

## Special Issue

# Achromatopsia: From Genetics to Therapy

### Message from the Guest Editors

Achromatopsia (rod monochromatism) is a rare (1 in 30,000–50,000) autosomal recessive disorder affecting cone photoreceptors. Pathogenic variants in six genes (*CNGA3*, *CNGB3*, *PDE6C*, *PDE6H*, *GNAT2*, *ATF6*) have been associated with achromatopsia to date. Currently, gene therapy trials are ongoing to correct the defects of *CNGA3* and *CNGB3* that account for almost 70% of cases. Novel therapeutic approaches can greatly benefit from a better understanding of the molecular basis of achromatopsia. Defining the genetic components and the mechanisms underlying disease progression has implications for patient selection and intervention timing.

This Special Issue aims to offer novel insights into the molecular pathogenesis of achromatopsia, the mechanistic role of the genetic components, and relevant genotype–phenotype correlations. We welcome contributions on new gene targets and mechanisms that improve our understanding of the disease. Topics also include novel molecular therapeutic strategies.

### Guest Editors

Prof. Dr. Francesca Simonelli

Prof. Dr. Francesco Testa

Dr. Marianthi Karali

### Deadline for manuscript submissions

closed (31 May 2022)



## International Journal of Molecular Sciences

---

an Open Access Journal  
by MDPI

---

Impact Factor 4.9  
CiteScore 9.0  
Indexed in PubMed



[mdpi.com/si/71247](https://mdpi.com/si/71247)

*International Journal of  
Molecular Sciences*  
Editorial Office  
MDPI, Grosspeteranlage 5  
4052 Basel, Switzerland  
Tel: +41 61 683 77 34  
[ijms@mdpi.com](mailto:ijms@mdpi.com)

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





# International Journal of Molecular Sciences

---

an Open Access Journal  
by MDPI

---

Impact Factor 4.9  
CiteScore 9.0  
Indexed in PubMed



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



## About the Journal

### Message from the Editor-in-Chief

The *International Journal of Molecular Sciences (IJMS)* is an open access journal, which was established in 2000. The journal aims to provide a forum for scholarly research on a range of topics, including biochemistry, molecular and cell biology, and molecular biophysics. *IJMS* publishes both original research and review articles, and regularly publishes special issues to highlight advances at the cutting edge of research. We invite you to read recent articles published in *IJMS* and consider publishing your next paper with us.

---

### Editor-in-Chief

Prof. Dr. José L. Quiles  
Department of Physiology, Institute of Nutrition and Food Technology  
"Jose Mataix", Biomedical Research Center, University of Granada,  
Avda. Conocimiento s/n, 18100 Armilla, Granada, Spain

---

### 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, PMC, MEDLINE, Embase, CAPus / SciFinder, and other databases.

#### Journal Rank:

JCR - Q1 (Biochemistry and Molecular Biology) / CiteScore - Q1 (Organic Chemistry)