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Rare Diseases - Molecular Mechanisms and Therapeutic Strategies (III)

Guest Editors:

Prof. Dr. Lidia Larizza
Experimental Research Laboratory of Medical Cytogenetics and Molecular Genetics, IRCCS Istituto Auxologico Italiano, Via Ariosto 13, 20145 Milan, Italy

Prof. Dr. Maria Vittoria Cubellis
Dipartimento di Biologia, Università Federico II, 80126 Napoli, Italy

Deadline for manuscript submissions:
closed (31 December 2020)

Message from the Guest Editors

Dear Colleagues,

A rare disease is any disease that affects a small percentage of the population. The quantification of "small" is variable and, and represents an artificial border that will necessarily change with the diffusion of genetic screenings. More than 5000 rare diseases have been described. Non-sense mutations, deletions and insertions, abolish the function of the affected proteins, but mis-sense mutations have variable effects that go from complete inactivation to mild reduction of activity. At present, more than 70,000 mis-sense mutations have been reported. Taken together, these findings imply that there are different genotypes and phenotypes for any given disease. Bare figures give a flavor of the great challenge represented by rare diseases both in terms of diagnosis and therapy.

We are looking for papers that look into rare diseases with a genetic, biochemical or bioinformatic approach. Papers addressing specific pharmacological therapies for rare diseases are warmly welcomed.

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Editor-in-Chief

Prof. Dr. Maurizio Battino
Department of Odontostomatologic and Specialized Clinical Sciences, Sez-Biochimica, Faculty of Medicine, Università Politecnica delle Marche, Via Ranieri 65, 60100 Ancona, Italy

Message from the Editor-in-Chief

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International Journal of Molecular Sciences Editorial Office
MDPI, Grosspeteranlage 5
4052 Basel, Switzerland

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