

BIO SKETCH

NAME : ZEITZ, Christina

POSITION TITLE : Dr.rer.nat., INSERM CR1

EDUCATION / TRAINING

INSTITUTION AND LOCATION	DEGREE	Month/Year	FIELD OF STUDY
Free Waldorf School, Stuttgart, Germany	Baccalaureate	May/1992	-
Alliance Française, Paris, France	Diplom of French Language	October/1993	French language
Albert Ludwigs University Freiburg, Germany	Intermediate Diplom (equivalent to Bachelor of Science)	September/1995	Chemistry
Free University of Berlin, Germany	Diplom (equivalent to Master of Science)	December/1999	Chemistry
Free University of Berlin, Germany	Dr.rer.nat (equivalent to PhD)	February/2004	Chemistry/Molecular Biology
Université Pierre et Marie Curie6	HDR (Diplôme d'habilitation à diriger des recherches)	May/2012	Life Sciences

POSITIONS AND EMPLOYMENTS

MONTH AA – MONTH BB	Name of the Institution - Title of the Position
February 1996 – March 1996	Internship, Blackholm Medical Diagnostic Laboratory, Blackholm, Schüll and Wolf, Heilbronn, Germany.
October 1996 - June 1997	Graduate student, Department of Biochemistry, University of Washington, Seattle, USA including laboratory work on biochemistry in the Department of Pathology: principal investigator: Prof. Lawrence LOEB, PhD M.D.
October 1997 – April 1999	Student assistant, molecular biology, Department of Human Molecular Genetics, Max-Planck-Institute for Molecular Genetics, Berlin, Germany: principal investigator: Prof. Dr. Hans-Hilger ROPERS.
April 1999 - December 1999	Diploma thesis on "Mutation analyses in patients with X-linked retinitis pigmentosa and studies on the RPGR transcript". Department of Human Molecular Genetics, Max-Planck-Institute for Molecular Genetics, Berlin, Germany in the laboratory of Prof. Dr. Wolfgang BERGER.
December 1999 –	Ph.D. on "Mebasis of X-chromosomal congenital night

September 2003	blindness", Department of Human Molecular Genetics, Max-Planck-Institute for Molecular Genetics, Berlin, Germany and Division of Medical Molecular Genetics and Gene Diagnostics, Institute of Medical Genetics, University of Zurich, Schwerzenbach, Switzerland in the laboratory of Prof. Dr. Wolfgang BERGER.
October 2003 – September 2007	Post-Doctoral Associate on "Different forms and inheritance of congenital stationary night blindness", the Division of Medical Molecular Genetics and Gene Diagnostics, Institute of Medical Genetics, University of Zurich, Schwerzenbach, Switzerland in the laboratory of Prof. Dr. Wolfgang BERGER.
October 2007 - present	Group leader on "Genotype phenotype correlations, gene identification and elucidation of the pathogenic mechanism in patients with different eye diseases", Department of Genetics, Institut de la Vision, Paris, France headed by Prof. José-Alain SAHEL.
October 2010	INSERM nomination: "stagiaire"
October 2011	INSERM nomination: "titularisée"

Scientific Interests :

Genetics, functional *in vitro* and *in vivo* analysis and therapeutically approaches in inherited stationary and progressive retinal disorders.

Other Experiences and Professional Memberships :

SCIENTIFIC MEMBER:

ARVO: The Association of Research in Vision and Ophthalmology:

2014-2017 elected member of the Annual Meeting Program Committee of the Biochemistry/Molecular Biology (BI) section

ASHG: The American Society of Human Genetics

ISER: International Society for Eye Research

ISGEDR: International Society for Genetic Eye Diseases and Retinoblastoma

SGOF: Société de Génétique Ophthalmologique Francophone

RCPG-Physio-Med: Récepteurs couplés aux protéines G – de la physiologie au médicament.

