Curriculum Vitae

Personal details

Full name: Alejandro Garanto
Date of birth: 12 September 1983
Place of birth: Barcelona, Spain
Nationality: Spanish
Research ID: D-5022-2014

Education

Since 2017 — Department of Human Genetics, Radboud University Medical Centre, Nijmegen (The Netherlands)
Donders Institute for Brain, Cognition and Behaviour, Nijmegen (The Netherlands)
Assistant Professor in Therapeutic RNA and DNA editing for eye disorders

2012-2017 — Department of Human Genetics, Radboud University Medical Centre, Nijmegen (The Netherlands)
Radboud Institute for Molecular Life Sciences (RIMLS), Nijmegen (The Netherlands)
Donders Institute for Brain, Cognition and Behaviour, Nijmegen (The Netherlands)
Postdoctoral researcher in the group of Dr. Rob Collin
Antisense oligonucleotide-based therapy for *CEP290*-associated LCA (2012-present)
Expanding the use of antisense oligonucleotide-based therapies for IRDs (2016-present)

2011-2012 — Genetics Department, University of Barcelona, Barcelona (Spain)
Postdoctoral research assistant in the group of Dr. Gemma Marfany
Contribution of Ubiquitin and Sumo signalling in the determination of photoreceptor fate

2006-2011 — PhD in Biology, Genetics Department, University of Barcelona (Barcelona, Spain)
"*CERKL*, generation of a knockout model of retinitis pigmentosa and functional studies" *Excellent Cum Laude*

2008 — Diploma of advanced studies (D.E.A) in Genetics (equivalent to the current Master), University of Barcelona (Barcelona, Spain). ;
Note: D.E.A was part of the old program of the PhD studentship in Spain.

2006 — B.Sc. Degree in Biology, University of Barcelona (Barcelona, Spain)
Speciality: Biomedicine

Research positions

Since April 2017	Department of Human Genetics, Radboud University Medical Centre, Nijmegen (The Netherlands) Donders Institute for Brain, Cognition and Behaviour, Nijmegen (The Netherlands) Assistant Professor in Therapeutic RNA and DNA editing
2012-2017	Department of Human Genetics, Radboud University Medical Centre, Nijmegen (The Netherlands) Radboud Institute for Molecular Life Sciences (RIMLS), Nijmegen (The Netherlands) Donders Institute for Brain, Cognition and Behaviour, Nijmegen (The Netherlands) Postdoctoral researcher in the group of Dr. Rob Collin Antisense oligonucleotide-based therapy for *CEP290*-associated LCA (2012-present) Expanding the use of antisense oligonucleotide-based therapies for IRDs (2016-present)
2011-2012	Genetics Department, University of Barcelona, Barcelona (Spain) Postdoctoral research assistant in the group of Dr. Gemma Marfany Contribution of Ubiquitin and Sumo signalling in the determination of photoreceptor fate
2006-2011	Genetics Department, University of Barcelona, Barcelona (Spain) PhD Student in the group of Prof. Dr. Roser Gonzàlez-Duarte and Dr. Gemma Marfany Functional studies and characterization of the Retinitis Pigmentosa-associated *CERKL*
2004-2006	Genetics Department, University of Barcelona, Barcelona (Spain) Undergraduate student (from Sept. 2004 to Jan. 2006) in the group of Prof. Dr. Roser Gonzàlez Mutational studies by sequencing analyses of Retinitis Pigmentosa causative genes
2004	Balagué Center, Barcelona (Spain) Internship (3 months: from June to Sept 2004) Technical training in a Clinical Laboratory

National and international stays

March 2014-May 2014	Stephen A Wynn Institute for Vision Research, University of Iowa, Iowa City (USA) Nine-week stay in the group of Dr. Budd A. Tucker
Jan 2011	Physiology Department, University of Alcalá de Henares (UAH), Madrid (Spain) Two-week stay in the laboratory of Prof. Pedro de la Villa and Dr. Román Blanco
Mar 2009-Aug 2009	Dean McGee Eye Institute (DMEI). Oklahoma University Health Sciences Center, Oklahoma City (USA) Six-month stay in the group of Prof. Dr. Robert E. Anderson
Aug 2008-Oct 2008	Dean McGee Eye Institute (DMEI). Oklahoma University Health Sciences Center, Oklahoma City (USA) Three-month stay in the group of Prof. Dr. Robert E. Anderson

2006 - 2011 RUBAM (Research Unit on Bioactive Molecules). Institute of Advanced Chemistry of Catalonia (IQAC). Spanish National Research Council (CSIC), Barcelona (Spain)
Visiting and performing experiments at the lab of Prof. Dr. Gemma Fabriàs and Dr. Josefina Casas on a regular basis

