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Genotypic and phenotypic Analysis in Slovenian Patients with Corneal Stromal Dystrophies

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Purpose: To assess the genetic profiles and clinical pictures of patients with stromal dystrophies and confirmed pathogenic mutations.
Methods: A systematic review and retrospective study were conducted at the Eye Clinic and the Clinical Institute of Genomic Medicine, University Medical Centre Ljubljana.
Results: Genetic analysis was indicated at the Cornea outpatient clinic, where severe cases of stromal corneal dystrophies are being followed-up before and after corneal transplantation. 7 patients with corneal stromal dystrophy underwent genetic analysis between 2015 and 2025 using the PanelAP (21-gene panel, https://nhsgms-panelapp.genomicsengland.co.uk/panels/658/v3.2) for corneal dystrophies. Genetic testing using next-generation sequencing revealed a causative pathogenic variant in 5 cases, while 2 cases were classified as having probable pathogenic variants. Pathogenic variants were identified in the TGFBI gene, whereas probable pathogenic variants were detected in the CHST6 and GSN genes. Clinical images and familial data of patients with genetically confirmed stromal dystrophies are presented.
Conclusions Ongoing advances in gene therapy, regenerative therapy and cell augmentation therapy may eventually result in the development of alternative, novel treatments for corneal dystrophies, which may substantially improve the quality of life of patients with these conditions and reduce the need for transplants.

Genotipska in fenotipska analiza pri slovenskih bolnikih s stromalnimi distrofijami roženice

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Namen: Genetska in klinična slika pri bolnikih s stromalnimi distrofijami in potrjenimi patogenimi mutacijami.
Metode: sistematični pregled, retrospektivna raziskava, Očesna klinika UKC Ljubljana in Klinični inštitut za genetiko UKC Ljubljana.
Rezultati: Iz Ambulante za refraktivno kirurgijo in bolezni roženice na Očesni kliniki UKC Ljubljana, kjer se vodijo pacienti zaradi težjih oblik stromalnih distrofij pred in po transplantaciji, je bilo v letih od 2015 do 2025 testiranih 7 pacientov s stromalno distrofijo. Genetsko testiranje z metodo sekvenciranja naslednje generacije in pregledom 21 genov v panelu za distrofijo roženice (https://nhsgmspanelapp.genomicsengland.co.uk/panels/658/v3.2) je v 5 primerih pokazalo vzročno patogeno različico gena, v 2 pa je bila le-ta verjetna. Patogene različice so bile ugotovljene v genu TGFBI, verjetno patogene pa v genih CHST6 in GSN. Prikazujemo klinične slike in družinske podatke pacientov z gentsko potrjenimi stromalnimi distrofijami roženice.
Zaključki: Neprestani napredki na področju genske terapije, regenerativne terapije in terapije z augmentacijo celic lahko v prihodnosti privedejo do razvoja alternativnih in inovativnih zdravljenj roženičnih distrofij, kar bi lahko bistveno izboljšalo kakovost življenja pacientov s temi stanji in zmanjšalo potrebo po trasnplantacijah.