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

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The International Journal of Molecular Sciences (*IJMS*, ISSN 1422-0067) 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, molecular biophysics, molecular medicine, and all aspects of molecular research in chemistry. *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.

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