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Discovering the World of Rare Diseases: 2nd Edition

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Deadline for manuscript submissions:
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Message from the Guest Editors

Rare diseases, although individually uncommon, can affect a significant proportion of the population (5–8% of the global population, with 7.9% of those younger than 25). In Europe, the European Medicines Agency specifies that rare disorders include those with a prevalence of less than 5 in 10,000 people (equivalent to less than 1 in 2,000).

The exact number of rare diseases is variable, ranging from 5,000 to 8,000, of which 80% have a genetic etiology.

A report from the 2005 European Conference on Rare Diseases, evaluating the life expectancy of patients affected with 323 rare diseases, concluded that 25.7% of rare diseases could be considered potentially lethal before 5 years of age, and that a further 36.8% lead to a reduced life expectancy. Conversely, only about a third (37.5%) are associated with a normal lifespan.

We are pleased to invite you and your coworkers to contribute to this Special Issue with original research reports, reviews, or meta-analyses on the topics of rare diseases, evaluating the incidence and prevalence of rare diseases in different countries, diagnostic approaches and delays, standard protocols of care, and national legislation and registries.

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Message from the Editor-in-Chief

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