Retinal dystrophies are a heterogeneous group of inherited diseases that lead to a progressive, severe and irreversible loss of vision by altering the anatomy and/or function of the retina. There is currently no causative cure, but research is being carried out to find ways of treating it with gene and cell therapies.

The retina is the part of the eye that is responsible for detecting light and turning it into visual signals that are then conducted to brain. The two type of photoreceptor cells, rods and cones are the beginning part of the visual pathway. These cells possessed very complicated and advanced structures and a cilium is one of them. Since retinal dystrophies pathogenesis is mostly related to photoreceptors' cilium, they are often called retinal ciliopathies. These diseases can cause damage to the photoreceptor cells, either, predominantly, the cones (responsible for detailed vision and color), rods (responsible for night and peripheral vision) or both at the same time. The most common retinal ciliopathy, Retinitis Pigmentosa, with prevalence of 1:3500, is the leading cause of inherited blindness worldwide, affecting upto 2.5 million people, very commonly since childhood, leading to irreversible disability. The majority of retinal ciliopathies are diseases located exclusively in the eye, but can sometimes be associated with extraocular manifestations (like Usher syndrome, Bardet-Biedl syndrome), in which case they are referred to as multi-syndromic retinal ciliopathies. Retinal ciliopathies are often not evident until their advanced stages, when their symptoms become apparent. The genetic information contained in the DNA of every person determines whether a retinal ciliopathy will develop, and, consequently, these hereditary conditions cannot be prevented. However, the modern science attempt to edit genetic information in our DNA to repair or erase damaged genes. In retinal ciliopathies, the symptoms of disease can be unfortunately determined by more than one gene, which makes the gene therapy approach difficult. These different genes involved into pathogenesis of retinal ciliopathies code certain proteins, which interact with each other's. Identifying the interactions between these proteins may help to understand complex mechanisms related to retinal ciliopathies and to propose a novel causative treatment.

The aim of the project is to experimentally simulate the retinal ciliopathy symptoms in cultured cells and in animal model by introducing mutant proteins, and to track interactions of these proteins with other protein networks. After we will recognize which interactions are altered, we hope to be able to propose targeted treatment that will repair protein network and alleviate symptoms of the disease. We hypothesize that although there are different genes involved in the pathogenesis of retinal ciliopathies, they may have common protein interactions, which would become one target for treatment of these diverse diseases. The project will employ the most advanced tools in proteomic and experimental ophthalmology research and will involve skilled researchers from both disciplines.