Permits/Accreditations

2012 Authorization to perform experimental work with animals in The Netherlands (Art. 9)
2012 Authorization to work with Genetically Modified Organisms in The Netherlands (ML-II,D-II)
2007 Authorization to perform experimental work with animals in Spain

Patents

2018 Antisense oligonucleotides rescue aberrant splicing of *ABCA4*
Patent application number 18184432.5

2016 Antisense oligonucleotides for the treatment of Stargardt disease
Patent application in Europe number 16203864.0

Grants

2018 Uitzicht Grant application
Splicing modulation therapy for recurrent *ABCA4* mutations underlying Stargardt disease
November 2018 – October 2022 (80.000 €)
2017 FFB Project Program Award (PPA)
Splice Modulation to Treat Inherited Retinal Diseases
2,5 million $. PI of Module 4 (468.000 $). May 2017-April 2022
2016 Off Road ZonMw
A novel approach to correct mutant transcripts in inherited retinal dystrophies
October 2016-March 2018 (125.000 €)
2015 Utizicht Grant application
Expanding splice modulation therapies for inherited retinal dystrophies
April 2016-March 2019 (241.074 €)

Fellowships

2018 Retinal Degeneration 2018 Travel Grant (300 $ plus registration fee (600 $) and accommodation (873 €)
2016 Pro Retina Germany Awardee, Retinal Degeneration Meeting 2016 Travel Grant (1.100 $ plus registration fee (630 $) and accommodation (1.100 €)
2016 EyeTN final conference: Vision beyond the genome Travel grant (550 €)
2016 ARVO 2016 International Travel Grant (1.100 $ plus registration fee)
2015 COST action travel grant (360 €)
2015 RIMLS travel grant award 2015 (1.000 €)
2014 EMBO short-term fellowship (5.420 €)
2014 Dondersfonds travel grant (1.000 €)
2013 Simonsfonds travel grant (400 €)
2009 FPI Short Stay Fellowship for 6 months (8.850 €)
2008 CIBERER Short Stay Fellowship for 3 months (3.000 €)
2007 FPI Fellowship for PhD studies
2006 Gestiones y Promociones inmobiliarias EGARA S.L Fellowship (56GE200602029) for PhD studies
2005 Internship Fellowship to collaborate with a Department at the University of Barcelona 2005-2006

Academic achievements: Published articles

Splice-modulating oligonucleotide QR-110 restores CEP290 mRNA and function in human c.2991+1655A>G LCA10 models
Dulla K., Aguila M., Lane A., Jovanovic K., Parfitt DA., Schulkens I., Chan HL., Schmidt I., Beumer W., Vorthoren L., Collin RWJ., **Garanto A.**, Duijkers L., Brugulat-Panes A., Semo M., Vugler AA., Biasutto P., Adamson P. & Cheetham ME.
Molecular Therapy Nucleic Acids (in press DOI: 10.1016/j.omtn.2018.07.010)
Impact factor (2016): 6.392 Area: Medicine, Research & Experimental Quartile: Q1

An FEVR-associated mutation in ZNF408 alters the expression of genes involved in the development of vasculature
Karjosukarso DW., van Gestel SHC., Qu J., Kouwenhoven EN., Duijkers L., **Garanto A.**, Zhou H.*& Collin RWJ.*
Hum Mol Genet. 2018 Jul 4 (in press)
Impact factor (2016): 5.340 Area: Genetics & Heredity Quartile: Q1
* Contributed equally to this work

Whole-Exome Sequencing in Age-Related Macular Degeneration Identifies Rare Variants in COL8A1, a Component of Bruch's Membrane
Corominas J., Colijn JM., Geerlings MJ., Pauper M., Bakker B., Amin N., Lores Motta L., Kersten E., **Garanto A.**, Verlouw JAM., van Rooij JGJ., Kraaij R., de Jong PTVM., Hofman A., Vingerling JR., Schick T., Fauser S., de Jong EK., van Duijn CM., Hoyng CB., Klaver CCW. & den Hollander AI.
Ophthalmology. Apr 26. pii: S0161-6420(17)33149-4 (2018)
Impact factor (2016): 8.204 Area: Ophthalmology Quartile: Q1

Identification and rescue of splice defects caused by two neighboring deep-intronic ABCA4 mutations underlying Stargardt disease
Albert S.*, **Garanto A.***, Sangermano R., Khan M., Bax N., Hoyng CB., Zernant J., Lee W., Allikmets R., Collin RWJ. & Cremers FPM.
American Journal of Human Genetics. Apr 5;102(4):517-527 (2018)
Impact factor (2016): 9.025 Area: Genetics & Heredity Quartile: Q1
* Contributed equally to this work

