

Current Research Topics

- Elucidation of the genetic basis of and identification of new genes for inherited ocular disorders with emphasis on hereditary retinal dystrophies and optic neuropathies
- The molecular basis of color vision and color vision deficiencies
- Functional analysis of mutant gene products, notably the cone CNG channel and the retinal phosphodiesterase by means of biochemical and cell biology methods and the development of bioassays to evaluate pharmaco-therapeutic approaches
- Characterization of the pathophysiological processes of degenerative retinal diseases and retinal mitochondriopathies at the cellular and system level
- Generation and characterization of animal models and reporter systems (mouse and zebrafish) for retinal degenerative diseases
- The quantitative genetics of retinal gene expression

Current Projects:

- Blue Cone Monochromacy
- DFG Trilateral - Genetic disorders in Arab societies of Israel and the Palestinian Authority
- ERMION - European Research Project on Mendelian Inherited Optic Neuropathies
- EyeTN - a Marie Curie Initial Training Network
- HOPE2 - Hereditary Retinal Disorders: From Patients towards Therapies
- IonNeurONet - German Network of Neurological and Ophthalmological Ion Channel Disorders
- Pharmacogenomics in Glaucoma

Institute for Ophthalmic Research Molecular Genetics Laboratory

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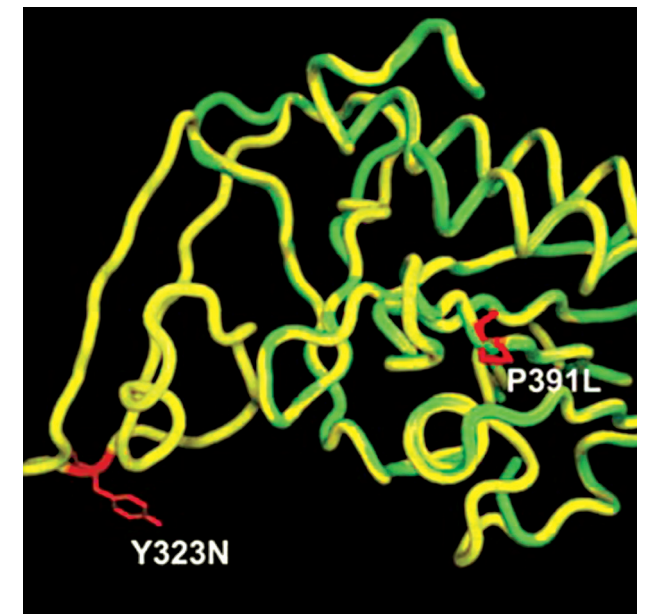
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How to find uns:



Wissinger Lab Molecular Genetics Laboratory





A large proportion of ocular conditions and ocular diseases in humans is governed or at least strongly influenced by genetic factors. In fact, the eye is amongst the organs that are most commonly afflicted by inherited disease. Our mission is “To uncover the genetic basis of inherited ocular disorders and to explain its clinical appearance and characteristics by constructing gene-to-function relationship at the molecular, cellular and physiological level”.

The Molecular Genetics Laboratory (MGL) at the Institute for Ophthalmic Research has been found-

ed as early as 1991. One year later a biobank and patient database has been established, and we have collected DNA and files of patients and family members with inherited ocular diseases, with a strong focus on inherited retinal disorders, hereditary optic neuropathy and familial glaucoma.

Today the biobank holds more than 23.000 DNA samples of >4000 families with inherited retinal dystrophies, >630 families with autosomal dominant or sporadic optic atrophy, >1150 families with Lebers hereditary optic neuropathy and >1100 families with hereditary glaucoma.



Bernd Wissinger

- Professor, Dr. rer. nat.
- Head of the Lab for Molecular Genetics of Sensory Systems



Susanne Kohl

- Dr. rer. nat.
- Deputy head of the Lab for Molecular Genetics of Sensory Systems

Important Publications

- Wei T, Schubert T, Paquet-Durand F, Tanimoto N, Chang L, Koeppen K, Ott T, Griesbeck O, Seeliger M, Euler T, Wissinger B (2012) Light driven calcium signals in mouse cone photoreceptors. *J Neurosci* 32: 6981-6994.
- Achilli A, Iommarini L, Olivieri A, Pala M, Kashani BH, Reynier P, La Morgia C, Valentino ML, Liguori R, Pizza F, Barboni P, Sadun F, De Negri AM, Zeviani M, Dollfus H, Moulignier A, Ducos G, Orssaud C, Bonneau D, Procaccio V, Leo-Kottler B, Fauser S, Wissinger B, Amati-Bonneau P, Torroni A, Carelli V (2012) Rare primary mitochondrial DNA mutations and synergistic variants in Leber's Hereditary Optic Neuropathy. *PLoS ONE* 7(8):e42242.
- Kohl S, Coppieters F, Meire F, Schaich S, Roosing S, Brennenstuhl C, Depasse F, Bolz S, Lukowski R, den Hollander AI, Cremers FPM, de Baere E, Hoyng C, Wissinger B (2012) Autosomal recessive achromatopsia caused by a homozygous nonsense mutation in the PDE6H gene, the gene encoding the α -subunit of the cone cGMP phosphodiesterase. *Am J Hum Genet* 91: 527-532.

Research toSee

The Institute for Ophthalmic Research

The Institute for Ophthalmic Research is headed by Prof. Marius Ueffing and cooperates closely with the University Eye Hospital (Prof. Karl-Ulrich Bartz-Schmidt) under the common roof of the Centre for Ophthalmology in order to perform translational research.

The Institute aims at uncovering the causes for degenerative, inflammatory and vascular diseases of the eye and the visual pathways at molecular, cellular and systemic levels.

The Institute houses several teams of scientists who work together to develop and evaluate concepts for therapy and treatment and optimise clinical and research diagnostics.

Thus, the Institute provides an efficient infrastructure which supports research and education and mediates contacts to other research institutions and to industry.

The Institute enjoys not only a variety of national and international scientific activities, like intense partnerships and cooperations, but also offers courses and seminar opportunities to students and young researchers.

The list of publications and sponsors are the evidence for the success of its activities.