# ISABELLE AUDO –PROFESSOR- GROUP LEADER

**EDUCATION**

| INSTITUTION AND LOCATION | DEGREE | YEAR(s) | FIELD OF STUDY |
|---|---|---|---|
| Paris XI University, France | Medical school | 1987-1993 | General medical education |
| Lille University, France | Residency | 1994-1998 | Ophthalmology |
| Paris VII University, France | Diploma in clinical visual electrophysiology | 1997-1998 | Ophthalmology |
| Laboratoire de physiopathologie cellulaire et moléculaire de la rétine, Strasbourg, France | M. S. | 1998-1999 | Molecular and cellular pharmacology |
| Department of Ophthalmology, University of Madison, Wisconsin, USA | Research fellowship | 1999-2003 | Ophthalmology |
| Paris VII University, France | Statistics degree (CESAM) | 2000-2001 | Statistics for biology |
| Louis Pasteur University, Strasbourg, France | PhD | 2003 | Molecular and cellular pharmacology |
| Paris VII University, France | European Master of genetics | 2006-2007 | Genetics |
| Paris VI University, France | Habilitation to direct research | 2013 | Accreditation to supervise PhD |

**Professional experience:**

2016-present **full professor** at Sorbonne Université and the CHNO of Quinze-Vingts, Paris, France
Group leader, team S6, A to Z, Department of genetics, Institut de la Vision, Paris, France.
Coordinator of the certified biobank NeuroSensCol for genetic research in Inherited sensory disorders
Head of the Electrodiagnostic Unit, CHNO of Quinze-Vingts, Paris, France.
Head of ophthalmic pediatrics, CHNO of Quinze-Vingts, Paris, France.
Deputy coordinator of the Center for Rare Diseases "inherited retinal diseases
I Investigator at the Clinical Investigation Center CIC1423
2008 - 2016 **Assistant professor**
2005 - 2008 **Postdoctoral Fellow**, Laboratoire de physiopathologie cellulaire et moléculaire de la rétine, Paris, France.
**Consultant at the Center for Rare Diseases** "Inherited retinal diseases"
2003 - 2005 **Medical Retina Fellow**, Moorfields Eye Hospital, London, UK.
1999 - 2003 **PhD Student**, Strasbourg University, France and University of Madison, Wisconsin, USA.

**Honors and Awards:**

2014 Reward from the Eye Foundation, part of the Foundation of France, total 50 000€
07/2008-08/2013 following FFB Marjorie Carr Adams Women's Career Development Award: 08/2008 – 08/2013. Total 65K$/years the first 2 years and then 75k$/years the following
2005-2007 Foundation Fighting Blindness Career Development Award et FAUN Fondation for research in Usher syndrome.
2003-present Honorary appointment in the Genetic Department, Institute of Ophthalmology, London, UK.
2003 Ph. D. with Honors (*magna cum laude*). Thesis granted with a price from the Comité d'étude international sur les vitamines/International committee on vitamin studies.
1999-2003 Doctoral Research Fellowships from the French Academy of Medicine, the 'Fondation de France' and the Philips Foundation.
1998 M. D. with Honors (*magna cum laude*).