Antisense oligonucleotide-based splicing correction in individuals with CEP290-associated Leber congenital amaurosis compound heterozygous for the c.2991+1655A>G mutation
Duijkers L., van den Born LI., Neidhardt J., Bax N., Pierrache L., Klevering JB., Collin RWJ. & **Garanto A.**
International Journal of Molecular Sciences. Mar 7;19(3). pii: E753 (2018)
Impact factor (2016): 3.226 Area: Biochemistry & Molecular Biology Quartile: Q2

ABCA4 midigenes reveal the full splice spectrum of all reported noncanonical splice site variants in Stargardt disease
Sangermano R.*, Khan M.*, Cornelis SS., Richelle V., Albert S., **Garanto A.**, Elmelik D., Qamar R., Lugtenberg D., van den Born LI., Collin RWJ., Cremers FPM.
Genome Research Jan;28(1):100-110 (2018)
Impact factor (2016): 11.922 Area: Genetics & Heredity Quartile: Q1
* Contributed equally to this work

Therapeutic effects of the mitochondrial ROS-redox modulator KH176 in a mammalian model of Leigh disease
de Haas R., Das D., **Garanto A.**, Renkema HG., Greupink R., van den Broek P., Pertijs J., Collin RWJ., Willems P, Beyrath J., Heerschap A., Russel FG. & Smeitink JA.

Scientific Reports Sep 15;7(1):11733 (2017)
Impact factor (2016): 4.259 Area: Multidisciplinary Sciences Quartile: Q1

Applications of antisense oligonucleotides for the treatment of inherited retinal diseases
Collin RWJ. & Garanto, A.
Current opinion in ophthalmology May;28(3):260-266 (2017)
Impact factor (2016): 2.920 Area: Ophthalmology Quartile: Q1

Antisense Oligonucleotide-based Splice Correction for USH2A-associated Retinal Degeneration Caused by a Frequent Deep-intronic Mutation.
Slijkerman RW., Vaché C., Dona M., García-García G., Claustres M., Hetterschijt L., Peters TA., Hartel BP., Pennings RJ., Millan JM., Aller E., **Garanto A.**, Collin RW., Kremer H., Roux AF.& Van Wijk E.
Molecular Therapy Nucleic Acids Nov 1;5(10):e381 (2016)
Impact factor (2016): 6.392 Area: Medicine, Research & Experimental Quartile: Q1

In vitro and in vivo rescue of aberrant splicing in CEP290-associated LCA by antisense oligonucleotide delivery. Human Molecular Genetics
Garanto A., Chung DC., Duijkers L., Corral-Serrano JC., Messchaert M., Xiao R., Bennett J., Vandenberghe LH. & Collin RWJ.
Human Molecular Genetics Apr 22 ddw118 (2016)
Impact factor (2016): 5.340 Area: Genetics & Heredity Quartile: Q1

Expression atlas of the deubiquitinating enzymes in the adult mouse retina, their evolutionary diversification and phenotypic roles
Esquerdo M., Grau-Bové X., **Garanto A.**, Toulis V., Garcia-Monclús S., Millo E., López-Iniesta MJ., Abad-Morales V., Ruiz-Trillo I. & Marfany G.
PLoS One Mar 2;11(3):e0150364 (2016)
Impact factor (2016): 2.806 Area: Multidisciplinary Sciences Quartile: Q1

Photoreceptor progenitor mRNA analysis reveals exon skipping due to the ABCA4 c.5461-10T>C mutation in Stargardt disease
Sangermano R., Bax N., Bauwens M., van den Born LI., de Baere E., **Garanto A.**, Collin RWJ., Goercharn-Ramlal ASA., Engelsman-van Dijk AHA., Rohrschneider K., Hoyng CB., Cremers FPM. & Albert S.
Ophthalmology Jun;123(6):1375-85 (2016)
Impact factor (2016): 8.204 Area: Ophthalmology Quartile: Q1

Whole exome sequencing reveals ZNF408 as a new gene associated with autosomal recessive retinitis pigmentosa with vitreal alterations
Avila-Fernandez A., Perez-Carro R., Corton M., Lopez-Molina MI., Campello L., **Garanto A.**, Fernandez-Sanchez L., Duijkers L., Lopez-Martinez MA., Riveiro-Alvarez R., Rodrigues Jacy da Silva L., Sanchez-Alcudia R., Martin-Garrido E., Reyes N., Garcia-Garcia F., Dopazo J., Garcia-Sandoval B., Collin RWJ., Cuenca N. & Ayuso C.
Human Molecular Genetics Jul 15;24(14):4037-48 (2015)
Impact factor (2015): 5.985 Area: Genetics & Heredity Quartile: Q1

Species-dependent splice recognition of a cryptic exon resulting from a recurrent intronic CEP290 mutation that causes congenital blindness
Garanto A., Duijkers L. & Collin RWJ.
International Journal of Molecular Sciences 16(3), 5285-5298 (2015)
Impact factor (2015): 3.257 Area: Biochemistry & Molecular Biology Quartile: Q2

