genes

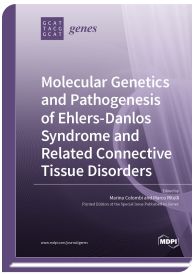
IMPACT FACTOR 3.5

Indexed in: PubMed

CITESCORE 5.2

Special Issue Reprint

Molecular Genetics and Pathogenesis of Ehlers–Danlos Syndrome and Related Connective Tissue Disorders



www.mdpi.com/books/reprint/2603

Edited by
Marina Colombi
Marco Ritelli

ISBN 978-3-03936-322-3 (Hardback)
ISBN 978-3-03936-323-0 (PDF)

Ehlers–Danlos syndromes (EDS) are a group of heritable connective tissue disorders (HCTDs) characterized by a variable degree of skin hyperextensibility, joint hypermobility and tissue fragility. The current EDS classification distinguishes 13 subtypes and 19 different causal genes mainly involved in collagen and extracellular matrix synthesis and maintenance. EDS need to be differentiated from other HCTDs with a variable clinical overlap, including Marfan syndrome and related disorders, some types of skeletal dysplasia and cutis laxa. The clinical recognition of EDS is not always straightforward, and, for a definite diagnosis, molecular testing can be of great assistance, especially in patients with an uncertain phenotype. Currently, the major challenging task in EDS is to unravel the molecular basis of the hypermobile EDS that is the most frequent form, and for which the diagnosis is only clinical in the absence of any definite laboratory test. This EDS subtype, as well as other EDS-reminiscent phenotypes, are currently investigated worldwide to unravel the primary genetic defect and related pathomechanisms. The research articles, case report, and reviews published in the Special Issue entitled “Molecular Genetics and Pathogenesis of Ehlers–Danlos Syndrome and Related Connective Tissue Disorders” focus on different clinical, genetic and molecular aspects of several EDS subtypes and some related disorders, offering novel findings and future research and nosological perspectives.

MDPI

Order Your Print Copy
You can order print copies at
www.mdpi.com/books/reprint/2603

MDPI Books

Publishing Open Access Books & Series

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.

MDPI AG
St. Alban-Anlage 66
4052 Basel
Switzerland
Tel: +41 61 683 77 34
www.mdpi.com/books
books@mdpi.com