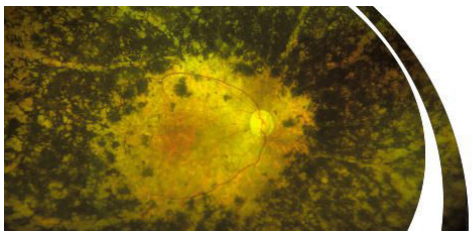

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clinical diagnosis shouldn't determine the IRD genes to be screened as use of large gene panels can maximise diagnostic yield.

Poster 36

Identification Of Novel Mutations In COL4A1 In Patients Presenting With Familial Retinal Arteriolar Tortuosity (fRAT) - Joanne Topping, University of Leeds
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Introduction: fRAT is a rare autosomal dominant disorder characterized by progressive tortuosity of the second- and third-order retinal arterioles. The aim of this study was to identify the molecular basis of fRAT in two unrelated patients.
Methods: Ophthalmologic evaluation included fundus examination, fluorescein angiography and OCT imaging. Whole exome sequencing (WES) or direct Sanger sequencing was performed to identify the mutations.
Results: Patient 1 is a white 37-year old male with fRAT and a history of spontaneous retinal hemorrhage, myocardial infarction and systemic hypertension. He was a member of a large family containing seven members affected with fRAT. WES revealed a novel heterozygous missense mutation in COL4A1, c.1582G>C, p.(G528R) which segregated with the phenotype in family members. Patient 2 is a 39-year old white male with isolated RAT. Sanger sequencing of exons 23-25 of COL4A1 revealed a novel heterozygous missense mutation c.1547G>A, p.(Gly516Asp). Both mutations are predicted to be pathogenic and are located in the same domain. The patients are being counseled with regard to the investigation of the systemic features of COL4A1 disease).
Conclusions: This study identified two novel mutations in COL4A1 in patients with fRAT and highlights the systemic phenotypic variability seen in patients with COL4A1 mutations.

Poster 37

A Recurrent Character And A High Frequency Of 11P13 Deletion Affecting *PAX6* Downstream Regulatory Regions In Aniridia Patients From Russia - Tatyana Vasilyeva, Research Centre for Medical Genetics
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Introduction: Aniridia (OMIM 106210) is autosomal dominant congenital panocular disorder caused by *PAX6* gene damage by large deletions and small mutations. The study aimed to determine *PAX6* mutation spectrum in Russian aniridia patients.
Methods: 121 unrelated families with congenital aniridia (147 patients) underwent ophthalmic examination and DNA testing: Sanger sequencing and MLPA followed by loss of heterozygosity of STR markers analysis.
Results: 16 patients from 10 unrelated families share the same 11p13 0.51.5 Mb deletion affecting *PAX6* downstream regulatory regions: 5 familial and 5 sporadic. The frequency of the deletion, 8.3% (10/121), is higher than hotspot c.718CT rate (7/121, 5.7%).
Conclusions: The high rate of the deletion in studied cohort suggests a common underlying mechanism of its formation and points to 11p13 genomic region instability. Supported by RFBR grant 17-04-00475.

Poster 38

Ophthalmic Manifestations of Heimler Syndrome Due To *PEX6* Mutations - Nutsuchar Wangtiraumnuay, MD, Queen Sirikit Natianal Institute of Child Health
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Introduction: Pigmentary retinal dystrophy and macular dystrophy have been previously reported in Heimler syndrome due to mutations in *PEX1*. Here we reported the ocular manifestations in Heimler syndrome due to mutations in *PEX6*.
Methods: Medical records were reviewed to identify patient demographics, ophthalmic and systemic findings, and results of diagnostic testing including whole genome sequencing.
Results: Both patients had heterozygous *PEX6* mutations. They exhibited pigmentary retinopathy, hyperfluorescent deposits, depletion of photoreceptors with intraretinal cystoid spaces. Full field electroretinograms showed abnormal photopic with abnormal mfERG.
Conclusions: Heimler syndrome due to biallelic *PEX6* mutations demonstrates a macular dystrophy with characteristic fundus autofluorescence and may be complicated by intraretinal cystoid spaces.