AD HOC PAPER REVIEWER:

JOURNALS:

American Journal of Human Genetics

European Journal of Human Genetics

GeneReviews

Genetics in Medicine

Human Genetics

Human Mutation

Human Molecular Genetics

Investigative Ophthalmology and Visual Sciences:

2013: "exceptional reviewer"

Molecular Vision
Nature Genetics
Ophthalmic Genetics
Orphanet Journal of Rare Diseases
PLOS ONE
PNAS
Progress in Retinal and Eye Research

GRANT APPLICATIONS:

BSF-United States-Israel Binational Science Foundation
FWO-Research Foundation Flanders, Belgium
Inserm – CNRS, France
Retina France, France
SNF-Swiss National Science Foundation
UitZicht, The Netherlands

HDR REVIEWER:

2014 for Université Paris Descartes, Paris, France

THESIS COMMITTEE MEMBER:

03.11.2010: Karin Littink Radboud University Nijmegen, The Netherlands: The power of homozygosity mapping: Discovery of new genetic defects in patients with retinal dystrophy
14.09.2012: Mariana Hajj, IGF – Institut de Génomique Fonctionnelle, CNRS UMR5203 – INSERM U661 – Univ.Montpellier 1 – Univ. Montpellier 2, Montpellier, France.

Selected Peer-reviewed Publications:

1. Peer-reviewed articles

2014

1. Manes G, Guillaumie T, Vos WL, Devos A, Audo I, **Zeit C**, Marquette V, Zanlonghi X, Defoort-Dhellemmes S, Puech B, Mohand Said S, Sahel JA, Odent S, Dollfus H, Kaplan J, Dufier JL, Le Meur G, Weber M, Faivre L, Behar Cohen F, Bérout C, Picot MC, Verdier C, Sénéchal A, Baudoin C, Bocquet B, Findlay JB, Meunier I, Dhaenens CM, Hamel CP (2014) High prevalence of PRPH2 in autosomal dominant retinitis pigmentosa in France and characterization of biochemical and clinical features. *Am J Ophthalmol*. Online 05.11.2014. Corresponding author: Manes G.
2. Burtscher V, Schicker K, Novikova E, Pöhn B, Stockner T, Kugler C, Singh A, **Zeit C**, Lancelot ME, Audo I, Leroy BP, Freissmuth M, Herzig S, Matthes J, Koschak A (2014)

Spectrum of Cav1.4 dysfunction in congenital stationary night blindness type 2. *Biochim Biophys Acta*. 1838:2053-2065. Corresponding author: Koschak A

3. Malaichamy S, Sen P, Sachidanandam R, Arokiasamy T, Lancelot ME, Audo I, **Zeitz C**, Soumittra N (2014) Molecular profiling of complete congenital stationary night blindness: a pilot study on an Indian cohort. *Mol Vis*. 20:341-351. Corresponding author: Soumittra N
4. El Shamieh S, Neuillé M, Terray A, Orhan E, Condroyer C, Démontant V, Michiels C, Antonio A, Boyard F, Lancelot ME, Letexier M, Saraiva JP, Léveillard T, Mohand-Saïd S, Goureau O, Sahel JA, **Zeitz C***, Audo I*. (2014) Whole-exome sequencing identifies KIZ as a ciliary gene associated with autosomal-recessive rod-cone dystrophy. *Am J Hum Genet* 94:625-33. Corresponding author: Audo I and **Zeitz C** * These authors contributed equally to this work
5. Neuillé M, El Shamieh S, Orhan E, Michiels C, Antonio A, Lancelot ME, Condroyer C, Bujakowska K, Poch O, Sahel JA, Audo I, **Zeitz C** (2014) Lrit3 deficient mouse (nob6): a novel model of complete congenital stationary night blindness (cCSNB). *PLoS One*. 9:e90342. Corresponding author: **Zeitz C**
6. Michalakis S, Shaltiel L, Sothilingam V, Koch S, Schludi V, Krause S, **Zeitz C**, Audo I, Lancelot ME, Hamel C, Meunier I, Preising MN, Friedburg C, Lorenz B, Zabouri N, Haverkamp S, Garrido MG, Tanimoto N, Seeliger MW, Biel M, Wahl-Schott CA (2014) Mosaic synaptopathy and functional defects in Cav1.4 heterozygous mice and human carriers of CSNB2. *Hum Mol Genet*. 23:1538-1550. Corresponding author: Wahl-Schott CA
7. Audo I, Bujakowska K, Orhan E, El Shamieh S, Sennlaub F, Guillonneau X, Antonio A, Michiels C, Lancelot ME, Letexier M, Saraiva JP, Nguyen H, Luu TD, Léveillard T, Poch O, Dollfus H, Paques M, Goureau O, Mohand-Saïd S, Bhattacharya SS, Sahel JA, **Zeitz C** (2014) The familial dementia gene revisited: a missense mutation revealed by whole-exome sequencing identifies ITM2B as a candidate gene underlying a novel autosomal dominant retinal dystrophy in a large family. *Hum Mol Genet*. 23:491-501. Corresponding author: Audo I

