“I have retinal degeneration. Are there any treatment options? Is it possible to stop the progress of this disease? I heard that it is hereditary, what is the risk that my child will also suffer from it?” These questions are posted on many social media, internet fora and mutual aid groups for blind or visually impaired persons. Psychological stress associated with eye diseases, especially degenerative ones, is enormous. At present, Polish ophthalmology does not have much to offer to these patients. **In practice, people suffering from degenerative diseases of the retina do not have access to genetic diagnosis, due to the high cost of the methods used in Poland at this point.** Moreover, these tests are based on genetic changes identified in populations of the West, which probably do not match the genetic profile of Polish patients. Currently, many clinical trials involving gene therapy for hereditary retinal diseases are conducted worldwide (e.g. for Leber congenital amaurosis or Stargardt disease), and many therapies are still being tested on animals. Admission to these programs, however, requires proof of carrying a mutation in a particular gene that is the targeted by the treatment.

Since over 120 genes were up to now associated with non-syndromic hereditary retinal disorders, standard diagnostic methods are not suitable for detecting changes in all genes. In recent years, a new method of detecting such mutations has revolutionised the diagnosis of genetic diseases worldwide. The technique, termed “next-generation sequencing”, enables the identification of the causative mutation in the patient by looking at many genes at a time. **In our study, we plan to use this technology to build specific genetic profile of Polish individuals suffering from degenerative diseases of the retina** and to determine the frequency of identified mutations in Polish population. Statistically, in 20% of patients suffering from retinal diseases the genetic cause is yet unknown. **In some patients in whom causative genetic changes cannot be detected, we are planning on searching for new genes involved in these diseases. In the future, the results if this research will serve to develop a cost-effective diagnostic plan.**