**Major recent Funding:**

2020-2025: « Investissements d'Avenir » IHU ForeSight IHU FOReSIGHT [ANR-18-IAHU-0001] par l'Agence Nationale de la Recherche; (450 000 EUR) " Combining massive parallel sequencing and transcriptomic data from patient-derived retinal organoids to unravel missing gene defects in inherited retinal disease " (PIs AUDO-ZEITZ)
2020: RETINA France (60 000 Eur) Pursuing the search for novel genetic defects underlying inherited retinal dystrophy.
2020-2025: Grant NIH-NEI avec mondule I: histoire naturelle de la dégénérescence rétinienne associée aux mutations sur BEST1, PI: AUDO
2018-2023 Grant NIH-NEI avec mondule I: histoire naturelle de la dégénérescence rétinienne associée aux mutations sur CNGB1, PI: AUDO, coPI ZEITZ)
2017-2019 « Investissements d'Avenir » Labex géré par l'Agence Nationale de la Recherche [réference : ANR-10-LABX-65] ; (400 000 EUR) "A comprehensive approach combining high throughput sequencing and patient transcriptomic data to unravel missing gene defects in inherited retinal disorders" (PIs AUDO-ZEITZ)
2019-2021 Foundation Fighting Blindness: Unraveling missing gene defects underlying extensively investigated IRDs through a comprehensive pipeline including disease modeling in patient-derived retinal organoids (100k$/year)
2017-2019 UNADEV-AVIESAN "Une approche innovante pour découvrir les défauts génétiques manquants dans les dystrophies rétiniennes", 200k€
2014-2019: Labex Lifesenses "Investigating the genetic bases of inherited retinal diseases", 150k€ per year
2016-2019: Retina France "Toward a better understanding of ITM2B in retinal degeneration using iPSC-disease modelling" 102k€

**Memberships**

ARVO: The Association of Research in Vision and Ophthalmology
ASHG: The American Society of Human Genetics
International Society for Clinical Electrophysiology of Vision (ISCEV).
Macular Society
ISGEDR: International Society for Genetic Eye Diseases and Retinoblastoma
European Fluorescein Angiography Club (eFAN club)
SFO: Société Française d'Ophtalmologie
SGOF : Société de Génétique Ophtalmologique Francophone from which I'm the president since October 2017

**Ad hoc reviewer**

for IOVS, Journal of Medical genetic, Plosone, Ophthalmology, Documenta in Ophthalmology, Jama Ophthalmology, Human Mutation, etc.

**Editorial boards and scientific committee:**

Orphanet Journal or Rare Diseases: section editor, Translational Vision Science and Technology, an ARVO Journal, associate editor
Scientific advisory board: Foundation Fighting Blindness, Retina France and Fondation de France, Expert group ITMO Neuroscience

**Academic duties:**

Member of the Institut de la Vision lab council
Member of the Quinze-Vingts hospital medical council
Member of the University commission for nomination of new professors and assistant professors (2016-2017)
Member of the University commission for the habilitation to direct research
Member of the University commission for authorization in cumulative activity

**Main publications within the last 2 years: H index 33**

. Mechaussier S", Almoallem B", Zeitz C, Van Schil K, Jeddawi L, Van Dorpe J, Dueñas Rey A, Condroyer C, Pelle O, Polak M, Boddaert N, Bahi-Buisson N, Cavallin M, Bacquet JL, Mouallem-Bézière A, Zambrowski O, Sahel JA, **Audo I*,** Kaplan J*, Rozet JM*, De Baere E°, Perrault I°. Loss of Function of RIMS2 Causes a Syndromic Congenital Cone-Rod Synaptic Disease with Neurodevelopmental and Pancreatic Involvement. American journal of human genetics, 2020: 106(6), 859–871. ", *,°: equal contribution

- Duncan JL, Liang W, Maguire MG, **Audo I**, Ayala AR, Birch DG, Carroll J, Cheetham JK, Esposti SD, Durham TA, Erker L, Farsiu S, Ferris FL 3rd, Heon E, Hufnagel RB, Iannaccone A, Jaffe GJ, Kay CN, Michaelides M, Pennesi ME, Sahel JA; Foundation Fighting Blindness Consortium Investigator Group. Baseline Visual Field Findings in the RUSH2A Study: Associated Factors and Correlation with Other Measures of Disease Severity. Am J Ophthalmol. 2020 May 21:S0002-9394(20)30254-3. doi: 10.1016/j.ajo.2020.05.024. Epub ahead of print.

