

# DNA Polymorphisms

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## Introduction

DNA polymorphisms are the various DNA sequences among individuals, groups, or populations. Polymorphism at the DNA level includes a good range of variations from single nucleotide change, many base pairs, and repeated sequences.

replication. XPD works by cutting and removing segments of DNA that are damaged thanks to things like cigarette smoking and inhalation of other environmental carcinogens. Asp312Asn and Lys751Gln are the 2 common polymorphisms of XPD that end in a change during a single aminoalkanoic acid. This variation in Asn and Gln alleles has been associated with individuals having a reduced DNA repair efficiency. Several studies are conducted to ascertain if this diminished capacity to repair DNA is said to an increased risk of carcinoma. These studies examined the XPD gene in carcinoma patients of varying age, gender, race, and pack-years. The studies provided mixed results, from concluding individuals who are homozygous for the Asn allele or homozygous for the Gln allele had an increased risk of developing carcinoma, to finding no statistical significance between smokers who have either allele polymorphism and their susceptibility to carcinoma. Research continues to be conducted to work out the connection between XPD polymorphisms and carcinoma risk.

Asthma is an disease of the lungs and quite 100 loci are identified as contributing to the event and severity of the condition. By using the normal linkage analysis, these asthma correlated genes were ready to be identified in small quantities using genome-wide association studies (GWAS). There are variety of studies looking into various polymorphisms of asthma-associated genes and the way those polymorphisms interact with the carrier's environment. One example is that the gene CD14, which is understood to possess a polymorphism that's related to increased amounts of CD14 protein also as reduced levels of IgE serum. A study was conducted on 624 children watching their IgE serum levels because it associated with the polymorphism in CD14.

The study found that IgE serum levels differed in children with the C allele within the CD14/-260 gene supported the sort of allergens they frequently exposed to. Children who were in regular contact with house pets showed higher serum levels of IgE while children who were regularly exposed to stable animals showed lower serum levels of IgE. Continued research into gene-environment interactions may cause more specialized treatment plans supported a person's surroundings.

Gene polymorphisms can occur in any region of the genome. the bulk of polymorphisms are silent, meaning they are doing not alter the function or expression of a gene. Some polymorphism is visible. for instance, in dogs the E locus can have any of 5 different alleles, referred to as E, Em, Eg, Eh, and e. Varying combinations of those alleles contribute to the pigmentation and patterns seen in dog coats.

A polymorphic variant of a gene can cause the abnormal expression or to the assembly of an abnormal sort of the protein; this abnormality may cause or be related to disease. for instance, a polymorphic variant of the gene encoding the enzyme CYP4A11, during which thymidine replaces cytosine at the gene's nucleotide 8590 position encodes a CYP4A11 protein that substitutes phenylalanine with serine at the protein's aminoalkanoic acid position 434. This variant protein has reduced enzyme activity in metabolizing arachidonic acid to the blood pressure-regulating eicosanoid, 20-hydroxyeicosatetraenoic acid. A study has shown that humans bearing this variant in one or both of their CYP4A11 genes have an increased incidence of hypertension, ischaemic stroke, and arteria coronaria disease.

Most notably, the genes coding for the main histocompatibility complex (MHC) are actually the foremost polymorphic genes known. MHC molecules are involved within the system and interact with T-cells. There are quite 800 different alleles of human MHC class I and II genes, and it's been estimated that there are 200 variants at the HLA-B HLA-DRB1 loci alone.

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