2013

1. Orhan E, Prézeau L, El Shamieh S, Bujakowska KM, Michiels C, Zagar Y, Vol C, Bhattacharya SS, Sahel JA, Sennlaub F, Audo I, **Zeitz C** (2013) Further insights into GPR179: expression, localization, and associated pathogenic mechanisms leading to complete congenital stationary night blindness. *Invest Ophthalmol Vis Sci*. 54:8041-8050. Corresponding author: **Zeitz C**
2. Storm T, **Zeitz C**, Cases O, Amsellem S, Verroust PJ, Madsen M, Benoist JF, Passemar S, Lebon S, Jønsson IM, Emma F, Koldsø H, Hertz JM, Nielsen R, Christensen EI, Kozyraki R (2013) Detailed investigations of proximal tubular function in Imlerslund-Gräsbeck syndrome. *BMC Med Genet*. 14:111. Corresponding author: Christensen EI

3. Manes G, Meunier I, Avila-Fernández A, Banfi S, Le Meur G, Zanlonghi X, Corton M, Simonelli F, Brabet P, Labesse G, Audo I, Mohand-Said S, **Zeitz C**, Sahel JA, Weber M, Dollfus H, Dhaenens CM, Allorge D, De Baere E, Koenekoop RK, Kohl S, Cremers FP, Hollyfield JG, Sénéchal A, Hebrard M, Bocquet B, Ayuso García C, Hamel CP (2013) Mutations in IMPG1 cause vitelliform macular dystrophies. *Am J Hum Genet.* 93:571-578. Corresponding author: Hamel C
4. Doumanov JA, **Zeitz C**, Dominguez Gimenez P, Audo I, Krishna A, Alfano G, Diaz ML, Moskova-Doumanova V, Lancelot ME, Sahel JA, Nandrot EF, Bhattacharya SS (2013) Disease-causing mutations in BEST1 gene are associated with altered sorting of bestrophin-1 protein. *Int J Mol Sci.* 14:15121-15140. Corresponding author: Doumanov J
5. **Zeitz C**, Jacobson SG, Hamel CP, Bujakowska K, Neuillé M, Orhan E, Zanlonghi X, Lancelot ME, Michiels C, Schwartz SB, Bocquet B; Congenital Stationary Night Blindness Consortium, Antonio A, Audier C, Letexier M, Saraiva JP, Luu TD, Sennlaub F, Nguyen H, Poch O, Dollfus H, Lecompte O, Kohl S, Sahel JA, Bhattacharya SS, Audo I (2013) Whole-exome sequencing identifies LRIT3 mutations as a cause of autosomal-recessive complete congenital stationary night blindness. *Am J Hum Genet.* 92:67-75. Corresponding author: **Zeitz C**
6. Bocquet B, Lacroux A, Surget M-O, Baudoin C, Marquette V, Manes G, Hebrard M, Sénéchal A, Roux A-F, Dhaenens C-M, Allorge D, Rozet J-M, Perrault I, Bonnefont J-P, Kaplan J, Dollfus H, Bonneau P, Audo I, **Zeitz C**, Paquis V, Calvas P, Arveiler B, Kohl S, Wissinger, B Blanchet C, Meunier I, Hamel C (2013) Prevalence of inherited retinal dystrophies and optic neuropathies in Southern France: assessment from a 21-year data management. *Ophthalmic Epidemiology.* 20:13-25. Corresponding author: Hamel C

2012

1. Falk MJ, Zhang Q, Nakamaru-Ogiso E, Kannabiran C, Fonseca-Kelly Z, Chakarova C, Audo I, Mackay DS, **Zeitz C**, Borman AD, Staniszewska M, Shukla R, Palavalli L, Mohand-Said S, Waseem7, Jalali S, Perin JC, Place E, Ostrovsky J, Xiao R, Bhattacharya SS, Consugar M, Webster AR, Sahel J-A, Moore AT, Berson EL, Liu Q, Gai X, Pierce EA (2012) *NMNAT1* Mutations Cause Leber Congenital Amaurosis. *Nature Genetics.* 44:1040-1045. Corresponding author: Pierce E
2. Audo I*, Bujakowska K*, Orhan E, Poloschek CM, Defoort-Dhellemmes, Drumare I, Kohl S, Luu TD, Lecompte O, Zrenner E, Lancelot, M-E, Antonio A, Germain A, Michiels C, Audier C, Letexier M, Saraiva J-P, Leroy BP, Munier FL, Mohand-Said S, Lorenz B, Friedburg C, Preising M, Kellner U, Renner AB, Moskova-Doumanova V, Berger W,

