Nevrooftalmologija in mrežnične distrofije, Thursday, May 15 2025, 14:25-15:30
Location: dvorana Grandis
Session: **Nevrooftalmologija in mrežnične distrofije / Neuro-ophthalmology and retinal dystrophies**
Chairs: Marko Hawlina and Martina Jarc Vidmar

OR-011
Natural history of Stargardt disease: The longest follow up cohort study
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Purpose: The study aimed to assess long term natural history of Slovenian Stargardt disease (STGD1) disease patients, inclouding electrophysiological (ERG) and fundus autofluorescence (FAF) progression rate with the median follow-up of 18 years (range 10-26 years).
Methods: 18 genetically confirmed STGD1 patients (5 male, 13 female) were included. Age at first exam was 22 years (range 7-46), age at last exam 40 years (range 17-72). Snellen best corrected visual acuity (VA), large-field pattern (PERG) and full-field electroretinography (ffERG) and fundus autofluorescence appearance (FAF) were analysed. Patients were classified into four Fishman stages and three electroretinography groups (Group 1-macular involvement, Group 2- macular involvement and generalised cone dysfunction and Group 3- macular involvement and generalised cone and rod dysfunction). Areas of definitely decreased autofluorescence (DDAF) were measured. Patients were further substratified based on genotype and phenotype-genotype corelation was performed.
Results: Median yearly VA loss was 0,009 per year (range 0,002-0.071), the median progression rate of DDAF area was 0.354 (range 0.002-4.359) mm2 per year. 8 out of 18 patients (44%) showed ERG progression, FAF appearance progressed in 10 out of 18 patients (56%). In 2 out of 18 patients (11%) FAF stayed stable and ERG progressed, in 4 out of 18 patients (22%) FAF progressed and ERG stayed stable, in 6 out of 18 patients (33%) both FAF and ERG progressed and in 6 out of 18 patients (33%) both FAF and ERG stayed stable. A total of 66.6% patients from group 1 showed ERG group transition during follow up, with 33.3% progressing to group 2 and 33,3% to group 3. 50% of patients from ERG group 2 progressed to ERG group 3. Patients harbouring p.(Gly1961Glu) or p.(Asn1868IIe)allele had significantly slower DDAF progression rate (0.07mm2 vs. 1.03mm2), when compared to patients with other genotypes, as well as significantly later age of onset (20 years vs.13 years).
Conclusions: Structural and functional parameters, together with genotype, should be considered when counselling patients regarding prognosis and monitoring Stargardt disease progression. Patients harbouring hypomorphic variants p.(Gly1961Glu) or p.(Asn1868IIe) presented with overall milder disease than patients with other genotypes.

Najdaljša kohortna študija spremljanja bolnikov s Stargardtovo distrofijo
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Namen : V študiji smo spremljali naravni potek bolezni in progresijo elektrofizoloških sprememb ter napredovanje sprememb v avtofluorescenci očesnega ozadja pri slovenskih bolnikih s Stargardtovo distrofijo. Povprečno smo bolnike spremljali 18 let (razpon od 10-26 let).
Metode : Vključenih je bilo 18 genetsko potrjenih bolnikov s STGD1 (5 moških, 13 žensk). Starost ob prvem pregledu je bila 22 let (razpon od 7-46) in pri zadnjem pregledu 40 let (razpon od 17-72). Analizirali smo najboljšo korigirano vidno ostrino (VO) po Snellenu, slikovni ERG (PERG, velika polja) in fotopični in skotopični ERG (SFERG) ter avtofluorescenco očesnega ozadja. Bolniki so bili razvrščeni glede na AF v štiri Fishmanove stopnje in glede na ERG v tri elektroretinografske skupine (skupina 1-makularne spremembe, skupina 2- okvara makule in generalizirana okvara čepnic, skupina 3- okvara makule in generalizirana okvara čepnic in paličnic). Izmerili smo področja definitivno zmanjšane avtofluorescence (DDAF). Bolniki so bili dodatno razdeljeni glede na genotip, primerjali smo fenotip in genotip.
Rezultati: Mediana letne izgube VO je bila 0,009 na leto (razpon 0,002-0,071), mediana stopnja napredovanja območja DDAF je bila 0,354 (razpon 0,002-4,359) mm2 na leto. Pri 8 od 18 bolnikov (44%) je prišlo do napredovanja ERG, spremembe v FAF smo opazili pri 10 od 18 bolnikov (56%). Pri 2 od 18 bolnikov (11%) je FAF ostala stabilna, prišlo je do napredovanja ERG, pri 4 od 18 (22%) je prišlo do napredovanja FAF, ERG je ostal stabilen, pri 6 od 18 (66%) je prišlo do napredovanja FAF in ERG, pri 6 od 18 (66%) pa sta tako FAF kot ERG ostala stabilna. Pri 66,6 % bolnikov iz ERG skupine 1 smo opazili napredovanje: 33,3 % so napredovali v ERG2 in v 33,3 % v ERG 3. 50 % bolnikov iz skupine ERG 2 je napredovalo v skupino ERG 3. Bolniki, ki so bili nosilci mutacije p.(Gly1961Glu) oz. p.(Asn1868IIe)alel, so imeli značilno počasnejšo stopnjo napredovanja DDAF v primerjavi z bolniki, ki so bili nosilci drugih mutacij (0,07 mm2 v primerjavi z

1,03 mm2), pri njih se je bolezen pojavila bistveno kasneje (v starosti 20 let v primerjavi s 13 leti).
Zaključki : Pri spremljanju bolnikov s Stargardtovo distrofijo in svetovanju le-tem glede prognoze in napredovanja bolezni je treba upoštevati strukturne in funkcionalne parametre skupaj z genotipom. Bolniki s hipomorfnimi različicami p.(Gly1961Glu) ali p.(Asn1868IIe) so imeli na splošno blažjo obliko bolezni kot bolniki z drugimi genotipi.