- Nassisi M, **Audo I***, Zeitz C*, Varin J, Wohlschlegel J, Smirnov V, Santiard-Baron D, Picaud S, Sahel JA. Impact of the COVID-19 lockdown on basic science research in ophthalmology: the experience of a highly specialized research facility in France. Eye (Lond). 2020 May 7. doi: 10.1038/s41433-020-0944-7. [Epub ahead of print]

-Varin J, Reynolds MM, Bouzidi N, Tick S, Wohlschlegel J, Becquart O, Michiels C, Dereure O, Duvoisin RM, Morgans CW, Sahel JA, Samaran Q, Guillot B, Pulido JS, **Audo I**, Zeitz C. Identification and characterization of novel TRPM1 autoantibodies from serum of patients with melanoma-associated retinopathy. PLoS One. 2020 Apr 23;15(4):e0231750.

- Lee EJ, Chiang WJ, Kroeger H, Bi CX, Chao DL, Skowronska-Krawczyk D, Mastey RR, Tsang SH, Chea L, Kim K, Lambert SR, Grandjean JM, Baumann B, **Audo I**, Kohl S, Moore AT, Wiseman RL, Carroll J, Lin JH. Multiexon deletion alleles of ATF6 linked to achromatopsia. JCI Insight. 2020 Apr 9;5(7).

- Augstburger E, Sahel JA, **Audo I**. Progressive chorioretinal involvement in a patient with light-chain (AL) amyloidosis: a case report. BMC Ophthalmol. 2020 Feb 21;20(1):59.

- Strunz T, Lauwen S, Kiel C; **International AMD Genomics Consortium (IAMDGC)**, Hollander AD, Weber BHF. A transcriptome-wide association study based on 27 tissues identifies 106 genes potentially relevant for disease pathology in age-related macular degeneration. Sci Rep. 2020 Jan 31;10(1):1584.

- Jaffal L, Joumaa WH, Assi A, Helou C, Cherfan G, Zibara K, **Audo I**, Zeitz C, El Shamieh S. Next Generation Sequencing Identifies Five Novel Mutations in Lebanese Patients with Bardet-Biedl and Usher Syndromes. Genes (Basel). 2019 Dec 16;10(12).

- Nassisi M, Mohand-Saïd S, Andrieu C, Antonio A, Condroyer C, Méjécase C, Dhaenens CM, Sahel JA, Zeitz C, **Audo I**. Peripapillary Sparing With Near Infrared Autofluorescence Correlates With Electroretinographic Findings in Patients With Stargardt Disease. Invest Ophthalmol Vis Sci. 2019 Dec 2;60(15):4951-4957

- Wohlschlegel J, Letellier C, Liu B, Méjécase C, Slembrouck-Brec A, Condroyer C, Michiels C, Sahel JA, Reichman S, Zeitz C, Goureau O, **Audo I**. Generation of human induced pluripotent stem cell lines from a patient with ITM2B-related retinal dystrophy and a non mutated brother. Stem Cell Res. 2019 Dec;41:101625.

- Kimchi A, Meiner V, Silverstein S, Macarov M, Mor-Shaked H, Blumenfeld A, **Audo I**, Zeitz C, Mechoulam H, Banin E, Sharon D, Yahalom C. An Ashkenazi Jewish founder mutation in CACNA1F causes retinal phenotype in both hemizygous males and heterozygous female carriers. Ophthalmic Genet. 2019 Oct;40(5):443-448.

. Nassisi M, Mohand-Saïd S, Andrieu C, Antonio A, Condroyer C, Méjécase C, Varin J, Wohlschlegel J, Dhaenens CM, Sahel JA, Zeitz C, **Audo I**. Prevalence of *ABCA4* Deep-Intronic Variants and Related Phenotype in An Unsolved "One-Hit" Cohort with Stargardt Disease. Int J Mol Sci. 2019 Oct 11;20(20). pii: E5053.

