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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**

Co-Authors:
Evangelia Panagiotou, MD, MSc, University of Leeds
Ambreen Kalhoro, MD, Department of Ophthalmology, St. James' University Hospital
Raj Mukherjee, MD, Department of Ophthalmology, St. James' University Hospital
Martin McKibbin, MD, Department of Ophthalmology, St. James' University Hospital
Carmel Toomes, PhD, University of Leeds

**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**

Co-Authors:
Anna A. Voskresenskaya, MD, Cheboksary branch of S. Fyodorov Eye Microsurgery Federal State Institution
Vitaly V. Kadyshev, PhD, Research Center for Medical Genetics
Nadezhda A. Pozdeyeva, DSC, Cheboksary branch of S. Fyodorov Eye Microsurgery Federal State Institution
Olga V. Khlebnikova, DSC, Research Center for Medical Genetics
Rena A. Zinchenko, DSC, Research Center for Medical Genetics, Pirogov Russian National Research Medical University
Andrey V. Marakhonov, PhD, Research Center for Medical Genetics, Moscow Institute of Physics and Technology
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 Wangtirumnuay, MD, Queen Sirikit National Institute of Child Health
Co-Authors:
Waleed Abed Alnabi, MD, Wills Eye Hospital
Mai Tsukikawa, MD, Sidney Kimmel Medical College at Thomas Jefferson University
Jenina Capasso, LCGC, Wills Eye Hospital
Reuven Sharony, MD, Meir Medical Center affiliated with the Sackler Faculty of Medicine, Tel Aviv University
Chris F Inglehearn, PhD, St. James's University Hospital, University of Leeds
Alex V Levin, MD, MHSc, Wills Eye Hospital

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.