… performing in Excellence

Current Research Topics

- Elucidation of the genetic cause of and identification of novel 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 OPA1 by means of biochemical and cell biology methods
- Characterization of the pathophysiological processes of degenerative retinal diseases and retinal mitochondriopathies at the cellular and systems level
- Generation and characterization of animal models and in vivo reporter systems for retinal degenerative disease
- Development of genetic therapies for retinal dystrophies and optic neuropathies

Current Projects:

- Genetics and molecular disease mechanisms in Blue Cone Monochromacy (BCM)
- DFG Trilateral - Genetic disorders in Arab societies of Israel and the Palestinian Authority
- EyeTN - a Marie Curie Initial Training Network
- RD-CURE - Bringing Gene Supplementation Therapy for Inherited PDE6A- and CNGA3-associated retinopathies into Clinical Practise
- Splicing defects and therapeutic rescue of splicing defects in inherited ocular disease

Contact

Institute for Ophthalmic Research
Molecular Genetics Laboratory

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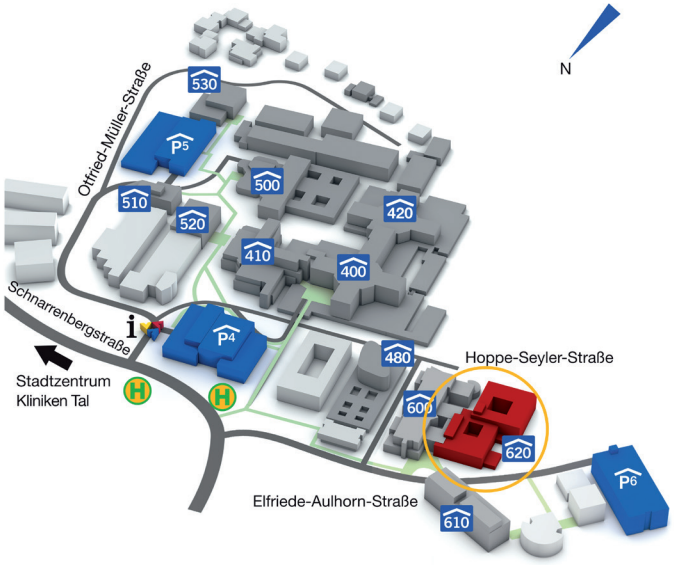

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Wissinger Lab
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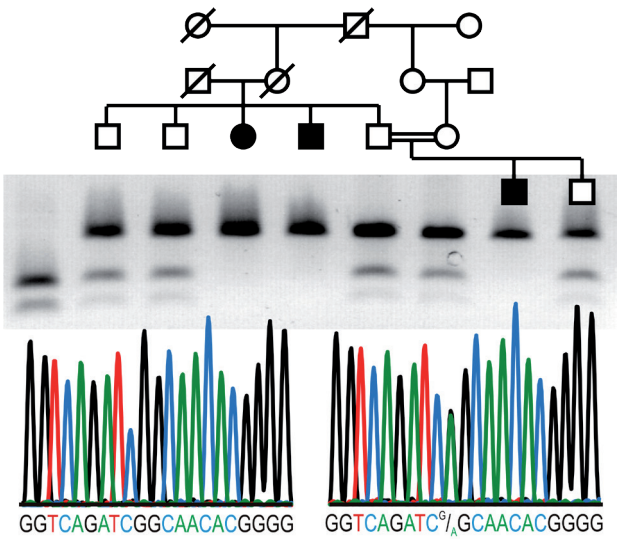

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A large proportion of ocular conditions and ocular diseases in humans is governed or at least strongly influenced by genetic factors. In fact, the eye is amongst the organs that are most commonly afflicted by inherited disease. The mission of the Molecular Genetics Laboratory (MGL) at the Institute for Ophthalmic Research 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 MGL hosts and maintains a large research biobank and patient database that contain DNA samples and medical genetic data files from patients and family members with inherited ocular diseases, with a strong focus on inherited retinal disorders, hereditary optic neuropathy and familial glaucoma. Currently the biobank archives more than 28.000 DNA samples and serves as a rich resource to study the genetic basis of ocular disease. In fact, samples from the MGL biobank have been key for the first description of more than 20 ocular disease genes and also initiated and stimulated further research in the group towards molecular mechanisms of disease, the generation and study of model systems, and the development of therapeutic strategies.

Bernd Wissinger

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

Susanne Kohl

- Dr. rer. nat.
- Deputy head of the Molecular Genetics Labratory

Recent Publications

- Bonifert T, Karle K, Tonagel F, Batra M, Wilhelm C, Theurer Y, Schoenfeld C, Kluba T, Kamenisch Y, Carelli V, Schicks J, Gonzalez M, Speziani F, Schüle R, Züchner S, Schöls L, Wissinger B, Synofzik M (2014) Pure and Syndromic Optic Atrophy Explained by Deep Intronic OPA1 mutations and an Intralocus Modifier. Brain 137: 2164-77
- Kohl S, Zobor D, Chiang WCJ, Weisschuh N, Staller J, Gonzalez Menendez I, Chang S, Beck SC, Garcia Garrido M, Sothilingam V, Seeliger MW, Stanzial F, Benedicenti F, Inzana F, Heon E, Vincent A, Beis J, Strom TM, Rudolph G, Roosing S, den Hollander AI, Cremers FPM, Lopez I, Ren H, Moore AT, Webster A, Michaelides M, Koenekoop RK, Zrenner E, Kaufman RJ, Tsang SH, Wissinger B, Lin JH (2015) Mutations in the Unfolded Protein Response regulator, ATF6, cause the cone dysfunction syndrome Achromatopsia. Nature Genetics 47(7):757-65.
- Buena-Atienza E, Rüther K, Baumann B, Bergholz R, Birch D, De Baere E, Dollfus H, Greally MT, Gustavsson P, Hamel CP, Heckenlively JR, Leroy BP, Plomp AS, Pott JWR, Rose K, Rosenberg T, Stark Z, Verheij JBGM, Weleber R, Zobor D, Weisschuh N, Kohl S, Wissinger B (2016) De novo intrachromosomal gene conversion from OPN1MW to OPN1LW in the male germline results in Blue Cone Monochromacy. Scientific Reports 6: 28253

Research to See

The Institute for Ophthalmic Research

Seeing is an essential part of human life. As a leading centre for vision research we conduct rigorous research in order to break new ground in understanding the principles of vision and the mechanisms of blinding diseases. We are confident that this research will enable us to rationally develop effective treatments that ultimately retain or restore vision.

Within the Center for Ophthalmology at the University of Tübingen Medical Centre, we and our colleagues at the University Eye Hospital jointly strive for scientific excellence, for speed in translating the advancements into patient's benefit, and for training and mentoring the next generation of leaders in our field.

As leaders and partners in multi-national collaborations, we work for continuous strengthening our ties to fellow international scientists in the public and private sector and to foundations, industry and patient organizations.

As an integral part of Tübingen's biomedical and neuroscience campus, we offer a scientific environment that favors creativity for generating groundbreaking ideas, their transfer into reality and their translation into diagnostics and therapy to help those that suffer from vision loss.