Morning programme

08:00	09:00	**REFRESHMENTS & REGISTRATION & MEET OUR SPONSORS**
09:00	09:10	Welcome by Bristol hosts
Session 1: Advances in clinical and population eye genetics. Chair: Denize Atan		
09:10	09:40	***Community genomic studies provide new insights into inherited eye disorders*** Dr Emma Baple, University of Exeter
09:40	10:10	***Recent developments in myopia genetics*** Professor Jeremy Guggenheim, University of Cardiff, & Professor Cathy Williams, University of Bristol
10:10	10:40	***Gene therapy for Leber hereditary optic neuropathy*** Dr Patrick Yu Wai Man, University of Cambridge
10:40	11:00	**REFRESHMENTS & POSTERS & MEET OUR SPONSORS**
Session 2: Session for Early Career Researchers in eye genetics. Chair: Gavin Reynolds		
11:00	11:10	***The contribution of common regulatory and protein-coding TYR variants in the genetic architecture of albinism*** David Green, University of Manchester
11:10	11:20	***Modelling dominant optic atrophy using pluripotent stem-cell derived retinal ganglion cells*** Joshua Harvey, Institute of Ophthalmology & MEH
11:20	11:30	***Fine-mapping of retinal vascular complexity loci identifies Notch signalling regulatory variants supporting a shared mechanism with myocardial infarction outcomes*** Ana Villaplana-Velasco, University of Edinburgh
11:30	11:40	***Electroretinography is highly specific in differentiating Pantothenate Kinase-Associated Neurodegeneration from mimics*** Robert Spaull, Neuroscience, GOSH
11:40	11:50	***Retinitis Pigmentosa GTPase Regulator (RPGR) regulates actin-mediated photoreceptor disc formation*** Roly Megaw, University of Edinburgh
11:50	11:55	**SHORT BREAK**
Session 3: Keynote speaker. Introduction by Denize Atan.		
11:55	12:45	***G=E: how Mendelian randomization can help identify modifiable causes of disease*** Professor George Davey Smith, University of Bristol
12:45	13:45	**LUNCH & POSTERS & MEET OUR SPONSORS**
13:30	13:45	**Open AGM of UK EGG committee will occur in the theatre during lunch**

Afternoon programme

Session 4: Joint session with BrisCEV. Introduction by Denize Atan.		
13:45	14:05	***Visual electrophysiology: a review of its role in diagnosis and phenotyping*** Ruth Hamilton, President of ISCEV and Secretary of BriSEV
Session 5: Clinical case discussions. Chair: Nervine Elmeshad		
14:05	14:15	***Cataract, abnormal electroretinogram and visual evoked potentials in a child with SMA-LED2 - extending the phenotype*** Agata Oliwa, University of Glasgow
14:15	14:25	***Visual electrophysiological and retinal findings in a case of Kenny-Caffey syndrome Type 2*** Linda Shi, GOSH, London
14:25	14:35	***CACNA1A and Pathologic Eye Movements: A Case Series and Literature Review*** Claire Chan, University of Bristol
14:35	14:45	***RPGR-related retinal dystrophy and systemic associations*** Ruofan Connie Han, John Radcliffe Hospital, Oxford
14:45	14:55	***An Alu transposable element causing Rod-Cone Dystophy in two Irish families*** Jacqueline Turner, Mater Misericordiae, Dublin
14:55	15:15	**REFRESHMENTS & POSTERS & MEET OUR SPONSORS**
Session 5: Patient session - Historical perceptions and patient perspectives on visual impairment. Chair: Nervine Elmeshad		
15:15	15:40	***Storying "diffability": art, narrative and journeys with difference*** Dr Andy Flack, University of Bristol
15:40	15:55	**Interview with Simon Prowting by Amanda Churchill**
Session 6: Advances in molecular genetics. Chair: Amanda Churchill		
15:55	16:25	***A molecular overview of the evolutionary history of vision*** Professor Davide Pisani, University of Bristol
16:25	16:55	***Modelling retinal development and disease using hPSC derived retinal organoids*** Dr Jorn Lakowski, University of Southampton
Session 7: BREAKING NEWS. Chair: Amanda Churchill		
16:55	17:05	***PERCEIVE STUDY: real-world safety and effectiveness of voretigene neparvovec*** Professor Dominik Fischer, Universities of Tübingen and Oxford and Oxford Eye Hospital
CLOSING SESSION		
17:05	17:15	Prizes for best oral and poster presentations & Closing remarks
17:15	Until late	**DRINKS RECEPTION IN WATERSHED BAR/RESTAURANT**