Review on Gene polymorphism

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Gene polymorphism is distinction in DNA sequence among individual patients, groups, or populations. Sources like SNPs, sequence repeats, insertions, deletions and recombination [1-4]. Genetic polymorphism is also the result of chance processes, or might have been induced by external agents (such as viruses or radiation) [5-8]. If deference in DNA sequence among patients has been shown to be related to disease, it will sometimes be known as genetic mutation. Changes in DNA sequence that are confirmed to be caused by external agents are typically known as "mutations" instead of "polymorphisms" [9-14].

Genetic mutations are a form of genetic polymorphism. Genetic mutation is a permanent alteration within the polymer sequence that produces up a gene, specified that the sequence differs from what is found in majority of individuals [15-19]. Mutations will have an effect on anywhere from one polymer (DNA) building block (base pair) to an oversized phase of a chromosome that features multiple genes [17,20-23].

Single nucleotide polymorphism may be a single base mutation in Deoxyribonucleic acid and SNPs are the common source of genetic polymorphism [24-27]. Genetic polymorphisms of CYP2C9 and CYP2C19 are shown to have clinical consequences leading to toxicity of some medicine within the affected individual, and will alter efficacy of alternative medicine [28-31].

Polymorphism in drug-metabolising enzymes plays importance for interindividual variations in drug medical care. Variant genes inflicting hyperbolic, diminished or abolished drug metabolism influences the drug response and risk for adverse drug reactions [29,32]. The genetic alterations embody single-nucleotide polymorphisms and gene copy number variations. This variability is currently significance throughout drug development and of importance for routine drug prescription. There are many different drugs contain pharmacogenomic labels where genotyping before prescription is necessary or recommended. Predictive genotyping of drug-metabolising genes can thus help to design individualised, safer and a lot of economical drug therapies [33-35].

Genetic variation or polymorphisms in the miRNA pathway are related to the prognosis and progression of diseases and drug responses and are increasing as powerful tools to review the biology of diseases [36,37]. MicroRNA polymorphisms might probably interfere with miRNA-mediated regulation of cellular functions and may available not only in the miRNA target gene but also in pri-, pre-, mature-miRNA sequences, in the genes involved in miRNA biogenesis and in miRNA cis-regulatory components. The
invention of the role of miRNA in drug resistance and miR-polymorphisms to predict drug response results to the development of a new branch in biomedical science called miRNA pharmacogenomics \[38-40\]. Detection of miRNA-polymorphisms will probably improve detection, treatment and prognosis in patients and has profound implications within the fields of pharmacogenomics and personalised medicine.

Pharmacogenetics is concentrated on pharmacological consequences of a single chromosomal mutation, pharmacogenomics tries to at the same time contemplate various genes and their mutual interaction \[41\]. Genotyping for CYP polymorphisms provides necessary genetic information that facilitates to understand the results of xenobiotic on human body. For drug metabolism, the main important polymorphisms are those of the genes coding for CYP2C9, CYP2C19, CYP2D6, and CYP3A4/5, which can result in therapeutic failure or severe adverse reactions \[42\]. Genes coding for CYP1A1, CYP1A2, CYP1B1, and CYP2E1 are among the foremost liable for the biotransformation of chemicals, particularly for the metabolic activation of pre-carcinogens.

The toxicity of medicine due to genetic polymorphism is major issue in pharmaceutical products development and clinical use. Pharmacogenetics is that the study of genetically controlled variations in drug response \[43\]. Gene polymorphism includes monogenic variation due to gene variation at a single gene, polygenic is due to the variation happened at two or more genes.

Polymorphisms of genes concerned in innate and adaptive immunity have become an object of major interest in regard to hematopoietic stem cell transplantation (HSCT) complications. Vitamin D receptor (VDR) gene polymorphisms and the risks for various breast and ovarian cancers have been reported in many epidemiological studies. SNPs occur normally throughout a person’s DNA. They act as biological marker to find out the genes that are associated with disease \[44\]. SNPs are produced within the gene or near to the regulatory region of a gene and acts as major role in disease by causing harmful to the gene’s function. Researchers have found SNPs that may help predict an individual’s response to certain drugs, susceptibility to environmental factors such as toxins, and risk of developing particular diseases.

Genetic polymorphisms of drug-metabolizing enzymes make to distinct subgroups within the population that differs in their ability to perform bound drug biotransformation reactions. Polymorphisms area unit generated by mutations within the genes for these enzymes, that causes slashed, increased, or absent accelerator expression or activity by multiple molecular mechanisms \[18,24,44\]. Genetic polymorphisms are represented for a large type of drug and xenobiotic metabolizing enzymes.

Genetic polymorphism has been coupled to 3 categories of phenotypes supported the extent of drug metabolism. in depth metabolism (EM) of a drug is characteristic of the conventional population; poor metabolism (PM) is related to accumulation of specific drug substrates and is typically an autosomal recessive trait requiring mutation and/or deletion of both alleles for phenotypic expression; and ultra-extensive metabolism (UEM) results in increased drug metabolism and is an autosomal dominant trait arising from gene amplification \[45\]. The main reason for interindividual variation of drug effects is genetic variation of drug metabolism.

Extreme genetic polymorphism maintained by equalization choice (so referred to as a result of several genes area unit maintained in a very balance by a mechanism of rare allele advantage) is intimately related to the vital task of self/non-self-discrimination. Wide disparate self-recognition systems of plants, animals and fungi share many general options, together with the upkeep of enormous numbers of alleles at comparatively even frequency, and persistence of this variation over terribly long term periods \[46\]. Due to the evolutionary dynamics of balanced polymorphism are very different from those of neutral genetic variation, data on balanced polymorphism have been used as a novel source for inference of the history of populations.

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