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Rare Disease Genes and Genomics

Guest Editor:

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Deadline for manuscript submissions:

closed (28 February 2021)

Message from the Guest Editor

Dear Colleagues,

Rare and undiagnosed diseases are an extremely important and rewarding topic of research not only in medical care but also in basic and translational science. Nevertheless, there are about 4000–6000 different rare diseases that remain undiagnosed and about 10,000 genes that have not yet been annotated. The best way to progress the science in this field is to work with those who are affected by these rare diseases.

In this Special Issue, papers reporting original research on humans related to new gene discovery, regulatory mechanisms, and discoveries associated with new genomic approaches will be published. New phenotypes that have not been observed to date are welcome as well. We welcome original research on rare cancers, but reviews are also invited.

Prof. Dr. Bela Melegh Guest Editor













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Editor-in-Chief

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Message from the Editor-in-Chief

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