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

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Deadline for manuscript submissions:
closed (30 December 2021)

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.

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Message from the Editor-in-Chief

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