UK-EGG Annual Conference Programme

Friday 9th June 2023

EDINBURGH

8:30 – 9:15	Registration
9:15 – 9:20	Welcome – **Wendy Bickmore**
9:20 – 10:20	***Session 1: Eye Developmental Disorders***
9:20 – 9:40	**Nikki Hall** - Eye developmental defects: pathology, diagnosis, and genetics
9:40 – 10:00	**Joe Rainger** - Closing the gap - understanding tissue fusion processes in optic fissure closure and coloboma
10:00 – 10:20	**Shipra Bhatia** - Functional assessment of disease-associated sequence variation in the noncoding genome
10:20 – 10:50	**BREAK, POSTERS, Meet the sponsors**
10:50 – 12:20	***Session 2: From the clinic***
10:50 – 11:40	**Interesting Clinical cases** 7-minute talks *Urvi Patel* - Ophthalmic findings guide investigations revealing systemic diagnosis in young patient with cardiac failure *Mauro Lecca* - Expanding the genotypic and phenotypic spectrum in a cohort of 65 individuals with congenital cataract *Celia Azevedo Soares* - Variable phenotypic expression of the m.8993T>G pathogenic variant in a family *Mohammad Anas* - Electroretinography guides re-analysis of whole genome data to find undetected variant in CACNA1F *Daniel Jackson* - Natural history of autosomal dominant RP1-retinitis pigmentosa
11:40 – 12:00	**Miguel Bernabeu** - TBC - research/ AI/retinal imaging/genetic eye diseases including AMD
12:00 – 12:20	**David Gilmour** - Clinical trials for the IRDs: a local PI's perspective
12:20 – 13:50	**LUNCH, POSTERS*, AGM, Meet the sponsors**
13:20 – 13:50	***All posters viewing session**
13:30 – 13:50	**AGM**
13:50 – 15:10	***Session 3: Patient experience/Genomic diagnosis***
13:50 – 14:10	**Libby Clegg** - patient voice with **Roly Megaw**
14:10 – 14:30	**Samantha Malka** - Genetic counselling in the era of whole genome sequencing
14:30 – 14:40	*Jacqueline Turner* - Investigation of variants of unknown significance runs the risk of patients having predictive testing without adequate pre-test counselling
14:40 – 14:50	*Dorine Bax* - Use of Optical Genome Mapping to advance Genetic Diagnosis in Developmental Eye Disorders
14:50 – 15:10	**Jamie Ellingford** - variant interpretation for improving genetic diagnosis of eye diseases
15:10 – 15:40	**BREAK, POSTERS, Meet the sponsors**
15:40 – 17:00	***Session 4: Research Insights***
	ECR talks
15:40 – 15:50	*Thomas Julian* - Phenome-wide Mendelian randomisation analysis provides causal insights into glaucoma and age-related macular degeneration.
15:50 – 16:00	*Siying Lin* - Spectrum of genetic variants in the commonest genes causing inherited retinal disease in a large molecularly characterised UK cohort
16:00 – 16:10	*Mihaly Badonyi* - Protein complex structural data enables phenotype-level prediction of missense variants in PRPH2
16:10 – 16:20	*Linda Nguyen* - Defining the mechanism of photoreceptor cell death in retinitis pigmentosa using the Pde6batrd2 mouse
16:20 – 17:00	**KEYNOTE: Elfride de Baere** Comparative 3D genome analysis between neural retina and RPE reveals differential cis-regulatory interactions at retinal disease loci
17:00 – 17:10	Prizes, Closing remarks
17:10 – close	DRINKS RECEPTION

Selected from submitted abstracts