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Diagnosis and Treatment of Rare Diseases

Guest Editor:

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Deadline for manuscript submissions:
closed (25 January 2024)

Message from the Guest Editor

Dear Colleagues,

Despite their low prevalence, clinically defined rare diseases (RDs), of which there are over 6000 affecting 300 million people worldwide, share certain characteristics (i.e., they have a significant diagnostic delay, they are usually chronic and incapacitating, and the majority have a genetic basis, high morbidity rate, and limited therapeutic options). Therefore, they usually have a deep impact on the quality of life of patients and caregivers.

Some of the priorities that deserve further development include: better knowledge of the natural history and pathophysiological mechanisms of RDs, improving access to multidisciplinary medical care and orphan drugs, increasing awareness among healthcare professionals, and enhancing genetic and molecular characterisation for a large number of undiagnosed RDs.

For this Special Issue, we welcome the submission of original research articles and up-to-date reviews on the described topics.

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Editors-in-Chief

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