Wissinger B, Hamel CP, Schorderet DF, De Baere E, Sharon D, Banin E, Jacobson SG, Bonneau D, Zalnonghi X, Le Meur G, Casteels I, Koenekoop R, Long VW, Meire F, Prescott K, de Ravel T, Simmons I, Nguyen H, Dollfus H, Poch O, Léveillard T, Nguyen-Ba-Charvet K, Sahel J-A, Bhattacharya SS, **Zeitz C** (2012) Whole-exome sequencing identifies mutations in *GPR179* leading to autosomal recessive complete congenital stationary night blindness. *Am J Hum Genet.* 90:321-330. Corresponding author: **Zeitz C**

3. Audo I, Bujakowska K, Léveillard T, Mohand-Saïd S, Lancelot M-E, Germain A, Antonio A, Michiels C, Saraiva J-P, Letexier M, Sahel J-A, Bhattacharya SS, Zeitz C (2012) Development and application of a next-generation-sequencing (NGS) approach to detect known and novel gene defects underlying retinal diseases. *Orphanet Journal of Rare diseases.* 7(1):8. Corresponding author: Audo I and **Zeitz C**

2011

1. Audo I, Bujakowska K, Mohand-Saïd S, Tronche S, Lancelot M-E, Antonio A, Germain A, Lonjou C, Carpentier C, Sahel J-A, Bhattacharya SS, **Zeitz C** (2011) A novel *DFNB31* mutation associated with Usher type 2 syndrome showing variable degree of auditory loss in a Portuguese consanguineous family. *Mol Vis.* 17:1598-1606. Corresponding author: Audo I and **Zeitz C**
2. Pieras JI, Barragán I, Borrego S, Audo I, González-del Pozo M, Bernal S, Gaget M, **Zeitz C**, Bhattacharya SS, Antiñolo G. Copy-Number variations in *Eys*. A significant event in the appearance of arRP. *Invest Ophthalmol Vis Sci.* 52:5625-5631. Corresponding authors: Antiñolo G, Bhattacharya SS.
3. Audo I, Lancelot ME, Mohand-Said S, Antoni A, Germain A, Sahel JA, Bhattacharya SS, **Zeitz C** (2011) Novel C2Orf71 mutations account for ~1% of cases in a large French arRP cohort. *Human Mut.* 32:E2091-2103. Corresponding author: Audo I and **Zeitz C**

2010

1. Barragán I, Borrego S, Pieras JI, González-del Pozo M, Santoyo J, Ayuso C, Baiget M, Millan JM, Mena M, Abd El-Aziz MM, Audo I, **Zeitz C**, Littink KW, Dopazo J, Bhattacharya SS, Antiñolo G (2010) Mutation spectrum in *EYS* in Spanish patients with autosomal recessive retinitis pigmentosa. *Human Mut.* E1772-E1800. Corresponding authors: Bhattacharya SS, Antiñolo G.
2. Riazuddin SA, Shahzadi A, **Zeitz C**, Ahmed ZM, Ayyagari R, Chavali VR, Ponferrada VG, Audo I, Michiels C, Lancelot ME, Nasir IA, Zafar AU, Khan SN, Husnain T, Jiao X, MacDonald IM, Riazuddin S, Sieving PA, Katsanis N, Hejtmancik JF (2010) A mutation

in SLC24A1 implicated in autosomal-recessive congenital stationary night blindness. *Am J Hum Genet.* 87:523-531. Corresponding Author: Riazuddin SA

