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

Institute for Ophthalmic Research
Molecular Genetics Laboratory

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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.

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Recent Publications
