

## Special Issue

# Recent Advances in Inherited Eye Disease

### Message from the Guest Editors

Inherited eye diseases (IED) represent a vast spectrum of blinding genetic disorders affecting all cells and tissue types of the globe. The genetic mechanisms that underlie these diseases are extremely complex and diverse, with hundreds of genes and many thousands of disease variants reported to date. Current diagnostic testing is focused on targeted gene panel analysis, and can identify causative variants in up to 60%–80% of cases where a clear clinical indication is present. However, despite the huge advances in knowledge and technology since the discovery of the first disease variant some 30 years ago, patients without a clear indication are often left without a molecular diagnosis, thus restricting their access to effective clinical management, counselling, and emerging therapeutics.

This Issue aims to advance the knowledge of the genetic etiology of IED and to inform future genomic studies, functional biologists, clinical diagnostic laboratories, and health care providers, and the wider field of Mendelian disease research. To this end, we invite papers reporting on novel findings in the genetics underlying any aspect of Mendelian IED.

---

### Guest Editors

Dr. Gavin Arno

1. UCL Institute of Ophthalmology, University College London, London WC1E 6BT, UK
2. Moorfields Eye Hospital, London WC1E 6BT, UK
3. GOSH NIHR Biomedical Research Centre, Great Ormond Street Hospital for Children NHS Foundation Trust, London WC1E 6BT, UK
4. Laboratory of Visual Physiology/Ophthalmic Genetics, National Institute of Sensory Organs, Tokyo 152-8902, Japan

Dr. Robert B Hufnagel

National Eye Institute, Bethesda, MD 20892-2510, USA

Dr. Jamie Ellingford

Manchester Centre for Genomic Medicine, The University of Manchester, Manchester M13 9PL, UK

---

### Deadline for manuscript submissions

closed (31 October 2019)

G C A T  
T A C G  
G C A T

## Genes

---

an Open Access Journal  
by MDPI

---

Impact Factor 2.8  
CiteScore 5.5  
Indexed in PubMed



[mdpi.com/si/27389](https://mdpi.com/si/27389)

*Genes*

Editorial Office

MDPI, Grosspeteranlage 5

4052 Basel, Switzerland

Tel: +41 61 683 77 34

[genes@mdpi.com](mailto:genes@mdpi.com)

[mdpi.com/journal/](https://mdpi.com/journal/)

[genes](https://mdpi.com/journal/genes)



G C A T  
T A C G  
G C A T

# Genes

---

an Open Access Journal  
by MDPI

---

Impact Factor 2.8  
CiteScore 5.5  
Indexed in PubMed



[mdpi.com/journal/  
genes](https://mdpi.com/journal/genes)



## About the Journal

### Message from the Editor-in-Chief

*Genes* is central to our understanding of biology, and modern advances such as genomics and genome editing have maintained genetics as a vibrant, diverse and fast-moving field. There is a need for good quality, open access journals in this area, and the *Genes* team aims to provide expert manuscript handling, serious peer review, and rapid publication across the whole discipline of genetics. Starting in 2010, the journal is now well established and recognised. Why not consider *Genes* for your next genetics paper?

---

### Editor-in-Chief

Prof. Dr. Selvarangan Ponnazhagan  
Department of Pathology, The University of Alabama at Birmingham,  
1825 University Blvd, SHEL 814, Birmingham, AL 35294-2182, USA

---

### Author Benefits

#### Open Access:

free for readers, with article processing charges (APC) paid by authors or their institutions.

#### High Visibility:

indexed within Scopus, SCIE (Web of Science), PubMed, MEDLINE, PMC, Embase, PubAg, and other databases.

#### Journal Rank:

JCR - Q2 (Genetics and Heredity) / CiteScore - Q2 (Genetics (clinical))