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## Diagnosis and Therapies for Rare Diseases

Guest Editors:

**Dr. Maria Monticelli**

**Dr. Bruno Hay Mele**

**Dr. Giuseppina Andreotti**

Deadline for manuscript  
submissions:

**10 September 2024**

### Message from the Guest Editors

Dear Colleagues,

We extend our invitation for you to contribute to the Special Issue “Diagnosis and Therapies for Rare Diseases”. Rare genetic disorders present unique diagnostic, treatment, and management challenges, underscoring the critical need for focused research efforts in this domain. This Special Issue aims to advance our understanding of rare diseases, spotlighting various significant aspects.

In particular, we seek contributions to identify known or novel disease-associated variants alongside advancements in related technologies crucial for accurate diagnosis. Additionally, we encourage submissions addressing the forefront of de novo drug discovery and drug repositioning, pivotal in pursuing treatments for currently incurable diseases and enhancing existing therapies burdened by substantial limitations.

Your expertise and insights would greatly enrich this endeavour, fostering progress in diagnosing and treating rare genetic diseases. We eagerly anticipate your valuable contributions to this vital area of research.

Dr. Maria Monticelli

Dr. Bruno Hay Mele

Dr. Giuseppina Andreotti

*Guest Editors*



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# Special Issue



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## Editor-in-Chief

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## Message from the Editor-in-Chief

Genes are central to our understanding of biology, and modern advances such as genomics and genome editing have maintained genetics as a vibrant, diverse and fastmoving 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.

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