

Special Issue

Rare Diseases: Molecular Mechanisms and Therapeutic Strategies

Message from the Guest Editors

A rare disease is any disease that affects a small percentage of the population. The quantification of "small" is variable and, in any case, 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.

Guest Editors

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