, 1995
- NCMLS award for the best thesis of the year, Nijmegen Center for Molecular Life Sciences, 2002
- PhD degree with the highest honor, 2002
- TALENT stipend, Netherlands Organisation for Scientific Research (NWO), 2002
- Ter Meulen Fund stipend, Royal Dutch Academy of Arts and Sciences (KNAW), 2007
- Aspasia Award, Netherlands Organisation for Scientific Research (NWO), 2009

### B. Selected peer-reviewed publications (in alphabetic order).

(Ten from 52 peer-reviewed publications; as of October 2009; with emphasis on papers published in last 5 yrs)

Boon CJF, Klevering BJ, LeRoy BP, Hoyng CB, Keunen JEE & **den Hollander AI** (2009). The spectrum of ocular phenotypes caused by mutations in the BEST1 gene. *Prog Retin Eye Res*, **in press**.

Boon CJF, Klevering BJ, Hoyng CB, Zonneveld MN, Nabuurs SB, Blokland E, Cremers FPM & **den Hollander AI**. (2008) Basal laminar drusen caused by compound heterozygous variants in the CFH gene. *Am J Hum Genet* **82**, 516-523.

Collin RWJ, Littink KW, Klevering BJ, van den Born LI, Koenekoop RK, Zonneveld MN, Blokland EAW, Strom TM, Hoyng CB, den **Hollander AI** & Cremers FPM (2008). Identification of a 2-megabases human orthologue of Drosophila eyes shut/spacemaker that is mutated in patients with retinitis pigmentosa. *Am J Hum Genet* **83**, 594-603.

**den Hollander AI**, Roepman R, Koenekoop RK & Cremers FPM (2008). Leber congenital amaurosis: genes, proteins and disease mechanisms. *Prog Retin Eye Res*, **27**, 391-419.

**den Hollander AI**, Koenekoop RK, Mohamed MD, Arts HH, Boldt K, Towns KV, Sedmak T, Beer M, Nagel-Wolfrum K, McKibbin M, Dharmaraj S, Lopez I, Ivings L, Williams GA, Springell K, Woods CG, Jafri H, Rashid Y, Strom TM, van der Zwaag B, Gosens I, Kersten FFJ, van Wijk E, Veltman JA, Zonneveld MN, van Beersum SEC, Maumenee IH, Wolfrum U, Cheetham ME, Ueffing M, Cremers FPM, Inglehearn CF & Roepman R (2007). Mutations in LCA5, encoding the ciliary protein lebercillin, cause Leber congenital amaurosis. *Nature Genet* **39**, 889-895.

**den Hollander AI**, Koenekoop RK, Yzer S, Lopez I, Arends ML, Voesenek KEJ, Zonneveld MN, Strom TM, Meitinger T, Brunner HG, Hoyng CB, van den Born LI, Rohrschneider K & Cremers FPM (2006). Mutations in the CEP290 (NPHP6) gene are a frequent cause of Leber congenital amaurosis. *Am J Hum Genet* **79**, 556-561.

**den Hollander AI**, Heckenlively JR, van den Born LI, de Kok YJM, van der Velde-Visser SD, Kellner U, Jurklies B, van Schooneveld M, Blankenagel A, Rohrschneider K, Wissinger B, Cruysberg JR, Deutman AF, Brunner HG, Apfelstedt-Sylla E, Hoyng CB & Cremers FPM (2001). Leber congenital amaurosis and retinitis pigmentosa with Coats-like exudative vasculopathy are associated with mutations in the crumbs homolog 1 (CRB1) gene. *Am J Hum Genet* **69**, 198-203.

**den Hollander AI**, ten Brink JB, de Kok YJM, van Soest S, van den Born LI, van Driel MA, van de Pol DJR, Payne AM, Bhattacharya SS, Kellner U, Hoyng CB, Westerveld A, Brunner HG, Bleeker-Wagemakers EM, Deutman AF, Heckenlively JR, Cremers FPM & Bergen AAB (1999). Mutations in a human homologue of *Drosophila* crumbs cause retinitis pigmentosa (RP12). *Nature Genet* **23**, 217-221.

Thiadens AA, **den Hollander AI**, Roosing S, Nabuurs SB, Zekveld-Vroom RC, Collin RW, De Baere E, Koenekoop RK, van Schooneveld MJ, Strom TM, van Lith-Verhoeven JJ, Lotery AJ, van Moll-Ramirez N, LeRoy BP, van den Born LI, Hoyng CB, Cremers FPM, Klaver CC (2009). Homozygosity mapping reveals PDE6C mutations in patients with early-onset cone photoreceptor disorders. *Am J Hum Genet* **85**, 240-247.

Wang H, **den Hollander AI**, Moayedi Y, Abulimiti A, Li Y, Collin RW, Hoyng CB, Lopez I, Bray M, Lewis RA, Lupski JR, Mardon G, Koenekoop RK & Chen R (2009). SPATA7 is an early onset retinal disease gene. *Am J Hum Genet* **84**, 380-387.

## C. Ongoing research Support

VIDI Innovative Research Award, Netherlands Organisation for Scientific Research (NWO). Elucidation of the molecular basis of basal laminar drusen and its predisposing role in age-related macular degeneration. 2008-2013. Role: PI.

Stichting Blindenhulp. Elucidation of the molecular basis of basal laminar drusen and its predisposing role in age-related macular degeneration. 2008-2009. Role: PI.

Junioronderzoekerronde 2008, Radboud University Nijmegen Medical Centre. A new approach in human genetics: homozygosity mapping in outbred populations. 2008-2012. Role: Co-investigator.

Foundation for Retinal Research, USA. LCA5 (lebercillin): molecular genetic, functional, mouse knock-out, and gene rescue studies. 2008-2012. Role: Co-investigator.

Individual Investigator Award, Foundation Fighting Blindness, USA. Identification of new genes for Leber congenital amaurosis and retinitis pigmentosa using a genome-wide approach. 2007-2010. Role: PI.

Stichting Wetenschappelijk Onderzoek Oogziekenhuis, Rotterdam. Aangeboren blindheid, retinitis pigmentosa en kegelsstaaf dystrofie: Identificatie van moleculaire oorzaken en genotype-fenotype correlatie. 2006-2009. Role: Co-investigator.

## Completed Research Support (2003 – 2008)

Individual Investigator Award, Foundation Fighting Blindness, USA. Ophthalmogenomics: a new genome-wide approach for inherited blindness. 2006. Role: PI.

VENI Innovative Research Award, Netherlands Organisation for Scientific Research (NWO). Ophthalmogenomics: a new genome-wide approach for inherited blindness. 2005-2008. Role: PI.

Stichting Wetenschappelijk Onderzoek Het Oogziekenhuis, Rotterdam. Identification of novel molecular causes of LCA and retinitis pigmentosa through genome-wide single nucleotide polymorphism analysis. 2004. Role: Co-investigator.

British Retinitis Pigmentosa Society. Identification of three genes involved in autosomal dominant retinal dystrophies. 2002-2005. Role: Co-investigator.

Foundation Fighting Blindness, USA. Isolation of novel retina and retinal pigment epithelium specific cDNAs through subtraction hybridization-PCR: Extended characterization of retina-specific genes and candidate gene analysis of selected chorioretinal diseases. 2002-2005. Role: Co-investigator.

Foundation Fighting Blindness, USA. Elucidation of the molecular processes underlying a severe form of autosomal recessive retinitis pigmentosa (RP12). 2000-2003. Role: Co-investigator.