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Student: Schließleder Gernot

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DEEP PHENOTYPING OF PROM1-ASSOCIATED RETINAL DEGENERATION

Gernot Schliessleder; Angelos Kalitzeos; Melissa Kasilian; Navjit Singh; Ziyuan Wang; Zhihong Jewel Hu; Manuel Grosspoetzl; SriniVas Sadda; Andreas Wedrich; Michel Michaelides; Rupert W. Strauss

Background/Aims: The purpose of this study was to investigate retinal structure in detail of subjects with autosomal-dominant (ad) and autosomal-recessive (ar) PROM1-associated retinal degeneration (PROM1-RD), study design: institutional, cross-sectional study

Methods: Four eyes from four subjects (three with ad and one with ar) PROM1-RD were investigated by ophthalmic examination including best-corrected visual acuity (BCVA) and multimodal retinal imaging: fundus autofluorescence (FAF), spectral-domain optical coherence tomography (SD-OCT) and adaptive optics scanning light ophthalmoscopy (AOSLO). Quantitative assessment of atrophic lesions determined by FAF, thickness of individual retinal layers and cone photoreceptor quantification was performed.

Results: BCVA ranged from 20/16 to 20/200. Initial pathologic changes included the presence of hyperautofluorescent spots on FAF imaging, while later stages demonstrated discrete areas of atrophy. In all patients, thinning of the outer retinal layers on SD-OCT with varying degrees of atrophy could be detected depending on disease-causing variants and age. Cone density was quantified both in central and/or at different eccentricities from the fovea. Longitudinal assessments were possible in two patients.

Conclusions: PROM1-RD comprises a wide range of clinical phenotypes. Depending on the stage of disease, the cone mosaic in PROM1-RD is relatively preserved and can potentially be targetted by cone-directed interventions.