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EDUCATION

2007-2010, expected date of thesis defence: November 2010

PhD, Ophthalmogenetics
The Rotterdam Eye Hospital, Rotterdam, the Netherlands
Dept. of Human Genetics, Radboud University Nijmegen Medical Centre, Nijmegen, the Netherlands

Dissertation: The power of homozygosity mapping: discovery of new genetic defects in patients with retinal dystrophy

Promotor:
Frans P.M. Cremers, PhD

Copromotores:
L. Ingeborgh van den Born, MD, PhD, Anneke I. den Hollander, PhD

September 2006-January 2007

Master research internship
McGill Ocular Genetics Centre, McGill University Health Centre, Montreal, Canada

Thesis: Phenotypic and molecular genetic analysis of three families with inherited retinal dystrophies

Supervisors:
Robert K. Koenekoop, MD, PhD; Frans P.M. Cremers, PhD; Anneke I. den Hollander, PhD

1999-2007

M.D., Medicine
Radboud University Nijmegen, Nijmegen, The Netherlands

PUBLICATIONS

Collin RWJ[*], Safieh C[*], **Littink KW**, Shalev SA, Garzozi HJ, Rizel L, Cremers FPM, den Hollander AI, Klevering BJ[#], Ben-Yosef T[#]. (2010)
Mutations in C2ORF71 cause autosomal recessive retinitis pigmentosa.
Am J Hum Genet, *in press*

Bandah-Rozenfeld D, **Littink KW**, Ben-Yosef T, Strom TM, Chowers I, Collin RWJ, den Hollander AI, van den Born LI, Zonneveld MN, Merin S, Banin E, Cremers FPM, Sharon D. (2010)
Novel null mutations in the EYS gene are a frequent cause of autosomal recessive retinitis pigmentosa in the Israeli population.
Invest Ophthalmol Vis Sci, *in press*

Littink KW, van den Born LI, Koenekoop RK, Collin RWJ, Zonneveld MN, Blokland EAW, Khan H, Theelen T, Hoyng CB, Cremers FPM, den Hollander AI, Klevering BJ. (2010)
Mutations in the EYS gene account for ~5% of autosomal recessive retinitis pigmentosa and cause a fairly homogeneous phenotype.
Opthalmology*, in press*

Littink KW, Pott JWR, Collin RWJ, Kroes HY, Verheij JBGM, Blokland EAW, de Castro Miró M, Hoyng CB, Klaver CCW, Koenekoop RK, Rohrschneider K, Cremers FPM, van den Born LI, den Hollander AI. (2010)
A novel nonsense mutation in CEP290 induces exon skipping and leads to a relatively mild retinal phenotype.
Invest Ophthalmol Vis Sci, Feb 3. [Epub ahead of print]

Littink KW, van Genderen MM, Collin RWJ, Roosing S, de Brouwer APM, Riemslag FCC, Venselaar H, Thiadens AAHJ, Hoyng CB, Rohrschneider K, den Hollander AI, Cremers FPM, den Born LI (2009)
A novel homozygous nonsense mutation in CABP4 causes congenital cone-rod synaptic disorder.
Invest Ophthalmol Vis Sci. 2009 May;50(5):2344-50

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 Mb human ortholog of Drosophila eyes shut/spacemaker that is mutated in patients with retinitis pigmentosa.
Am J Hum Genet. 2008 Nov;83(5):594-603

∗ *These first authors contributed equally*
# *These last authors contributed equally*

PRESENTATIONS

Nonsense mutation in *CEP290* leads to relatively mild phenotype
Dutch Ophthalmology Conference, March 2010

Intrafamilial clinical variability explained by nonsense-associated altered splicing and by a *MERTK* modifier allele in a family carrying splice site mutations in *CEP290*.
ARVO-NED, November 2009
2nd price for best presentation

Genetic causes of hereditary blindness & why it is necessary to search for them.
NVBS, Retina information and contact day for patients, Utrecht, November 2009

Identification of *EYS*; a novel 2-megabases human orthologue of *Drosophila* eyes shut that is mutated in patients with retinitis pigmentosa.
ARVO-NED, November 2008

Congenital blindness, retinitis pigmentosa and cone-rod dystrophy: Identification of molecular causes and genotype-phenotype correlation.
1st young researcher's workshop on Vision and Neurodegeneration, Potsdam, Germany. March 2007

POSTERS

Identification of novel genetic defects in outbred cone-rod dystrophy patients using homozygosity mapping.
ARVO 2009 Annual Meeting, Fort Lauderdale, Florida, USA. May 2009

Identification of novel genetic defects in outbred cone-rod dystrophy patients using homozygosity mapping.
ProRetina Meeting, Potsdam, Germany. April 2009

Novel homozygous nonsense mutation in *CABP4* causes severe cone dysfunction and relatively spared rod function
21st Course in medical genetics, Bertinoro, Italy. May 2009

COURSES

May 2008	21st Course in Medical Genetics, Bertinoro, Italy
Sept 2007	Excellence in communication. Course at masterprogram of "Molecular Mechanisms of Diseases", Radboud University Nijmegen
2002-2004	Participant in "Honours Program" Radboud University Nijmegen