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Genotype-Phenotype Study in Disease

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Message from the Collection Editors

The study of inter-individual variability is essential for precision medicine. The study of polymorphic variants has become decisive in the understanding of mechanisms that underpin various multifactorial diseases (i.e., diabetes, obesity, cancer, and cardiovascular disease).

All diseases have different degrees of genetic heterogeneity, for which the severity of the disease varies from individual to individual. The same pathological phenotype could be determined by different mutations in different genes. In multifactorial diseases, however, genetic heterogeneity is much more widespread and frequent, so genes contribute to a different extent to the expression of phenotype.

Some genes do not cause disease but rather affect or cause particular aspects of the disease by behaving as modifying genes.

More recently it has been proposed that genetic variation may also explain some of the other features of clinical phenotype, such as disease duration.

Although the genotype–phenotype link is not always clear, it allows improved therapy, evaluation of individual drug response, and understanding of the adaptive response of organisms to environmental stimuli.













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Message from the Editor-in-Chief

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