3. Audo I, Friedrich A, Mohand-Saïd S, Lancelot M-E, Antonio A, Moskova-Doumanova V, Poch O, Bhattacharya SS, Sahel J-A, **Zeit** C (2010) An Unusual Retinal Phenotype Associated With A Novel Mutation in *RHO*. *Arch Ophthalmol.* 128:1036-1045. Corresponding author: Audo I
4. Audo I, Sahel J-A, Mohand-Saïd S, Lancelot M-E, Antonio A, Moskova-Doumanova V, Nandrot E, Jordan Doumanov, Barragan I, Antinolo G, Bhattacharya SS, **Zeit** C (2010) *EYS* is a major gene for rod-cone dystrophies in France. *Human Mut.* E1 406-435. Corresponding author: **Zeit** C
5. Audo I, Manes G, Mohand-Saïd S, Friedrich A, Lancelot M-E, Antonio A, Moskova-Doumanova V, Poch O, Zanlonghi X Hamel C, Sahel J-A, Bhattacharya SS, **Zeit** C (2010) Spectrum of rhodopsin mutations in French autosomal dominant rod-cone dystrophy patients. *Invest Ophthalmol Vis Sci.* 51:3687-3700. Corresponding author: **Zeit** C

2009

1. Audo I, Kohl S, Leroy BP, Munier FL, Guillonneau X, Mohand-Saïd S, Bujakowska K, Nandrot EF, Lorenz B, Preising M, Kellner U, Renner AB, Bernd A, Antonio A, Moskova-Doumanova V, Lancelot ME, Poloschek CM, Drumare I, Defoort-Dhellemmes S, Wissinger B, Léveillard T, Hamel CP, Schorderet DF, De Baere E, Berger W, Jacobson SG, Zrenner E, Sahel JA, Bhattacharya SS, **Zeit** C (2009) TRPM1 Is Mutated in Patients with Autosomal-Recessive Complete Congenital Stationary Night Blindness. *Am J Hum Genet.* 85: 720-729. Corresponding author: **Zeit** C
2. **Zeit** C, Labs S, Lorenz B, Forster U, Üksti J, Kroes HY, De Baere E, Leroy BP, Cremers FPM, Wittmer M, van Genderen MM, Sahel J-A, Audo I, Poloschek CM, Mohand-Saïd S, Fleischhauer J, Hüffmeier U, Veselina Moskova-Doumanova, Levin AV, Hamel CP, Leifert D, Munier FL, Schorderet DF, Zrenner E, Friedburg C, Wissinger B, Kohl S, Berger W. (2009) Genotyping microarray for CSNB-associated genes. *Invest Ophthalmol Vis Sci.* 50: 5919-5926. Corresponding author: **Zeit** C
3. Leroy BP, Budde B, Wittmer M, De Baere E, Berger W, **Zeit** C (2009) A common *NYX* mutation in Flemish patients with X-linked CSNB. *BJO.* 93: 692-696. Corresponding author: **Zeit** C * These authors contributed equally to this work

2. Peer-reviewed reviews:

2014

1. **Zeitz C**, Robson AG, Audo I (2014) Congenital stationary night blindness: An analysis and update of genotype-phenotype correlations and pathogenic mechanisms. *Prog Retin Eye Res.* Oct 13. [Epub ahead of print] Corresponding author: **Zeitz C**

2012

1. Bujakowska K, Audo I, Mohand-Saïd S, Lancelot M-E, Antonio A, Germain A, Léveillard T, Letexier M, Saraiva J-P, Lonjou C, Carpentier W, Sahel J-A, Bhattacharya SS, Zeitz C (2012) *CRB1* mutations in inherited retinal dystrophies. *Human Mut.* 33: 306-315. Corresponding author: Audo I and **Zeitz C**
2. Audo I, Mohand-Saïd S, Dhaenens C-M, Germain A, Orhan E, Antonio A, Hamel C, Sahel J-A, Bhattacharya SS, **Zeitz C** (2012) *RP1* and autosomal dominant rod-cone dystrophy: novel mutations, a review of published variants, and genotype-phenotype correlation. *Human Mut.* 33:73-80. Corresponding author: Audo I and **Zeitz C**

2010

3. Audo I, Bujakowska K, Mohand-Saïd S, Lancelot ME, Moskova-Doumanova V, Waseem NH, Antonio A, Sahel JA, Bhattacharya SS, **Zeitz C** (2010) Prevalence and novelty of *PRPF31* mutations in French autosomal dominant rod-cone dystrophy patients and review of published reports. *BMC Med Genet.* 11:145. Corresponding author: **Zeitz C**