Expression of the SUMO pathway genes in the mouse retina.
Abad-Morales V., Domènech EB., **Garanto A.** & Marfany G.
Biology Open 4:224-232 (2015)
Impact factor (2015): 2.135 Area: Biology Quartile: Q2

Unexpected CEP290 mRNA splicing in a humanized knock-in mouse model for Leber congenital amaurosis
Garanto A., van Beersum SEC., Peters TA., Roepman R., Cremers FPM. & Collin RWJ.
PLoS One. 8(11): e79369 (2013).
Impact factor (2013): 3.534 Area: Multidisciplinary Sciences Quartile: Q1

Specific sphingolipid content decrease in Cerkl knockdown mouse retinas
Garanto A., Mandal NA., Egido-Gabás M., Marfany G., Fabriàs G., Anderson RE., Casas J. & Gonzàlez-Duarte R.
Experimental Eye Research 110:96-106 (2013).
Impact factor(2013): 3.017 Area: Ophthalmology Quartile: Q1

Targeted knockdown of Cerkl, a retinal dystrophy gene, causes mild affectation of the retinal ganglion cell layer
Garanto A., Vicente J., Riera M., De La Villa P., Gonzàlez-Duarte R., Blanco R. & Marfany G.
Biochim Biophys Acta - Molecular Basis of Disease 1822: 1258-1269 (2012).
Impact factor (2012): 4.910 Area: Biochemistry & Molecular Biology Quartile: Q1

High transcriptional complexity of the retinitis pigmentosa CERKL gene in human and mouse
Garanto A., Riera M., Pomares E., Permanyer J., De Castro-Miró M., Sava F. Abril JF., Marfany G. & Gonzàlez-Duarte R.
Investigative Ophthalmology & Visual Science 52: 5202-5214 (2011).
Impact factor (2011): 3.597 Area: Ophthalmology Quartile: Q1

Overexpression of CERKL, a gene responsible for Retinitis Pigmentosa in humans, protects cells from apoptosis induced by oxidative stress
Tuson M.*, **Garanto A.***, Gonzalez-Duarte R. & Marfany G.
Molecular Vision 15: 168-180 (2009)
Impact factor (2009): 2.541 Area: Ophthalmology Quartile: Q1
* Contributed equally to this work

Academic achievements: book chapters

Antisense oligonucleotide-based splice correction of a deep-intronic mutation in CHM underlying choroideremia
Garanto A., van der Velde-Visser SD., Cremers FPM. & Collin RWJ.
Retinal Degenerative Diseases. Adv Exp Med Biol. 2018;1074:83-89

Design and in vitro use of antisense oligonucleotides to correct pre-mRNA splicing defects in inherited retinal dystrophies
Garanto A. & Collin RWJ.
Retinal Gene Therapy. Methods Mol. Biol. 2018;1715:61-78

Combining zebrafish and mouse models to test the function of deubiquitinating enzymes (Dubs) genes in development: Role of USP45 in the retina.
Toulis V., Garanto A. & Marfany G.
Proteostasis. Methods Mol. Biol. 2016;1449:85-101

Antisense oligonucleotide therapy for inherited retinal dystrophies
Gerard X., Garanto A., Rozet JM. & Collin RWJ.
Retinal Degenerative Diseases. Adv Exp Med Biol. 2016;854:517-24

Invited speaker

2017 Guest lecturer: Master in Genetics and Genomics (University of Barcelona). 29 November (Barcelona, Spain)
RNA therapies for inherited retinal diseases: design & proof of concept studies

2017 Invited Speaker: Spanish Society of Genetics. 24-27 October 2017 (Gijón, Spain)
Splice modulation therapy for rare inherited diseases

2017 Pecha Kucha talk at the Health Valley Event. 9 March 2017 (Nijmegen, Netherlands)
Development of therapeutic approaches for inherited retinal degenerations

2014 Guest lecturer: Master in Genetics (University of Barcelona). 23 November (Barcelona, Spain)
Correcting mRNA splicing as a retinal dystrophy gene therapy

Academic achievements: conference contributions (presenter underlined)

Identification and correction of pre-mRNA splicing defects underlying Stargardt disease due to deep-intronic variants in ABCA4. **Garanto A.**, Sangermano R., Albert, S., Khan M., Bauwens M., Naessens, S., Allikmets R., ven den Born LI., Hoyng CB., De Baere E., Cremers FPM. & Collin RWJ. Retinal degeneration meeting 2018. September 2018. **Oral presentation**. Killerney, Ireland

