UK EGG Annual Meeting Programme

21st September 2021

8:30 – 9:15 Registration

9:15 – 9.20 Welcome by **Professor Mariya Moosajee**

9.20 – 10.20 Session 1: Clinical Session "Advances in genetic eye disease"

- 9:20 – 9:40 **Professor Andrew Webster** – *The future of WGS for IRDs*
- 9:40 – 10:00 **Professor Stephan Beck** – *Connecting dots for genomic medicine*
- 10:00 – 10:20 **Dr Anthony Khawaja** – *Complex genetics of primary open-angle glaucoma and key endophenotypes*

10:20 – 11:00 Coffee break and meet the associations

11.00 – 12.00 Session 2: Free paper/ Research Session- ECRs

- 11.05 – 11.15 **Dr Valentina Cipriani** – *Beyond Factor H: the influence of genetic variation associated with age-related macular degeneration on circulating FHL-1 and FHR protein levels*
- 11.15 – 11.25 **Ms Philippa Harding** – *Transcriptome-wide investigation of microphthalmia patient-derived iPSC organoid models reveals molecular disruption associated with missense PAX6 variant p.(Asn124Lys)*
- 11.25 – 11.35 **Dr Imran H. Yusuf** – *Gene therapy rescues cone and rod function in a pre-clinical model of CDHR1-associated retinal degeneration through restoration of photoreceptor outer segments*
- 11.35 – 11.45 **Dr Maria Toms** – *Using non-viral S/MAR DNA vectors to restore protein expression in models of choroideremia*
- 11.45 – 11.55 **Dr Nikolaos Tzoumas** – *Loss-of-Function Complement Factor I Variants Associated with Macular Thinning and Age-Related Macular Degeneration in the UK Biobank*

12:00 – 12:45 Keynote Talk: **Professor Tara Moore** – *Progressing gene therapy for corneal dystrophies*

12:45 – 13:45 Lunch break, poster session and meet the associations

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13.20 – 13.40 AGM

13.40 – 15.20 Session 3: Genetic counselling and interesting cases session

- 13.45-14.05 **Moorfields Genetics Team** – *Live MDT session*
- 14:05 – 14.15 **Dr Salwah Rehman** – *Novel association of CRX p.(Arg90Trp) mutation with cone dystrophy and axial length shortening*
- 14.15 – 14.25 **Ms Laura Whelan** – *Atypical peroxisome biogenesis disorder associated with a nonsense variant in PEX5 detected as part of genetic testing for inherited retinal disease.*
- 14:30 – 14:50 **Ms Eshika Haque** – *"A to Z" of pre-implantation diagnosis*
- 14.50 – 15.00 **Dr Penny Clouston** – *Lab update*
- 15.00 – 15:20 **Mr Sol Woodroffe** – *Patient session*

15:20 – 15:50 Coffee break and meet the associations

15.50 – 16.50 Session 4: Future of genomic ophthalmology session

- 15:50 – 16:10 **Professor Wendy Bickmore** – *Chromosome organisation and gene regulation*
- 16:10 – 16:30 **Professor Majlinda Lako** – *A single cell atlas of human cornea that defines its development, limbal progenitor cells and their interactions with the immune cells*
- 16.30 – 16.50 **Professor Pearse Keane** – *Cloud-based pipelines for research in inherited retinal disease*

16:50 – 17:00 Prize presentation and closing remarks

17.00 – 18.00 Networking and drinks reception

Register on https://ukegg.com/#registration for face-to-face or virtual attendance.

For any queries, please email **UKeyegenetics@gmail.com**