



*Special Issue Reprint*

## **Advances in Rare Diseases Biomarkers**

[www.mdpi.com/books/reprint/10087](http://www.mdpi.com/books/reprint/10087)

Edited by  
Andrea Bernini

ISBN 978-3-7258-2412-0 (Hardback)  
ISBN 978-3-7258-2411-3 (PDF)



A rare disease is a health condition with a lower prevalence than common diseases. The World Health Organization defines a rare disease as one that strikes fewer than 65 per 100,000 people. However, their combined effect is significant: around 7,000 rare diseases affect approximately 350 million people worldwide.

Biomarkers play a crucial role in diagnosing and monitoring rare diseases, which are often challenging to detect and understand due to their low prevalence and diverse clinical manifestations. Biomarkers serve as measurable indicators of biological processes or conditions in rare diseases, offering valuable insights into disease mechanisms and progression. These markers may include genetic mutations, protein levels, or other molecular signatures unique to a rare condition. The discovery and validation of such biomarkers contribute to early detection and the development of targeted therapies, allowing for more effective and personalized treatment approaches.

As technology advances, the integration of omics technologies, such as genomics, proteomics, and metabolomics, has further expanded the repertoire of potential biomarkers, fostering a deeper understanding of rare diseases and paving the way for innovative diagnostic and therapeutic strategies. Biomarkers enhance our ability to navigate diagnostic challenges and promise to improve patient care and foster breakthroughs in treatment modalities.



Order Your Print Copy  
You can order print copies at  
[www.mdpi.com/books/reprint/10087](http://www.mdpi.com/books/reprint/10087)

MDPI Books offers quality open access book publishing to promote the exchange of ideas and knowledge in a globalized world. MDPI Books encompasses all the benefits of open access – high availability and visibility, as well as wide and rapid dissemination. With MDPI Books, you can complement the digital version of your work with a high quality printed counterpart.



## Open Access

Your scholarly work is accessible worldwide without any restrictions. All authors retain the copyright for their work distributed under the terms of the Creative Commons Attribution License.



## Author Focus

Authors and editors profit from MDPI's over two decades of experience in open access publishing, our customized personal support throughout the entire publication process, and competitive processing charges as well as unique contributor discounts on book purchases.



## High Quality & Rapid Publication

MDPI ensures a thorough review for all published items and provides a fast publication procedure. State-of-the-art research and time-sensitive topics are released with a minimum amount of delay.



## High Visibility

Due to our global network and well-known channel partners, we ensure maximum visibility and broad dissemination. Title information of books is sent to international indexing databases and archives, such as the Directory of Open Access Books (DOAB), and the Verzeichnis Lieferbarer Bücher (VLB).



## Print on Demand and Multiple Formats

MDPI Books are available for purchase and to read online at any time. Our print-on-demand service offers a sustainable, cost-effective and fast way to publish MDPI Books printed versions.