BIOGRAPHICAL SKETCH

NAME
Ronald Roepman, Ph.D.

POSITION
Associate Professor, Head Molecular Biology of Ciliopathies research group (10 scientists), Department of Human Genetics and Nijmegen Centre for Molecular Life Sciences, Radboud University Nijmegen Medical Centre, Nijmegen, The Netherlands.

EDUCATION/TRAINING

Institution and location	Degree	Year(s)	Field of study
Wageningen University	M.Sc.	1993	Cellular and Molecular Biology
Radboud University Nijmegen	PhD	2000	Medical Science/Genetics

A. POSITIONS AND HONORS.

Positions and Employment

- *1993–1994* Research fellow, Dutch Red Cross Blood Transfusion Service (CLB), Amsterdam.
- *1994-2000* PhD student, Department of Human Genetics, Radboud University Nijmegen Medical Centre / Nijmegen Centre for Molecular Life Sciences (RUNMC/NCMLS).
- *1997–1998* Research fellow (KNAW Ter Meulen Fonds exchange fellowship); Department of Toxicology and Pharmacology, Medical College of Wisconsin, Milwaukee, USA.
- *2000–2002* Post-doctoral fellow, Department of Human Genetics, (RUNMC/NCMLS).
- *2002–2007* Assistant professor (5-year tenure track), Head Functional Genomics of the Retina research group, Department of Human Genetics, (RUNMC/NCMLS).
- *2005–2006* Pro Retina Germany Research Fellowship; GSF Institute of Human Genetics, Neuherberg/Munich, Germany.
- *2007-2009* Assistant professor, Head Functional Genomics of the Retina research group, Department of Human Genetics, (RUNMC/NCMLS).
- *2010-present* Associate professor (tenured position), Department of Human Genetics, (RUNMC/NCMLS).

Honors and awards

- Royal Netherlands Academy of Arts and Sciences (KNAW) (1997), Ter Meulen Fonds exchange fellowship.
- Fight For Sight/Fighting Blindness America (1997), student research fellowship.
- Concerted Action of the European Communities 'Prevention of Blindness' (1999), Molecular and Clinical Research in Photoreceptor Disorders: short-time exchange fellowship.
- Pro Retina Germany (2005), research fellowship.
- Netherlands Organisation for Scientific Research (NWO) (2007), 'Vidi' award.
- Principal Investigator, Radboud University Nijmegen Medical Centre and Nijmegen Centre for Molecular Life Sciences, 2008.

Other academic activities

- Reviewer for Nat. Genet., Am. J. Hum. Genet., J. Clin. Invest., Proc. Natl. Acad. Sci. USA, Hum. Mol. Genet., Hum. Mutat., Clin. Genet., Invest. Ophthalmol. Vis. Sci., Mol. Vis., Exp. Eye Res., Ped. Nephrol., J. Neurosci., J. Neurochem., Eur. J. Cell Biol.; international funding agencies.
- Supervisor of 3 Ph.D. Thesises, of which 2 cum laude (top 5%).
- Member of Association for Research in Vision and Ophthalmology (1998-2010)
- Member of American Society of Human Genetics (1995-2010)

B. SELECTED PEER-REVIEWED PUBLICATIONS (in chronological order).

1. **Roepman, R.**, [8 co-authors], and Berger W. (1996). Identification of a gene disrupted by a microdeletion in a patient with X-linked retinitis pigmentosa. *Hum. Mol. Genet.*, 5, 827-833.
2. **Roepman, R.**, [8 co-authors], and Berger, W. (1996). Positional cloning of the gene for X-linked retinitis pigmentosa: homology with the guanine-nucleotide-exchange factor RCC1. *Hum. Mol. Genet.*, 5, 1035-1041.
3. **Roepman, R.**, [6 co-authors], and Ferreira, P.A. (2000). The retinitis pigmentosa GTPase regulator (RPGR) interacts with novel transport-like proteins in the outer segments of rod photoreceptors. *Hum. Mol. Genet.*, 9, 2095-2105.

