Understanding and reprogramming developmental visual disorders: from anophthalmia to cortical impairments, (ImprovVision)

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Diseases of vision can originate in genetic defects, that affect the embryonic development of the various components of the “visual system”: the eyes, their connection to the brain by nerves, and the brain areas that process visual information and convert it into a mental image of what we see. How genes shape our visual system, and how their malfunctioning leads to neurodevelopmental diseases of vision, is very poorly understood, and is the general question addressed by our Project. Our laboratories generated mutant mouse strains, in which specific genes are mutated, that allow to study in detail the specific contribution of individual genes to visual development. We address, in particular, genes encoding transcription factors, proteins that control the activity of many genes in parallel that, altogether, give rise to the organized development of the visual system. We already know about the importance of these genes, because their mutation impairs visual system development in mice and in patients and discovered that the genes we study are functionally connected within a “gene regulatory network”. We will use cell cultures, mouse, zebrafish models and human samples to investigate the full consequences that genetic defects already known to cause NDVD have on the rest of the brain, thus determining the full extent of visual abnormalities. This knowledge will be then translated to patients and further applied to evaluate if cell reprogramming at postnatal stages could improve visual function in models of neurodevelopmental diseases of vision. We are confident that, if successful, the ambitious goal of *ImprovVision* will lead the field of vision disease beyond the current state of the art.