

## Special Issue

# Research in Genetic Causes of Adult-Onset Disorders

### Message from the Guest Editor

Advances in genomic technologies have revolutionized our understanding of monogenic adult-onset disorders, enabling accurate diagnoses and therapeutic development. Neurodegenerative diseases, cancer-predisposing syndromes, cardiomyopathies, and autosomal dominant polycystic kidney disease are prominent examples. This Special Issue focuses on recent research into the genetic causes of adult-onset disorders, emphasizing genomic approaches, mechanistic studies validating genomic findings, specific therapeutic approaches related to the genetic cause, penetrance, and variant expression of pathogenic within families. We welcome the submission of original research, case studies, up-to-date reviews, and discussions on topics including:

- Methods to validate variants of uncertain significance;
- Methods to detect and validate non-canonical variants;
- Expansion of the phenotypic spectrum and its relationship with dual molecular diagnosis;
- The clinical and molecular characterization of family members;
- Estimates of disease penetrance using population cohort data;
- Gene-tailored therapeutic options (including pharmacogenomics).

### Guest Editor

Dr. Claudio Graziano

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### Deadline for manuscript submissions

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## International Journal of Molecular Sciences

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### Message from the Editor-in-Chief

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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### Editor-in-Chief

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