4. **Roepman, R.**, [6 co-authors], and Cremers, F.P.M. (2005). Interaction of nephrocystin-4 and RPGRIP1 is disrupted by nephronophthisis or Leber congenital amaurosis-associated mutations. *Proc. Natl. Acad. Sci. USA*, **102**, 18520-18525.
5. van Wijk, E., [11 co-authors], **Roepman, R.***, and Kremer, H.* (2006). The DFNB31 gene product whirlin connects to the Usher protein network in cochlea and retina by direct association with USH2A and VLGR1. *Hum. Mol. Genet.*, 15, 751-765.**equal authors.*
6. Kremer, H., van Wijk, E., Märker, T., Wolfrum, U., and **Roepman, R.** (2006). Usher syndrome: molecular links of pathogenesis, proteins and pathways. *Hum. Mol. Genet.*, 15, R262-R270.
7. Gosens, I, [13 co-authors], and **Roepman, R.** (2007). MPP1 links the Usher protein network and the Crumbs protein complex in the retina. *Hum. Mol. Genet.*, 16, 1993-2003.
8. den Hollander, A.I.*, Koenekoop, R.K.*, Mohamed, M.D.*, Arts, H.H.*, [26 co-authors], Cremers, F.P.M.*, Inglehearn, C.F.*, and **Roepman, R.*** (2007). Mutations in LCA5, encoding the novel ciliary protein lebercilin, cause Leber congenital amaurosis. *Nat. Genet.*, 39, 889-895. *equal authors
9. Arts, H.H., [17 co-authors], and **Roepman, R.** (2007). Mutations in the gene encoding the basal body protein RPGRIP1L, a nephrocystin-4 interactor, cause Joubert syndrome. *Nat. Genet.*, 39, 882-888.
10. Gosens, I., den Hollander, A.I., Cremers, F.P.M., and **Roepman, R.** (2008). Composition and function of the Crumbs protein complex in the mammalian retina. *Exp. Eye Res.*, 86, 713-726.
11. den Hollander, A.I., **Roepman, R.,** Koenekoop, R.K., Cremers, and F.P.M. (2008). Leber congenital amaurosis: genes, proteins and disease mechanisms. *Prog. Retin. Eye Res.*, 27, 391-419.
12. Gorden, N.*, Arts, H.H.*#, [29 co-authors], **Roepman, R.**, Moens, C.B., Glass, I.A., and Doherty, D. (2008). CC2D2A is mutated in Joubert syndrome and interacts with the ciliopathy-associated basal body protein CEP290. *Am. J. Hum. Genet.*, 83, 559-571.*Equal 1st authors; #Roepman group.
13. van Wijk, [11 co-authors], **Roepman, R.***, and Kremer, H.* (2009). Usher syndrome and Leber congenital amaurosis are molecularly linked via a novel isoform of the centrosomal ninein-like protein. *Hum. Mol. Genet.*, 18, 51-64 *equal authors
14. Coene, K.L.M.*, **Roepman, R.***, [15 co-authors], and de Brouwer, A.P.M. (2009). OFD1 is mutated in X-linked Joubert syndrome and interacts with LCA5-encoded lebercilin. *Am. J. Hum. Genet.* 85, 465-481. *Equal authors.
15. Kersten, F., [8 co-authors], **Roepman, R.*** and Kremer, H.* (2010) Whirlin associates with the Cav1.3 ({alpha}1D) channels in photoreceptors, defining a novel member of the Usher protein network. *Invest Ophthalmol Vis Sci.,* 51, 2338-2346. *equal authors

C. RESEARCH SUPPORT.

Ongoing Research Grants (total € 2,847K to Roepman)

1. *Radboud University Nijmegen Medical Centre* (2006–2010) “Elucidation of the pathogenetic mechanisms in Usher syndrome: new tools for diagnostics and therapy” (PIs: Kremer, Keunen and Roepman).
2. *Netherlands Organisation for Scientific Research (NWO)* (2007-2012) “Cilia (mal)function in the retinal system”.
3. *Foundation for Retinal Research (FRR), USA* (2007-2012) “LCA5 (lebercilin): molecular genetic, functional, mouse KO, and gene rescue studies” (PIs: Bennett, den Hollander, Koenekoop, Nishina, Cremers and Roepman).
4. *Foundation Fighting Blindness (FFB), USA* (2008-2011); “Unraveling the molecular disease mechanism of LCA5 and associated ciliary blinding disorders”.
5. *EC-FP7, HEALTH-2009-2.1.2-1 (2010-2015)*: “SYSCILIA - A systems biology approach to dissect cilia function and its disruption in human genetic disease” (18 PIs from 16 institutions; €11M total budget; coordinator: R. Roepman).

Finished Research Grants (total € 834K to Roepman)

1. *EC-FP6 (2005–2009)* “Functional Genomics of the Retina in Health and Disease (EVI-GenoRet)”.
2. *Dutch Blindness Foundations* (2006–2007) “Analysis of the RPGR/RPGRIP1 protein complex in the 'connecting cilia' of photoreceptors – from function to therapy”.
3. *Pro Retina Germany* (2006–2007) “Proteomic analysis of RPGR/RPGRIP1-associated protein complexes – generating novel handles for therapeutic approaches” (PIs: Ueffing and Roepman).
4. *British Retinitis Pigmentosa Society* (2006-2009) “Identification of the pathogenic mechanisms in Usher syndrome: new handles for therapy and diagnostics” (PIs: Kremer and Roepman).
5. *Dutch Kidney foundation (2005–2009)* “Identification and characterization of novel proteins in the kidney cilia that are associated with nephronophthisis and allied syndromes” (PIs: Knoers and Roepman).