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Chapter

Introductory Chapter: Population Genetics - The Evolution Process as a Genetic Function

Rafael Trindade Maia and Magnólia de Araújo Campos

1. Introduction

Population genetics is defined as the sub-area of biology that studies the distribution and change in frequency of alleles. The population genetics is also the basis of evolution, and it has been established as a science; its main founders were JBS Haldane, Sir Ronald Fisher, and Sewall Wright. Since 1966, from the pioneering work of Fisher, Haldane, and Wright, the population genetics had accumulated a large mathematical theory, statistical tools, laboratory techniques, molecular markers, and huge information of polymorphisms in databanks [1]. The main concept in population genetics is focused on the Hardy-Weinberg theorem (also known as Hardy-Weinberg theorem or Hardy-Weinberg law). This central theorem preconizes that if the population size is large, with random mating, and mutation, selection, and migration are not significant, the allelic frequencies do not change over the generations. If not, the allelic and genotype frequencies will change from one generation for applied studies and decisions can be provided by accessing the genetic variation in populations.

2. Evolutionary processes that interfere on genepool

Population genetics is an extremely useful tool for studies of microevolution, population dynamics, and conservation genetics. When accessing the genetic constitution of a population, several parameters can be conserved such as phenotypic frequencies, genotype frequencies, allelic frequencies, gene flow, heritability, genetic correlation, genetic diversity, heterozygosity, and several other indicators that allow an understanding of the genetic dynamics of the population in study. Through this information, it is possible to improve strategies for proper management and control or even more efficient conservation actions.

Among the evolutionary processes that interfere in the gene pool of populations, altering their genetic constituent, they include mutation, migration (with gene flow), natural selection, and genetic drift [2].

Mutation is the classic source of genetic variation, generating new alleles in a population. They direct the evolution and have different probabilities depending on the type of mutation, and this has implications for the evolution of genotypes. For example, if a G-A mutation occurs more frequently than A-G genotypes with the nucleotide A tend to be more common in the population. In summary, one can classify the mutations at synonymous and non-synonymous, that is, those that

entail changes in amino acid and those which do not cause changes in the peptide chain, also called neutral mutations. According to the theory proposed by Kimura neutrality, most mutations which attach the populations are of neutral type, since most non-synonymous mutations cause deleterious effects on the phenotypes of individuals [3].

Genetic drift is the change in gene frequency due to an event of random selection of individuals, usually in small populations (**Figure 1**). Genetic drift can have serious consequences for the population and can cause alleles to permanently disappear from a population, reducing their genetic variability. Drift can occur in two ways: the founder effect and bottleneck effect. The founder effect is the decrease of genetic diversity due to a new population established by few individuals [4, 5],



Figure 1. *Genetic drift. Source: Google images.*

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while bottleneck effect is the reduction of population size by environmental events (earthquakes, famines, floods, disease, fires, or droughts) [6].

The impacts of genetic drift in a population will vary according to the effective population size (N_e) , which can be estimated by the following equation:

$$N_e = \frac{4N_f N_m}{N_f + N_m} \tag{1}$$

where N_e is the effective population, N_m is the number of breeding males, and N_f is the number of breeding females.

Migration with reproductive success (**Figure 2**) allows the introduction of new alleles into the population, thus altering their genetic structure. Gene flow is one of the main parameters to evaluate the degree of structuring between populations of the same species [7].

Natural selection, a process observed by Charles Darwin, is the adaptation of organisms to the environment. It acts through the selection of genotypic changes in a given population. Through natural selection, mutations that increase the chances of individuals surviving and procreating tend to be preserved and multiplied in populations (positive selection), and mutations that decrease population fitness (deleterious mutations) tend to be eliminated (purified selection).

The future of population genetics is very promising. The crucial progress in this field has showed that today, it is particularly relevant for the comprehension of genetic variation among populations from many species. Advances in population genetics will impact strongly the way we deal with biodiversity, pathogenic species, crops, and many other species.

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