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Introductory Chapter: Genetic Variation - The Source of Biological Diversity

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1. Introduction

Genetic diversity is usually defined as the number of genetic characteristics (alleles and genotypes) in a species [1]. In this context, analyzing the genetic diversity in populations is essential to understand evolutionary and adaptative process for most species [2]. The genetic diversity is also very useful to implement conservation strategies and crop management. This is also the source of disease resistance of natural populations, as it is the font of drug resistance by many pathogens.

Genes are DNA fragments that encodes some biological information, usually coding a protein or a RNA. Genes can be represented as a sequence of nucleotides that can be expressed in a living organism. Most genes have small nucleotide sequence differences among individuals. These differences are called genetic polymorphism [3]. Some of these polymorphisms may affects how proteins works and how the proteins interacts with subtracts and other proteins. The different gene forms caused by genetic polymorphisms are called alleles.

The genetic diversity has three different sources: mutation, recombination and immigration of genes. Mutation is the driving force of genetic variation and evolution. There are three types of DNA mutations: base substitutions (also called point mutations), deletions and insertions (**Figure 1**) [4].

2. Base substitutions (point mutations)

The point mutations are also subdivided in three groups: (1) Silent mutations: when the nucleotide substitution does not change the aminoacid in the polypeptide sequence; (2) Missense mutations: when occurs aminoacid change, that can be classified in conservative (when the change results in an amino acid from the same physical–chemical group) and non-conservative (when the change results in a different physicochemical aminoacid group); (3) Non-sense mutations: when the nucleotide modification results in a stop codon (**Figure 2**).

Single mutations are very important in population genetics and evolution. They are the mainly source of DNA polymorphic sites, which provides information for many inferences and analysis such as nucleotide and haplotype diversity, allelic diversity, genetic distance, heterozygosity, etc. These parameters are very important to elucidate evolutionary process in populations across time and space. Genes with high levels of polymorphism can be applied to genetic population studies, while genes with moderate and low polymorphic levels can be used for phylogeographic and phylogenetic inferences [5].

Figure 1.
Illustrative scheme of DNA mutations types. Source: Google Images.

Figure 2.
Types of point mutations (nucleotide substitution) in DNA molecule. Source: Google Images.

Single mutations are also important for health: many missense mutations can be deleterious and resulting in a disease or metabolic disorder. Another thing to be considered is that single mutations can also provide adaptative vantages such as pathogen resistance, xenobiotic tolerance and fitness improvement [6]. So, detecting point mutations in the organisms can be very useful to implement many strategies such as biodiversity conservation, crop management and infectious disease monitoring.

3. Recombination

In eukaryotes, genetic recombination is the aleatory change of genetic material resulting from the meiosis process, also called crossing-over or permutation. This type of recombination consists in break and rejoining homologous regions of pared chromosomes between the Prophase I and Metaphase I from meiosis division (**Figure 3**). Many combinations can be performed among gene exchange between two individuals [7]. Although prokaryotic species does not have chromosomes conjugation is performed by these beings by one of these three process: (a) Conjugation: when the DNA is transferred by tube after cells contact; (b) transduction: the DNA is inserted accidentally from one bacterium to another by a virus; and (c) transformation: when the bacterium receives exogenous DNA from the environment [8].

Recombination is very important because it makes new combinations of the existent alleles. Many effects of the DNA rearrangement can be good for species and populations, once it can improve adaptation. However, some recombination events can be unfavorable if it breaks apart important and beneficial alleles in the sisters chromatids [9]. The recombination rate is positively correlated with

Figure 3.
Crossing-over scheme in homologous chromosomes. Source: Google Images.

nucleotide diversity, which increases genetic variation and resulting in purifying deleterious mutations.

4. Deletions and insertions

Nucleotide deletions and insertions are types of mutation that changes the number of DNA base in genome. Deletions changes the base number by removing pieces of DNA and insertions alters the base number by adding pieces of DNA [10]. Often, these kind of mutations results in a gene that encodes a protein that does not function properly (**Figure 4**). When the deletions/insertions change the gene's reading frame they are called *frameshit* mutations.

Insertions and deletions can be particularly hazardous when occurs in an exon region, which is the coding segment of a gene. Due to multiple new aminoacid after translation, the protein function may be affected [11]. Knowing the rate of insertion-deletion mutations are crucial to understanding evolutionary process, such as natural selection, especially in coding regions due to the protein disruption that is usually caused.

5. Gene immigration

Gene immigration, or gene flow, is the transfer of genetic material from one population to another by migration of individuals or gametes. This can alter genetic diversity by changing allelic frequencies in populations [12]. Gene flow is essential to prevent population diverging. When gene flow is interrupted by physical (geographical) barriers, allopatric speciation tends to occur. Population gene flow can be measured by the formula:

$$Nm = \frac{\left(\frac{1}{F_{st}} \right) - 1}{4}$$

Figure 4.
Illustrative example of a DNA insertion modifying all the subsequent codons. Source: Google Images.

Where Nm refers to the number of migrants per generation; F_{st} is the degree of genetic differentiation.

When F_{st} is 1, there is a strong differentiation among populations. Gene flow is also very important to reduce genetic drift effects. Due to this particularity, the gene flow is extremely important for conservation genetics.

6. Final considerations

Genetic diversity is a very important feature of living organisms. It serves for population adapting to environment, once that how higher is the allelic variation, it is more likely that individuals display adaptative characteristics that suits to the environment. So, genetic diversity is essential for species survival.

Molecular markers, amplification and DNA sequencing technologies are improving an incredible advance in access genetic variation. The number of sequences and genomes deposited in the databases grows exponentially, generating an enormous amount of information to be studied and analyzed. The knowledge resulting from this information has innumerous applications and has been promoting a huge revolution in several areas of the biological, agrarian and health sciences. The way we understand, analyze and deal with biodiversity has been intensely modified and deepened by the advancement of genetics. In this context, knowledge about the genetic diversity of organisms will bring solutions to various problems and issues involving living beings.

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