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Polymorphism

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Synonyms

Alterations; Alternate forms; Variant

Definition

Polymorphisms are alterations of genetic material that results in a variety of forms (alleles) that might take on different functional roles. Genetic polymorphisms result from several mechanisms. A common source of polymorphisms is mutations due to errors in DNA replications and repair. For example, replication slippage may cause variable number tandem repeat (VNTR) polymorphisms at microsatellites. Polymorphisms may also arise from exposure to mutagens (for e.g., radiation) in the external and internal environment.

Polymorphisms occur in different sizes. A single nucleotide polymorphism (SNP) is the result of a single base pair change. Larger alterations may result from restriction fragment length polymorphisms (RFLPs), simple sequence repeats (SSRs), or copy number variants (CNVs). Association studies employ genetic polymorphisms to identify genetic risks of disorders. For instance, several CHRNA5/A3/B4 polymorphisms have been associated with early onset tobacco and alcohol initiation (Schlaepfer et al. 2008).

Cross-References

- ▶ DNA
- ▶ Gene
- ▶ Genotype

References and Readings

Schlaepfer, I. R., Hoft, N. R., Collins, A. C., Corley, R. P., Hewitt, J. K., Hopfer, C. J., et al. (2008). The CHRNA5/A3/B4 gene cluster variability as an important determinant of early alcohol and tobacco initiation in young adults. *Biological Psychiatry, 63*, 1039–1046.

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