Special Issue

Diagnosis and Therapy of Rare Diseases

Message from the Guest Editors

This Special Issue on the "Diagnosis and Therapy of Rare Diseases" is dedicated to advancing our understanding and management of these medically challenging conditions.

The diverse and dynamic nature of cutting-edge research in rare diseases, such as exploring therapies targeting specific genetic mutations to improve lung function in cystic fibrosis or gene therapies in conditions such as spinal muscular atrophy, focuses on improving diagnostics, treatment options, and the overall quality of life for individuals affected by these conditions.

We welcome original research, reviews, case studies, and discussions concerning the ethical and legal considerations associated with rare disease management. Researchers, clinicians, and experts in the field are encouraged to share their insights into the molecular mechanisms, biomarkers, and multidisciplinary approaches that facilitate earlier, faster, and more accurate diagnosis, along with ensuring treatment availability, accessibility, and affordability.

Guest Editors

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Deadline for manuscript submissions

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Editor-in-Chief

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