Streszczenie w języku angielskim

Title: Analysis of photoreceptor morphology in rare inherited retinal dystrophies, including Stargardt disease and cone-rod dystrophy – a 6-year observation with the use of adaptive optics

Background: Inherited retinal dystrophies (IRDs) are genetic disorders recognized as rare diseases. They lead to progressive, bilateral and irreversible vision loss. Stargardt disease (STGD), cone-rod dystrophy (CRD) and cone dystrophy (CD) are conditions affecting primarily macula.
Visualization of IRDs tends to be challenging. In many cases, it is not precise enough to state a diagnosis and to monitor the progression. Adaptive optics is a new non-invasive imaging technique enabling to visualize single photoreceptors.

Objectives: The aim of this dissertation was to determine characteristic features of macular cones in inherited retinal dystrophies with the use of adaptive optics, and to compare them with healthy eyes. Another aim was to track the progression of cone changes in inherited retinal degenerations in 6-year observation.

Methods: The cross-sectional study comprised of 36 eyes with STGD, 8 eyes with CRD and 14 healthy controls. The study was held on 9 eyes with CD as well.
6-year observation was conducted in 38 eyes with STGD, 8 eyes with CRD and also in 10 eyes with CD.
Cone parameters were evaluated, such as density (DM), spacing (SM), regularity (REG) and Voronoi analysis of hexagonal cells (N%6). The parameters were correlated with best corrected visual acuity (BCVA). An analysis to identify the factors that increase the probability of incomplete data acquisition was performed.
The images were obtained with the use of adaptive optics camera Rtx1™ (Imagine Eyes, France).

Results: There were significant differences in the DM, SM, REG, and N%6 parameters between the healthy and IRD-affected eyes ($p<0.001$ for DM, SM, and REG; $p=0.008$ for N%6).
No correlation was found between DM, SM and REG parameters compared to age, sex and BCVA in the study group ($p>0,05$).
DM, SM and BCVA changed significantly over a 6-year observation period ($p<0,001$ for DM, $p<0.001$ for SM, $p=0.001$ for BCVA). The decrease in the DM and increase in the SM parameters were significantly higher in females than in males ($p=0.025$ for DM, $p=0.021$ for SM).
There was a correlation found between DM change and SM change ($p<0.001$), which stems from the definition of SM and DM and between DM change and REG change ($p=0,036$). DM deterioration was not correlated with BCVA change ($p=0.847$), age ($p=0.223$) nor from initial DM ($p=0.302$), initial SM ($p=0.231$) and initial REG ($p=0.276$).
The mean DM and SM differed significantly between the diagnoses of STGD, CRD and CD ($p=0.006$ for DM, $p=0.002$ for SM), which has not been confirmed in following analyses. There was no significant difference noted between photoreceptor parameters (REG and N%6) and the diagnoses.
Progression rate in DM, SM and REG did not differ significantly in the analysis between STGD, CRD and CD ($p=0,338$ for DM, $p=0,308$ for SM, $p=0,475$ for REG).

Complete data acquisition was less frequent in eyes with lower DM ($p=0.004$) and higher SM ($p=0.013$).

Conclusions: The results outlined above lead to conclusion that photoreceptors' morphology in rare inherited retinal dystrophies differ from the ones in healthy eyes. Longitudinal analysis shows progression of degeneration in 6-years observation.
The change in cones' parameters is not correlated with best corrected visual acuity.
Low cone density and high cone spacing are risk factors for incomplete data acquisition.
Data obtained with adaptive optics offer the potential for the long-term observation of disease progression.