. Boulanger-Scemama E, Mohand-Saïd S, El Shamieh S, Démontant V, Condroyer C, Antonio A, Michiels C, Boyard F, Saraiva JP, Letexier M, Sahel JA, Zeitz C, **Audo I**. Phenotype Analysis of Retinal Dystrophies in Light of the Underlying Genetic Defects: Application to Cone and Cone-Rod Dystrophies. Int J Mol Sci. 2019 Sep 30;20(19). pii: E4854

. Khateb S, Mohand-Saïd S, Nassisi M, Bonnet C, Roux AF, Andrieu C, Antonio A, Condroyer C, Zeitz C, Devisme C, Loundon N, Marlin S, Petit C, Bodaghi B, Sahel JA, **Audo** I. PHENOTYPIC CHARACTERISTICS OF ROD-CONE DYSTROPHY ASSOCIATED WITH MYO7A MUTATIONS IN A LARGE FRENCH COHORT. Retina. 2019 Aug 29. [Epub ahead of print]

. Augstburger E, Orès R, Mohand-Said S, Mrejen S, Keilani C, Antonio A, Condroyer C, Andrieu C, Sahel JA, Zeitz C, **Audo I**. Outer retinal alterations associated with visual outcomes in Best vitelliform macular dystrophy. Am J Ophthalmol. 2019 Dec;208:429-437.

. Khateb S, Nassisi M, Bujakowska KM, Méjécase C, Condroyer C, Antonio A, Foussard M, Démontant V, Mohand-Saïd S, Sahel JA, Zeitz C, **Audo I**. Longitudinal Clinical Follow-up and Genetic Spectrum of Patients With Rod-Cone Dystrophy Associated With Mutations in PDE6A and PDE6B. JAMA Ophthalmol. 2019 Jun 1;137(6):669-679.

. Méjécase C, Hummel A, Mohand-Saïd S, Andrieu C, El Shamieh S, Antonio A, Condroyer C, Boyard F, Foussard M, Blanchard S, Letexier M, Saraiva JP, Sahel JA, Zeitz C, **Audo I**. Whole exome sequencing resolves complex phenotype and identifies CC2D2A mutations underlying non-syndromic rod-cone dystrophy. Clin Genet. 2019 Feb;95(2):329-333

. Nassisi M, Mohand-Saïd S, Dhaenens CM, Boyard F, Démontant V, Andrieu C, Antonio A, Condroyer C, Foussard M, Méjécase C, Eandi CM, Sahel JA, Zeitz C, **Audo I**. Expanding the Mutation Spectrum in ABCA4: Sixty Novel Disease Causing Variants and Their Associated Phenotype in a Large French Stargardt Cohort. Int J Mol Sci. 2018 Jul 27;19(8).

. Orès R, Mohand-Said S, Dhaenens CM, Antonio A, Zeitz C, Augstburger E, Andrieu C, Sahel JA, **Audo I**. Phenotypic Characteristics of a French Cohort of Patients with X-Linked Retinoschisis. Ophthalmology. 2018 Oct;125(10):1587-1596.

. **Audo I**, Mohand-Said S, Boulanger-Scemama E, Zanlonghi X, Condroyer C, Démontant V, Boyard F, Antonio A, Méjécase C, El Shamieh S, Sahel JA, Zeitz C. MERTK mutation update in inherited retinal diseases. Hum Mutat. 2018 Jul;39(7):887-913. doi: 10.1002/humu.23431.

. Matet A, Kohl S, Baumann B, Antonio A, Mohand-Said S, Sahel JA, **Audo I**. Multimodal imaging including semiquantitative short-wavelength and near-infrared autofluorescence in achromatopsia. Sci Rep. 2018 Apr 4;8(1):5665.

. Boulanger-Scemama E, Sahel JA, Mohand-Said S, Antonio A, Condroyer C, Zeitz C, **Audo I**. AUTOSOMAL DOMINANT VITREORETINOCHOROIDOPATHY: When Molecular Genetic Testing Helps Clinical Diagnosis. Retina. 2019 May;39(5):867-878.

. Méjécase C, Mohand-Saïd S, El Shamieh S, Antonio A, Condroyer C, Blanchard S, Letexier M, Saraiva JP, Sahel JA, **Audo I**, Zeitz C. A novel nonsense variant in REEP6 is involved in a sporadic rod-cone dystrophy case. Clin Genet. 2018 Mar;93(3):707-711