Human Genetic Variation

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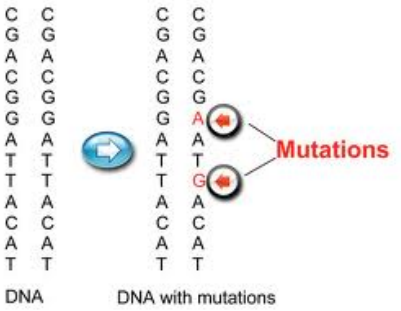
Genetic Diversity of Humans

The total amount of genetic characteristics observed within the human species is known as Human genetic variation. Genetic differences are found in humans and it is noted between 2 humans and with the population level.

Multiple variants of gene, in the human population lead to polymorphism. We can't see any two genetically identical humans. Genetic fingerprinting is the latest perfect technology to find out the differences between any two or more individuals even from closely related individuals. Geographical area also plays an important role in the genetic variations.

Causes of differences between individuals include the exchange of genes during meiosis and various mutational events. Natural selection has an adaptive advantage to individuals in a specific environment.



Mutations

It is also the major cause of genetic variations. The main cause of mutation is is genetic drift and this causes the random changes in the gene pool. Founder effect and genetic drift had an important influence for the differences between human populations.

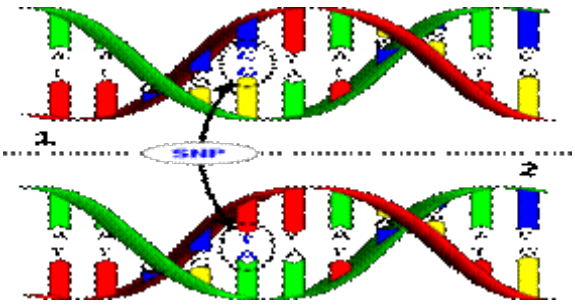
For evolutionary significance and medical applications knowing about genetic variation is very important. The study about the variations help the scientists to understand about population migrations and the biological relations between different individuals.

Genetic variation

Genetic variation occurs both within and among populations.

Measures of variation

"Genetic variation among individual humans occurs on many different scales, ranging from gross alterations in the human karyotype to single nucleotide changes."



Single Nucleotide Polymorphisms– SNP

DNA molecules differ by a single base-pair location.

Nucleotide diversity is based on single mutations called Single Nucleotide Polymorphisms (SNPs).

The nucleotide diversity between humans is about 0.1%, which is 1 difference per 1,000 base pairs. It is estimated that a total of 10

million SNPs exist in the human population of which at least 1% are functional.

Copy number variation:
It is estimated that approximately 0.4% of the genomes of unrelated people typically differ with respect to copy number. With copy number variation human to human genetic variation is estimated to be at least 0.5%. Copy number variations are usually inherited but there is a hence for it to arise during development.

Epigenetics:
This type of variation arises from chemical tags, the epigenetic markings attached to DNA. These act as switches that control how the reading of genes.

Genetic variability:
Individual genotypes in a population to vary from one another.

Variable Number Tandem Repeats
A variable number tandem repeat (VNTR) is a location in a genome where a short nucleotide sequence is organized as a tandem repeat. Their analysis is useful in

- Genetics research
- Biology research
- Forensics
- DNA fingerprinting.

There are two principal families of VNTRs:

1. microsatellites - repeats of sequences less than about 5 base pairs in length
2. minisatellites- involve longer blocks

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