Antisense oligonucleotide-based restoration of ABCA4 splicing defects caused by deep-intronic mutations associated with Stargardt disease. **Garanto A.**, Sangermano R., Albert, S., Khan M., Bauwens M., Naessens, S., Allikmets R., ven den Born LI., Hoyng CB., De Baere E., Cremers FPM. & Collin RWJ. ARVO 2018 Annual Meeting. May 2018. **Poster**. Honolulu, Hawaii (USA)

Antisense oligonucleotide-based restoration of ABCA4 splicing defects caused by deep-intronic mutations associated with Stargardt disease. **Garanto A.**, Sangermano R., Albert, S., Khan M., Bauwens M., Naessens, S., Allikmets R., ven den Born LI., Hoyng CB., De Baere E., Cremers FPM. & Collin RWJ. NVGCT Symposium 2018. March 2018. **Oral presentation**. Lunteren, Netherlands.

Developing new antisense oligonucleotide-based therapeutic approaches for inherited retinal dystrophies. **Garanto A.**, Albert S., Naessens S., Duijkers L., Sangermano R., Bouwens M., De Baere E., Cremers FPM. & Collin RWJ. Retinal Degeneration 12th Pro Retina Research colloquium Potsdam 2017. April 2017. **Poster & Oral presentation** (one of the 8 best abstracts out of 56). Potsdam, Germany.

Expanding the use of antisense oligonucleotide-based therapies for inherited retinal dystrophies. **Garanto A.**, Albert S., Duijkers L., Sangermano R., Cremers FPM. & Collin RWJ. NVGCT Symposium 2017. March 2017. **Oral presentation**. Lunteren, Netherlands.

Antisense oligonucleotide-based splice correction is an effective therapeutic approach for CEP290-associated LCA. **Garanto A.**, Chung DC., Duijkers L., Corral-Serrano JC., Messchaert M., Xiao R., Bennett J., Vandenberghe LH. & Collin RWJ. Retinal degeneration meeting 2016. September 2016. **Oral presentation**. Kyoto, Japan

Expanding the use of antisense oligonucleotide-based therapies for inherited retinal dystrophies. **Garanto A.**, Albert S., Duijkers L., Sangermano R., Cremers FPM. & Collin RWJ. Retinal degeneration meeting 2016. September 2016. Poster. Kyoto, Japan

Antisense oligonucleotide-based splice modulation for CEP290-associated LCA. **Garanto A.**, Chung DC., Duijkers L., Corral-Serrano JC., Messchaert M., Xiao R., Bennett J., Vandenberghe LH. & Collin RWJ. EyeTN final conference: Vision Beyond the Genome. June 2016. **Oral presentation**. Montpellier, France

Identification and validation of intronic ABCA4 mutations in Stargardt patients by using induced pluripotent stem cell-derived photoreceptor progenitor cells. Albert S., Sangermano R., Bax N., Bauwens M., van den Born LI., de Baere E., **Garanto A.**, Collin RWJ. & Cremers FPM. ARVO 2016 Annual Meeting. May 2016. Poster. Seattle, Washington (USA)

Antisense Oligonucleotide Delivery is an Effective Therapeutic Approach for CEP290-Associated LCA. **Garanto A.**, Chung DC., Duijkers L., Xiao R., Bennett J., Vandenberghe LH. & Collin RWJ. ARVO 2016 Annual Meeting. May 2016. **Oral presentation**. Seattle, Washington (USA)

Antisense Oligonucleotide Delivery is an Effective Therapeutic Approach for CEP290-Associated LCA. **Garanto A.**, Chung DC., Duijkers L., Xiao R., Bennett J., Vandenberghe LH. & Collin RWJ. The Oligo Meeting (11th Annual Meeting of the Oligonucleotide Therapeutics Society). October 11-14 2015. **Oral presentation**. Leiden (Netherlands)

Investigating the role of C2orf71 in the pathogenesis of retinitis pigmentosa. Corral Serrano JC, Letterboer S., **Garanto A.**, Roepman R. & Collin RWJ. ARVO 2015 Annual Meeting. May 2015. Poster. Denver, Colorado (USA)

Antisense oligonucleotide- based therapy for CEP290-associated LCA. **Garanto A.**, Duijkers LEM., Xiao R., Bennet, J., Vandenberghe LH. & Collin RWJ., Radboud Science Day. April 13th 2015. Laptop presentation. Nijmegen (Netherlands)

Expression of the SUMO pathway genes in the mouse retina. Abad-Morales V., Domènech EB., **Garanto A.** & Marfany G. First Proteostasis Meeting. November 2014. Poster. Valencia (Spain)

Deubiquitinating enzymes in retinal health and neurodegeneration: expression analysis and functional studies. Toulis V., Garcia-Monclús S, **Garanto A.** & Marfany G. First Proteostasis Meeting. November 2014. Poster. Valencia (Spain)

