# UK-EGG Webinar 3: UK Eye Genetics - Early Career Researchers-

# Pushing Ocular Genetic Frontiers

5.30 - 7.00pm, Thursday, 15th April, 2021

## Agenda

5:30 Introduction

**Rapid fire poster session**

5:35 3 minute posters followed by an open panel questions to speakers

- *An illustrative case of bilateral ectopia lentis due to mutations of the ADAMTSL4 gene*- Aditi Das, Great Ormond Street Hospital, London
- *High myopia referrals to the Genetic Eye Clinic: Are we being short-sighted*?- Melody Redman, Yorkshire Regional Genetics Service, Leeds.
- *Exome Sequencing of mendelian genes in syndromic and non syndromic patients with Microphthalmia Anophthalmia Coloboma identifies potentially causal mutations in 45%*- Irina Balikova, University Hospital Gasthisberg, Belgium
- *EPHA2 segregates with bilateral microphthalmia and congenital cataracts in two unrelated families*- Philippa Harding, UCL Institute of Ophthalmology, London
- *A systematic review of genetic mutations and clinical features associated with Late-Onset Retinal Degeneration*- Randa Li, University of Edinburgh, Edinburgh.
- *Ocular features associated with DYRK1A variants*- Cécile Méjécase, UCL Institute of Ophthalmology, London
- *Translational readthrough of ciliopathy genes BBS2 and ALMS1 using patient fibroblasts*- Jonathon Eintracht, UCL Institute of Ophthalmology, London
- *Genome-wide study of retinal vascular complexity identifies 8 novel loci and unravels shared genetic with cardiovascular events*- Ana Villaplana-Velasco, University of Edinburgh, Edinburgh.
- *Metabolomic analysis of choroideremia patients plasma reveals systemic lipid metabolism dysfunction and oxidative stress*- Dulce Lima Cunha, UCL Institute of Ophthalmology, London

**Oral presentations**

6:10pm: *A Qualitative study exploring the support requirements of families with inherited retinal dystrophies and how they obtain information*- Manara Gul, University of Cardiff, Cardiff

6:22pm: *Retrospective natural history study of a UK cohort of cerebrotendinous xanthomatosis*- Suzannah Bell- Moorfields Eye Hospital, London

6:34pm: *Characterisation of foveal development and cone photoreceptor specialisation in SLC38A8 mutations*- Helen Kuht, The University of Leicester, Leicester

6:46pm: *The role of BMP3 in the development of myopia*- Amy Findlay, University of Edinburgh, Edinburgh.