2016 Publications

Vitamin A deficiency due to bi-allelic mutation of RBP4: There's more to it than meets the eye.

Mutations in REEP6 Cause Autosomal-Recessive Retinitis Pigmentosa.

Mutations in CPAMD8 Cause a Unique Form of Autosomal-Recessive Anterior Segment Dysgenesis.

The Effect on Retinal Structure and Function of 15 Specific ABCA4 Mutations: A Detailed Examination of 82 Hemizygous Patients.

FUNCTIONAL AND ANATOMICAL OUTCOMES OF CHOROIDAL NEOVASCULARIZATION COMPLICATING BEST1-RELATED RETINOPATHY.


Development of an optimized AAV2/5 gene therapy vector for Leber congenital amaurosis owing to defects in RPE65.

Reevaluation of the Retinal Dystrophy Due to Recessive Alleles of RGR With the Discovery of a Cis-Acting Mutation in CDHR1.

The New Pretender: A Large UK Case Series of Retinal Injuries in Children Secondary to Handheld Lasers.

OCT angiography in the management of choroidal neovascular membrane secondary to Sorsby fundus dystrophy.

Phenotype and Progression of Retinal Degeneration Associated With Nullizigosity of ABCA4.

Clinical and Genetic Features of Choroideremia in Childhood.

Cone Photoreceptor Structure in Patients With X-Linked Cone Dysfunction and Red-Green Color Vision Deficiency.

Expanding the Phenotype of TRNT1-Related Immunodeficiency to Include Childhood Cataract and Inner Retinal Dysfunction.
Visual Acuity Loss and Associated Risk Factors in the Retrospective Progression of Stargardt Disease Study (ProgStar Report No. 2).

Comparison of Short-Wavelength Reduced-Illuminance and Conventional Autofluorescence Imaging in Stargardt Macular Dystrophy.

Recessive Retinopathy Consequent on Mutant G-Protein β Subunit 3 (GNB3).

Novel heterozygous mutation in YAP1 in a family with isolated ocular colobomas.

Nonsyndromic Retinal Dystrophy due to Bi-Allelic Mutations in the Ciliary Transport Gene IFT140

Investigation of SLA4A3 as a candidate gene for human retinal disease.

Molecular and Clinical Findings in Patients With Knobloch Syndrome.

Cone opsins, colour blindness and cone dystrophy: Genotype-phenotype correlations.

Differentiating drusen: Drusen and drusen-like appearances associated with ageing, age-related macular degeneration, inherited eye disease and other pathological processes.

Unilateral BEST1-Associated Retinopathy.

Advanced diagnostic genetic testing in inherited retinal disease: experience from a single tertiary referral centre in the UK National Health Service.


Phenotypic features of CRB1-associated early-onset severe retinal dystrophy and the different molecular approaches to identifying the disease-causing variants.

The Effect of Multispot Laser Panretinal Photocoagulation on Retinal Sensitivity and Driving Eligibility in Patients With Diabetic Retinopathy.

Nonsyndromic Retinal Dystrophy due to Bi-Allelic Mutations in the Ciliary Transport Gene IFT140.

Effects of Intraframe Distortion on Measures of Cone Mosaic Geometry from Adaptive Optics Scanning Light Ophthalmoscopy.

Preserved visual function in retinal dystrophy due to hypomorphic RPE65 mutations.
Lamination of the Outer Plexiform Layer in Optic Atrophy Caused by Dominant WFS1 Mutations.

RPGR-associated retinopathy: clinical features, molecular genetics, animal models and therapeutic options.

The Natural History of the Progression of Atrophy Secondary to Stargardt Disease (ProgStar) Studies: Design and Baseline Characteristics: ProgStar Report No. 1.

Mutations in CACNA2D4 Cause Distinctive Retinal Dysfunction in Humans.

2017 Publications

Quantitative Analysis of Retinal Structure Using Spectral-Domain Optical Coherence Tomography in RPGR-Associated Retinopathy.

Childhood-onset Leber hereditary optic neuropathy.

Mutations in the Spliceosome Component CWC27 Cause Retinal Degeneration with or without Additional Developmental Anomalies.

Association of Steroid 5α-Reductase Type 3 Congenital Disorder of Glycosylation With Early-Onset Retinal Dystrophy.

DETAILED RETINAL IMAGING IN CARRIERS OF OCULAR ALBINISM.

THE FUNDUS PHENOTYPE ASSOCIATED WITH THE p.Ala243Val BEST1 MUTATION.


Clinical Characterization of CNGB1-Related Autosomal Recessive Retinitis Pigmentosa.

Comprehensive Rare Variant Analysis via Whole-Genome Sequencing to Determine the Molecular Pathology of Inherited Retinal Disease.

Retinitis pigmentosa-associated cystoid macular oedema: pathogenesis and avenues of intervention.

Stargardt disease: clinical features, molecular genetics, animal models and therapeutic options.