Research Profiles: ***Bernd Wissinger***

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Date of birth: 13.02.1962, Boxberg/Baden, Germany

Academic education and professional experience:

1982 - 1987	Undergraduate studies in Biology at the Eberhart-Karls-University of Tübingen
1986 - 1987	Diploma-thesis in the lab of PD Dr. Axel Brennicke at the Dept. of Botany, Eberhart-Karls-University of Tübingen
1988 - 1991	Promotion (summa cum laude) at the Faculty of Biology, Eberhart-Karls-University of Tübingen. Ph. D. thesis in the lab of Prof. Dr. Axel Brennicke at the Institut für Genbiologische Forschung mbH, Berlin-Dahlem
1991	Postdoctoral research fellow at the the Institut für Genbiologische Forschung mbH, Berlin-Dahlem
1992 - 1993	Research Fellow at the Forschunggstelle für Experimentelle Ophthalmologie, Universitäts-Augenklinik Abt.II, Tübingen (Ärztl. Direktor: Prof. Dr. Eberhart Zrenner)
since 1994	Head of the Molekulargenetisches Labor, Universitäts-Augenklinik Abt.II, Tübingen, (Ärztl. Direktor: Prof. Dr. Eberhart Zrenner)

Relevant awards and fellowships, major administrative and board activities

1988 - 1990	Stipendship of the Graduiertenförderung Baden-Württemberg
1999 – 2001	Coordinator of the States' Research Program: „Fundamentals in Color Vision - A multidisciplinary Approach "
2003	Offer for a position (C3 Assistant Professorship) as Research Group Leader at the Max-Planck-Institut für Molekulare Genetik, Berlin (refused)
since 2005	Coordinator of the Clinical Research Group „Hereditary Retinal Disorders: Clinics, Genetics and Animal Models "

Other Scientific Functions

Reviewer for numerous scientific journals: Proc Natl Acad Sci USA, Hum Mol Genet, Am J Hum Genet, Invest Ophthalmol Vis Sci, Genomics, and others

Reviewer for several scientific agencies: Fight-for-sight Foundation, Wellcome Trust, Israel Science Foundation, and others

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Main Research topics:

- Molecular Genetics of Hereditary Retinal Disorders and Optic Neuropathies
- Genetics of Colour Vision Deficiencies
- Physiology of Phototransduction
- Biology and Development of Cone Photoreceptors
- Transcription and Transcriptional Regulation in the Retina
- Animal Models for Retinal Disorders
- Mitochondrial Biology and Mitochondriopathies

Current Research Funding

Deutsche Forschungsgemeinschaft, European Union (6^{th} framework program), Thyssen Foundation, Hildebrandt Foundation

Selected publications

Kohl S, Marx T, Giddings I, Jägle H, Jacobson S, Apfelstedt-Sylla E, Zrenner E, Sharpe LT, Wissinger B (1998) Total color blindness is caused by mutations in the gene encoding the α-subunit of the cone photoreceptor cGMP gated cation channel. **Nature Genet** 19:257-259.

Kohl S, Baumann B, Broghammer M, Jägle H, Sieving P, Kellner U, Spegal R, Anastasi M, Zrenner E, Sharpe LT, Wissinger B (2000) Mutations in the *CNGB3* gene encoding the β-subunit of the cone photoreceptor cGMP gated channel are responsible for Achromatopsia (*ACHM3*) linked to chromosome 8q21. **Hum Mol Genet** 9:2107-2116.

Alexander C, Votruba M, Pesch U, Thiselton D, Mayer S, Moore T, Rodriguez M, Kellner U, Leo-Kottler B, Auburger G, Bhattacharya S, Wissinger B (2000) OPA1, a gene encoding a dynamin-related GTPase, is mutated in autosomal dominant optic atrophy linked to chromosome 3q28. **Nature Genet** 26:211-215.

Jacobi FK, Leo-Kottler B, Mittelviefhaus K, Zrenner E, Meyer J, Pusch CM, Wissinger B (2001) Segregation patterns and heteroplasmy prevalence in Leber's hereditary optic neuropathy. **Invest Ophthalmol Vis Sci** 42:1208-1214.

Pesch UEA, Leo-Kottler B, Mayer S, Jurklies B, Kellner U, Apfelstedt-Sylla E, Zrenner E, Wissinger B (2001) OPA1 mutations in patients with autosomal dominant optic atrophy and evidence for semi-dominant inheritance. **Hum Mol Genet** 10:1359-1368.

Kohl S, Baumann B, Rosenberg T, Kellner U, Lorenz B, Vadala M, Jacobson SG, Wissinger B (2002) Mutations in the cone photoreceptor G-protein alpha-subunit gene *GNAT2* in patients with achromatopsia. **Am J Hum Genet** 71: 422-425.

Pesch UEA, Fries JE, Bette S, Kalbacher H, Wissinger B, Alexander C, Kohler K (2004) Opa1, the disease gene for autosomal dominant optic atrophy, is specifically expressed in ganglion cells and intrinsic neurons of the retina. **Invest Ophthalmol Vis Sci** 45:4217-4225.

Bette, S., Mittelbronn, M., Schlaszus, H., Wissinger, B., Meyermann, R. (2005) OPA1, associated with autosomal dominant optic atrophy is widely expressed in the human brain. **Acta Neuropathol (Berl)**. 109(4): 393-399.

Schimpf, S., Schaich, S., Wissinger, B. (2005) Identification of mutations in exon and intron sequences of the OPA1 gene involving the activation of cryptic splice sites. **Hum Genet** 118(6):767-771.

Dadgar, S., Hagens, O., Dadgar, S.R., Haghighi, E.N., Schimpf, S., Wissinger, B., Garshasbi (2006) Structural modell of the OPA1 GTPase domain may explain the molecular consequences of a novel mutation in in family with autosomal dominant optic atrophy. **Exp Eye Res** 83(3): 702-706.

Alavi, M., Bette, S., Schimpf, S., Schüttauf, F., Schraermeyer, U., Wehrl, H.F., Rüttiger, L., Beck, S.C., Tonagel, F., Pichler, B.J., Knipper, M., Peters, T., Laufs, J., Wissinger, B. (2007) A splice site mutation in the murine *OPA1* features pathology of autosomal dominant optic atrophy. **Brain** 130: 1029-1042.

Schimpf, S., Fuhrmann, N., Schaich, S., Wissinger, B. (2007) A comprehensive cDNA study and quantitative transcript analysis of OPA1 mutations associated with autosomal dominant optic neuropathy. **Hum Mut** (re-submitted)