The Israeli Consortium for Inherited Retinal Diseases by Professor Dror Sharon

Vision is definitely the most important sense of humans in the modern world, and indeed, the human eye is a complex organ, and even Charles Darwin, in his famous book "The Origin of Species" struggled a lot with the question how did the eye, with it versatile and complex forms, has evolved. The most important ocular tissue is the retina, which is a neuronal tissue, containing the cells responsible for light reception, called photoreceptors. These cells convert light to electric signals that are undergoing preliminary processing in the retina, and are being transferred to the brain via the optic nerves in order to create visual perception.
The photoreceptors are under heavy metabolic pressure, and therefore any damage to their function may cause them to degenerate, a process which by its nature is irreversible. Degeneration of a large number of photoreceptor causes significant damage to the activity and structure of the retina, and may lead to blindness. Over the last 25 years, numerous genetic mutations (over 250) were identified as the cause of hereditary retinal diseases, in which vision is dramatically affected, sometimes even causing in blindness. The involvement of numerous genes in these diseases complicates both identification of the causative gene and the development of treatment (whether for delaying deterioration or even preventing the disease). Lirot ("to see") Association is the leading association in Israel that is focused on prevention of blindness as its main goal, and acts in several areas in order to achieve this goal. One of the main project of the association is the establishment of the Israeli Consortium for Iherited Retinal Diseases, combined of ophthalmologists who are experts in retinal diseases, retinal disease researchers, centers for human molecular genetics, electrophysiological diagnostic centers and a bioinformatics laboratory (belonging to twelve academic centers in Israel), in order to map inherited retinal diseases in the Israeli population. This project is unique, and only recently a similar consortium was reported to have been established in Ireland. The Israeli consortium centers work in tight collaboration, and its main focus is recruiting to the genetic study the majority of families with these diseases, and identify the genetic factors for retinal diseases in the Israeli population. So far over 2000 families were recruited for the study, and additional 250 families join every year. The recruited patients sign a research participation consent form, undergo a thorough clinical and electrophysiological diagnostic in order to accurately determine the type of the disease, provide (and sometimes their family members as well) a blood sample, and research genetic analysis is performed in order to identify the gene causing the disease. As part of this research, many genes that cause inherited retinal diseases in Israel were identified, as well as three novel genes that were recently reported by the members of the consortium as causing retinal diseases.
These days the consortium members act to establish a national database which will enable to perform research more efficiently and also to check a possible correlation between the gene causing the disease and its severity (genotype – phenotype correlation). Moreover, the database will be important towards the next phase of the study during which various treatments will be tested on the various disease types. Since the diseases are genetically and clinically complex, we anticipate that based

on disease type and severity, different treatments will be provided, similar to personalized medicine modalities that become common in treating other diseases in humans. Already today there is experimental treatment for some types of retinal diseases, including gene therapy, stem cells treatment and nutritional treatment. The existence of such a database will enable a quick identification of all patients compatible with a certain treatment, whether it was developed by the consortium or by other institutions around the globe, and to measure the efficiency of the treatment on those patients.

This unique research is the pinnacle of Lirot Association's activity, which established it together with the US Foundation Fighting Blindness, recognizing the many advantages in Israel for eyes medicine and research, and which has been providing financial support to this research for the last three years. Lirot Association is diligently working this days to obtain additional support for this research from Israeli sources, such as the Israeli Ministry of Health as well as private and institutional benefactors interested in promoting blindness prevention research, in order to make the most of this important collaboration, which will enable many patients to see.