Jana Sajovic

Genomic and transcriptomic elucidation of persons with Stargardt disease.

Short Bio	I am a medical doctor, and I graduated in 2019 at the Faculty of Medicine, University of Ljubljana, Slovenija. After my graduation, I decided to start a PhD study in the interdisciplinary doctoral programme in Biomedicine at the University of Ljubljana. While doing my PhD, I am also employed as a young researcher at Eye Hospital, University medical centre Ljubljana, Slovenia.
Home Institution	Eye Hospital, University medical centre Ljubljana, Ljubljana, Slovenia
Host Institution	University Radboud University Medical Cente, Department of Human Genetics, Nijmegen, The Netherlands
Project description	Causal ABCA4 variants were identified in the majority of persons with autosomal recessive STGD. Until recently, in 20-35% of STGD cases, no or only one variant was identified in the protein-coding elements of ABCA4. Some missing mutations were found in the intronic regions that do not encode parts of the ABCA4 protein, and which constitute 95% of the ABCA4 gene. Identification of all causal variants on both alleles is important as clinical trials are under development, some of which are mutation-specific. Genetic analysis of the ABCA4 gene, finding known, and new mutations are, therefore, the main aim of our project. A considerable part for myself is the analysis of approximately 70 patients with STGD and STGD-like cases from Slovenia. Findings will give us valuable information on how mutations are distributed in the Slovenian population, which is only 2 million.
Personal statement	The EJP fellowship at Radboud University Medical Center in Nijmegen is a great asset for my further research and PhD study. It represents a significant impact on my academic growth and career opportunities as a future medical doctor. From the analysis of 70 samples from Slovenian STGD and STGD-like patients, a pool of new data will be provided, from which we will analyse Slovenian patients epidemiologically and check what their genotype-phenotype correlations are. This will be a significant achievement for University Eye Hospital in Ljubljana. The data will be presented at many meetings and published in scientific journals, which will engage various ophthalmologists at my home institution, and raise the awareness for rare genetic eye diseases. What is more, my training in Nijmegen will also be a huge achievement for other Slovenian

	scientists and doctors as I will be able to transfer the advanced methods, learnt in the Netherlands, into Slovenian space.