

European Society of Ophthalmology



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Project Abstract

The European Society of Ophthalmology requests a brief abstract of the project that each EuLDP participants has worked on during the program. The compiled abstracts will be included on the SOE website and will be given to incoming EuLDP classes as background material. Please e-mail your abstract to E-mail: secretariat@soevision.org

*Please include the headings if appropriate: **Title, Purpose, Methods, Results and Conclusion***

Title of Project:

Setting up a register of patients with inherited retinal dystrophies in Hungary

Purpose:

Inherited retinal dystrophy (IRD) is a group of rare diseases with genetic background. The clinical features as well as the molecular genetic changes show heterogeneity: there are more than 400 genes involved. Until recently, there was almost no therapeutic options, but evolving gene therapies make it crucial to set up proper clinical and genetic diagnosis of these patients.

Methods:

The first phase of this work was to design the spreadsheet used in the database – which clinical and molecular data to have as obligatory / optional data. The aim was to make a simple but detailed, well-searchable sheet. The second phase is to build up a secure IT platform (server, dedicated access) to handle the data. The third part (parallel with the first two phases) is the negotiation with the university centers about sharing the (anonymized) data.

Results:

The spreadsheet was designed using the most important papers and our own 20 years' experience in the field. The platform is now being set up in cooperation with IT experts. The third part of the project is carried out by organizing symposia in the topic during national Ophthalmological meetings.

Conclusion:

Setting up a national register is challenging and needs proper planning, cooperation with experts in various fields as well as colleagues around the country.