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Clinical Molecular Genetics in Hematologic Diseases

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Deadline for manuscript submissions: 20 March 2025

Message from the Guest Editors

Hematologic diseases comprise a wide spectrum of disorders affecting blood and blood-forming organs, including both cancerous and noncancerous diseases. Many noncancerous hematologic diseases are caused by inherited genetic aberrations. For example, Factor Five Leiden thrombophilia, a blood clotting disorder manifesting as deep vein thrombosis in the legs or pulmonary embolism in adults, is caused by a heterozygous or homozygous c.1601G>A (p.Arg534Gln) variant in the F5 gene, which is inherited from an affected parent in an autosomal dominant manner. Hemophilia A, a type of bleeding disorder manifesting mostly as prolonged bleeding time after injuries and spontaneous bleeding, is caused by various mutations of the F8 gene and inherited in an X-linked manner. Many cancerous hematologic diseases, or hematologic malignancies, have been characterized by recurrent genetic alterations presenting in various forms. The precise detection of these genetic aberrations plays a critical role in the establishment of diagnosis and the optimal clinical management of hematologic diseases.



mdpi.com/si/213750







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Message from the Editor-in-Chief

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