Antisense oligonucleotide- based therapy for CEP290-associated LCA. **Garanto A.**, Duijkers LEM., Xiao R., Bennet, J., Vandenberghe LH. & Collin RWJ., 2014 European Society of Gene and Cell Therapy (ESGCT) meeting. October 2014. Poster. The Hague (Netherlands)

Deubiquitinating enzymes in retinal health and neurodegeneration: expression analysis and functional studies. Toulis V., Garcia-Monclús S, **Garanto A.** & Marfany G. Biochemical Basis of Healthy Aging. September 2014. Oral communication. Spetses (Greece) (Best oral presentation award)

Antisense oligonucleotide- based therapy for CEP290-associated LCA. **Garanto A.**, Duijkers LEM., Xiao R., Vandenberghe LH. & Collin RWJ. XVI International Symposium on Retinal Degeneration RD2014. July 2014. Poster. Pacific Grove, California (USA) (Best Poster Award)

Deubiquitinating enzymes in retinal health and neurodegeneration: expression analysis and functional Studies. Toulis V., Garcia-Monclús S, Vystavělová V., **Garanto A.** & Marfany G. XXII Jornades de la Societat Catalana de Biologia. June 2014. Poster. Barcelona (Spain).

*Expression atlas of SUMO pathway genes in the mouse retina. Abad-Morales V., Domènech EB., **Garanto A.** & Marfany G.* XXII Jornades de la Societat Catalana de Biologia. June 2014. Oral communication. Barcelona (Spain).

AAV-mediated antisense oligonucleotide-based therapy for CEP290-associated LCA. **Garanto A.**, Duijkers LEM., Xiao R., Vandenberghe LH. & Collin RWJ. American Society of Gene and Cell Therapy (ASGCT) meeting. May 2014. Poster. Washington (USA) (Outstanding Poster Award)

AAV-mediated antisense oligonucleotide-based therapy for CEP290-associated LCA. **Garanto, A.**, Duijkers LEM., Xiao R., Vandenberghe LH. & Collin, RWJ. ARVO 2014 Annual Meeting. May 2014. Poster. Orlando, Florida (USA)

Specific sphingolipid content decrease in Cerkl knockdown mouse retinas. **Garanto A.**, Mandal NA., Egido-Gabás M., Marfany G., Fabriàs G., Anderson RE., Casas J. & Gonzàlez-Duarte R. Sphingolipid Intenational Meeting. June 2013. Poster. Assisi (Italy)

Antisense oligonucleotide-based therapy for CEP290-associated LCA. **Garanto A.**, Cremers FPM., Collin RWJ. ARVO 2013 Annual Meeting. May 2013. Poster. Seattle, Washington (USA)

Antisense oligonucleotide-based therapy for CEP290-associated LCA. **Garanto A.**, Cremers FPM. & Collin RWJ. 1st IGMD Science Day. March 2013. Oral presentation. Nijmegen (The Netherlands)
Expression pattern and map of SUMO and ubiquitin pathway enzymes in the mouse retina. Esquerdo M., Abad V., Millo E., Garcia-Monclus S., Toulis, V., Iniesta MJ., **Garanto A.** & Marfany G. Global questions in advanced biology (Societat Catalana de Biologia). July 2012. Poster. Barcelona (Spain)

Cerkl knockdown murine model shows a mild affectation of the retinal ganglion cell layer. Gonzàlez-Duarte R., **Garanto A.**, Vicente-Tejedor J., Riera M., De La Villa P., Blanco R. & Marfany G. ARVO 2012 Annual Meeting. May 2012. Poster #1620. Fort Lauderdale, Florida (USA)

High transcriptional complexity of the Retinitis Pigmentosa CERKL gene in human and mouse. Marfany G., **Garanto A.**, Riera M., Pomares E., Permanyer J., De Castro-Miró M., Abril JF. & Gonzàlez-Duarte R. ARVO 2011 Annual Meeting. May 2011. Poster. Number 5404. Fort Lauderdale, Florida (USA)

High transcriptional complexity of the Retinitis Pigmentosa CERKL gene in human retina. Riera M., **Garanto A.**, Pomares E., Permanyer J., Abril JF., Marfany G. & Gonzàlez-Duarte R. IV Reunión Anual del CIBERER. October 2010. Oral presentation. Madrid (Spain)

CERKL function and contribution to pathogenesis. Riera M., **Garanto A.**, Esteban I., Fathinajafabadi A., Marfany G., Knecht E. & Gonzalez-Duarte R. XIVth International Symposium on Retinal Degeneration. July 2010. Poster. Number 91. Montremblant, Quebec (Canada)

CERKL, an orphan lipid kinase. Garanto A., Marfany G., Mandal NA., Anderson RE. & Gonzàlez-Duarte R. 4th Annual Student & Postdoctoral Fellow Workshop. June 2009. Oral presentation. Pag. 22. Oklahoma City, OK (USA)

