Special Issue

Primary Ciliary Dyskinesia: Genetics, Molecular Mechanisms, Diagnostic and Therapeutic Perspectives

Message from the Guest Editors

Primary ciliary dyskinesia (PCD) is the flagship ciliopathy caused by the genetically determined dysfunction of motile cilia. The symptoms are not specific, and the genetic background is heterogeneous. The whole ciliome (cilia genome) consists of thousands of genes. including those encoding proteins involved in cilia biogenesis—a multi-step process involving signaling factors. The pace of identification of PCD-related genes experienced a rapid acceleration during the last several years, mostly due to the introduction of new technologies such as whole-exome sequencing. To date, PCD-causing mutations in about 50 genes have been identified, and the number of PCD-related genes is bound to increase. Even though the genetic cause of approximately one-third of PCD cases remains unknown, the current knowledge can be used to design schemes of molecular testing that enable correct and effective PCD diagnosis. We warmly invite the contribution of original and review papers to this Special Issue of IJMS, which is basically devoted to PCD, but is in fact intended to present a wide spectrum of ciliary research related to primary ciliary dyskinesia.

Guest Editors

Prof. Michał P. Witt

Prof. Ewa Ziętkiewicz

Dr. Zuzanna Bukowy-Bieryłło

Deadline for manuscript submissions

closed (30 December 2021)



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International Journal of Molecular Sciences Editorial Office MDPI, Grosspeteranlage 5 4052 Basel, Switzerland Tel: +41 61 683 77 34 ijms@mdpi.com

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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.

Editor-in-Chief

Prof. Dr. Maurizio Battino

Department of Odontostomatologic and Specialized Clinical Sciences, Sez-Biochimica, Faculty of Medicine, Università Politecnica delle Marche, Via Ranieri 65, 60100 Ancona, Italy

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