

## Special Issue

# Clinical and Molecular Aspects of Rare Disease

### Message from the Guest Editors

Over the last decade, our knowledge about clinical and molecular aspects of rare diseases has profoundly deepened; the advances in genetic testing and genome sequencing have allowed us to find a personalized, tailored therapeutic approach, thus prolonging the survival and improving the quality of life of hundreds of patients. Disciplines such as genomics, transcriptomics and proteomics have provided pathophysiological insights into stratifying patients according to their specific clinical phenotype and molecular mechanisms; therefore, the groups for whom tailored therapy is most beneficial at earlier disease stages can be identified. This Special Issue aims to gather and present novel research in the field of rare cardiovascular diseases. Articles may include the molecular basis of complex cardiovascular conditions, new frontiers in gene testing and genome editing, gene therapy for rare conditions, new genotype–phenotype correlations, personalized, targeted pharmacological approaches in syndromic conditions and applications of translational research to clinical aspects of rare diseases.

---

#### Guest Editors

Dr. Giuseppe Limongelli

Prof. Dr. Giulia Frisso

Dr. Michele Lioncino

Dr. Martina Caiazza

---

#### Deadline for manuscript submissions

closed (20 December 2022)

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/122383](https://mdpi.com/si/122383)

*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/  
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))