Overexpression of CERKL, a Retinitis Pigmentosa gene, protects cells from apoptosis under oxidative stress conditions. **Garanto A.**, Tuson M., Gonzàlez-Duarte R. & Marfany G. European Human Genetic Conference 2008. June 2008. Poster. Pag. 277. Barcelona (Spain)

Functional characterization of CERKL, a Retinitis Pigmentosa gene. **Garanto A.**, Tuson M., Gonzàlez-Duarte R. & Marfany G. ARVO 2008 Annual Meeting. April 2008. Poster. Pag. 280. Fort Lauderdale, Florida (USA)

Construcción de un modelo murino knockout condicional para el gen Ceramida Kinasa-Like (CERKL). **Garanto A.**, Tuson M., Pomares E., Permanyer J., Méndez P., Marfany G. & Gonzàlez-Duarte R. 1ª Reunión Anual del CIBERER. November 2007. Poster. Pag. 47. Barcelona (Spain)

Construcción de un modelo murino knockout condicional para el gen Ceramida Kinasa-Like (CERKL). **Garanto A.**, Tuson M., Marfany G. & Gonzàlez-Duarte R. XXXVA congreso de la Sociedad Española de Genética. September 2007. Poster. Pag. 146. León (Spain)

Construction of a conditional knockout mouse model of the Ceramide Kinase-Like gene (CERKL). **Garanto A.**, Tuson M., Gonzàlez-Duarte R. & Marfany G. Sphingolipid Intenational Meeting. November 2006. Oral presentation. Pag. 22. Calella de Mar (Spain)

Co-supervision of:

Master students & Bachelor students

2018 *Splice correction therapy for a recurrent mutation in ABCA4 underlying Stargardt disease*
Lea Weiss (Molecular Mechanisms of Disease Master Student, Radboud University)

2018 *Design and assessment of bifunctional AONs for exon inclusion*
Koen Jakobs (Medical Biology Master Student, Radboud University)

2017 *Expanding splice modulation therapies for inherited retinal dystrophies*
Núria Torres Farràs (Erasmus Bachelor Student, University of Barcelona, Spain)

2017 *Development of an innovative therapeutic approach for inherited retinal dystrophies*
Gözde Büyükkahraman (Molecular Mechanisms of Disease Master Student, Radboud University)

2016 *Development of a Splice Modulation Therapeutic Approach for Stargardt Disease*
Hind Almushattat (Biomedical Sciences Student, University of Amsterdam)

2015 *Expanding splice modulation therapies for inherited retinal dystrophies*
Ruben van Osch (Molecular Mechanisms of Disease Master Student, Radboud University)

2014 *Molecular genetic analysis of Indonesian LCA patients & AON optimization for CEP290-associated Leber Congenital Amaurosis*
Widya Eka Nugraha (Genetics Master's program student, Semarang, Indonesia)

2014 *Antisense oligonucleotide delivery in the mouse retina*
Manon Oud (Molecular Mechanisms of Disease Master Student, Radboud University)

2013 *Identification of ZNF408 transcriptional targets*
Dyah Karjosukarso (Molecular Mechanisms of Disease Master Student, Radboud University)

2012 *Expression analysis of deubiquitinating enzyme families in the mouse retina*
Erica Millo (Erasmus master student, Italy)

2012 *Expression of the deubiquitinating enzymes in the retina and their contribution to the transcriptional regulation of photoreceptor specific genes*
Mariona Esquerdo (Genetics Master's program student, University of Barcelona)

2012 *Retinal expression of genes involved in neurodegeneration*
Silvia Garcia-Monclús (Biomedical Sciences bachelor student, University of Barcelona)

2012 *Mutational screening of genes involved in the stress pathways in the King-Kopetzky Syndrome*
Judit Domingo (Biomedical Sciences bachelor student, University of Barcelona)

2012 *Mutational screening in a patient with King-Kopetzky Syndrome*
Luz Jubierre (Biomedical Sciences bachelor student, University of Barcelona)

2011 *Expression of SUMO and related enzymes in the retina*
Victor Abad (Genetics Master's program student, University of Barcelona)

Technicians

Since January 2013 co-supervision of the work of the technician in the group, Lonneke Duijkers.
Jan-Dec 2017- co-supervision of the work of the technician in the group, Simon van Reijmersal
Since August 2017 supervision of Anita Hoogendoorn

PhD candidates

Co-promoter of one PhD student (Julio Corral Serrano) defence is expected in 2018

Thesis defence committe member

2017 Marta de Castro Miro (University of Barcelona). September 13th.

Attended conferences, courses and workshops

2018 ARVO 2018 Annual Meeting – Honolulu, Hawaii (USA)
2018 Dutch Society of Gene and Cell Therapy (NVGCT) 2018 – Lunteren (The Netherlands)
2017 Young Researcher Vision Camp 2017 – Leibertingen (Germany)
2017 Retinal Degeneration 12th Pro Retina Research colloquium 2017- Potsdam (Germany)
2017 Dutch Society of Gene and Cell Therapy (NVGCT) 2017 – Lunteren (The Netherlands)
2016 Donders Perception Day – Nijmegen (The Netherlands)
2016 Retinal degeneration meeting 2016 – Kyoto (Japan)
2016 EyeTN final conference: Vision Beyond the Genome – Montpellier (France)
2016 ARVO 2016 Annual Meeting – Seattle, Washington (USA)
2015 11th Annual Meeting of the Oligonucleotide Therapeutics Society - Leiden (The Netherlands)
2015 Animal models for exon skipping workshop - Munich (Germany)
2015 Training school on patient communication - Leiden (The Netherlands)
2015 Radboud Science Day - Nijmegen (The Netherlands)
2014 European Society of Gene and Cell Therapy conference - The Hague (The Netherlands)
2014 ARVO 2014 Annual Meeting - Orlando, Florida (USA)
2013 Academic writing course - Nijmegen (The Netherlands)
2013 ARVO 2013 Annual Meeting – Seattle, Washington (USA)
2013 1st IGMD (Institute for Genetic and Metabolic Disease) Science Day – Nijmegen (the Netherlands)
2013 DOPS-meeting, Dutch Opthalmology PhD-students – Nijmegen (The Netherlands)
2010 4th Course of Human Genetics. Spanish Society of Genetics – Barcelona (Spain)
2009 4th Course of "Expression and purification of recombinant proteins" – Sevilla (Spain)
2009 Advances on the Molecular Diagnosis and Therapy of Retinal Dystrophies – Barcelona (Spain)
2009 4th Annual Student & Postdoctoral Fellow Workshop – Oklahoma City, Oklahoma (USA)
2009 ARVO 2009 Annual Meeting – Fort Lauderdale, Florida (USA)
2008 European Human Genetic Conference – Barcelona (Spain)
2008 XVI Molecular Biology Conference (Catalan Society of Biology) – Barcelona (Spain)
2008 ARVO 2008 Annual Meeting – Fort Lauderdale, Florida (USA)
2007 1st Annual Meeting CIBERER – Barcelona (Spain)
2007 XXXVA meeting of the Spanish Society of Genetics – León (Spain)
2007 XV Molecular Biology Conference (Catalan Society of Biology) – Barcelona (Spain)
2007 Training for research personnel who use animals for experiments and other purposes, Generalitat de Catalunya (Spain)
2006 Sphingolipid Intenational Meeting – Calella de Mar, Barcelona (Spain)
2005 XIII Molecular Biology Conference (Catalan Society of Biology) – Barcelona (Spain)

Teaching experience:

2017 RNA therapies for inherited retinal diseases: design & proof of concept studies. Degree: Master in Genetics. 1,5 hours. November (University of Barcelona)
2017 Supervisor medical students for their research training
2017 MMD Masterclass: Molecular Therapies. 40 hours. Degree: Master in Molecular Mechanisms of Diseases (MMD). January (Radboud University). (Deputy coordinator of the course).
2015 EyeTN training course on Antisense Oligonucleotides. 30 hours. September (Radboud university medical center).
2015 Radboud Summer School - Lessons from the Eye: How to Translate Your Basic Research into Clinical Applications. 3 hours. Genetic approaches for IRDs. August (Radboud University).
2015 Gene Therapy for retinal dystrophies. Radboud honours student program. 10 hours. January (Radboud University)
2014 Correcting mRNA splicing as a retinal dystrophy gene therapy. Degree: Master in Genetics. 1,5 hours. November (University of Barcelona)
2014 Gene Therapy for ciliopathies. Degree: Medical Biosciences. 1,5 hours. June (Radboud university)
2014 Gene Therapy for ciliopathies. Degree: Medical Biotechnology, course KMP1. 3 hours. May (Radboud university)
2014 Antisense oligonucleotide-based therapy. IGMD honours student program. 4 hours. January (Radboud University)
2009 Molecular Genetics. Degree: Biology. 25 hours. (University of Barcelona)
2008 Advanced Genetic Engineering. Degree: Biology. 30 hours. (University of Barcelona)
2008 Human Molecular Genetics. Degree: Biochemistry. 15 hours. (University of Barcelona)
2007 Advanced Genetic Engineering. Degree: Biology. 45 hours. (University of Barcelona)
2007 Molecular Genetics. Degree: Biochemistry. 20 hours. (University of Barcelona)

Languages
Catalan: Native
Spanish: Native
English: High level
Dutch: Basic knowledge