

EVOLUTIONARY AND GENETIC ASPECTS OF BIODIVERSITY

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Summary

All living beings usually develop from single cells under the instruction of their genomes. All biodiversity has hence a developmental genetic basis and, ultimately, an evolutionary origin. Genes, evolution and biodiversity are thus intimately intertwined biological concepts. Here some basic genetic and evolutionary principles are outlined that appear essential to understand the origin and character of biodiversity. Genetic diversity is defined, recognized as an important aspect of biodiversity, and ways to measure it are briefly outlined.

Some basic terms and principles of population genetics, such as fitness, natural selection and inbreeding are described that are essential for understanding genetic aspects of evolution and biodiversity. Both chance events (genetic drift) and deterministic processes (natural selection) are introduced as important driving forces of evolution. Evolutionary mechanisms generating biodiversity are explained, with a focus on

speciation, which is considered a bridge between microevolution and macroevolution.

1. Introduction

Living beings have two especially stunning features, **complexity** and **diversity**. Attempts to understand the origin, structure and function of these features is at the very heart of biology. Given the laws of physics and chemistry all systems that make copies of themselves (replicators) may require a minimal amount of complexity. Currently living beings are the only efficient replicators we know of. Their diversity might just be a spin-off of that complexity. In any case this diversity cannot solely be explained from a functional (i.e. engineer's) point of view—almost all features of living organisms are a product of both necessity and chance during evolution.

All living beings—large and small ones, relatively complex and seemingly simple ones—usually develop from single cells under the instruction of their genomes. All biodiversity has hence a developmental **genetic basis** and, ultimately, an **evolutionary origin**. Genes, development, evolution and biodiversity are thus intimately intertwined biological concepts. To understand biodiversity, some basic knowledge about genetics as well as developmental and evolutionary biology is required.

Biodiversity is the sum total of all biotic variation. It has three main components: genetic diversity, species richness and ecosystem diversity. **Genetic diversity** is the most fundamental level of biodiversity; it refers to the degree of variation in all the genes for all individuals. **Species richness** means the number of species which occur within a particular taxonomic level (i.e. genus level, family level, etc.) in a geographic area.

Ecosystem diversity is the aggregate of different environmental types in a region. Ecosystems are the collection of all the plants and animals within a particular area, each differing in species composition, physical structure and function. For convenience, genetic and evolutionary aspects are in the following treated in separate subchapters, even though there is broad overlap of both, for example within population genetics.

2. Genetic Aspects of Biodiversity

2.1. Genes, Genotypes, and Hereditary Mechanisms

The hereditary information of living beings is carried by deoxyribonucleic acid (**DNA**) molecules. DNA consists of two complementary strands twisted around each other, and jointly constituting the famous double-helix structure. Each strand of DNA is a linear polynucleotide consisting of four types of **nucleotides**: adenine (A), guanine (G), thymine (T) and cytosine (C).

According to a very useful definition a **gene** is a sequence of DNA that is essential for a specific function. Three types of genes are distinguished: many genes are transcribed into ribonucleic acid (**RNA**) and then translated into proteins and are thus called protein-coding genes. Some genes are only transcribed, but not translated, and are hence called RNA-specifying genes. Genes that are not even transcribed are aptly termed

untranscribed genes. **Transcription** is the synthesis of a complementary RNA molecule on a DNA template and hence an essential step of gene expression. The process is carried out by enzymes called DNA-dependent RNA polymerases (RNAP).

The entire sequence of DNA in an organism is called its **genome**. The genome carries the genetic information of an organism in the linear sequence of the nucleotides. Many sexually reproducing organisms have two copies of each gene and are hence **diploid**. Different versions of the same gene are called **alleles**. A gene with only one type of allele is **monomorphic**, otherwise it is **polymorphic** (i.e. the gene has more than one allelic variant).

Different combinations of alleles are produced as a result of the physical exchange of DNA between two different chromosomes, a process termed **recombination**. New alleles originate by **mutation**, i.e. changes in the nucleotide sequence of DNA. Mutations can be neutral—having no effect on the fitness of an organism, or they can have negative effects; only in rare cases does a mutation increase the fitness of an organism—these cases are of enormous evolutionary importance, however.

DNA encodes the characteristics of different organisms at least in part by specifying the structure of **proteins** in the cells. Proteins are made from a linear sequence of **amino acids**. This amino acid sequence is synthesized by organelles termed ribosomes under the instruction of messenger RNA (mRNA) molecules that have previously been transcribed from DNA. The process of protein synthesis is called **translation**. During translation a triplet of nucleotides (termed codon) specifies a single amino acid.

Of the 64 possible codons 3 specify the end of proteins; the 61 others specify 20 different amino acids according to rules that are called ‘genetic code’. To meet the requirements of a cell protein synthesis is a highly regulated process. Regulation of gene expression can occur from the earliest steps of transcription until the maturation of the synthesized protein e.g. by posttranslational modifications.

The heritable form of a gene is called its **genotype**. The physical manifestation of a gene is called its **phenotype**. Almost 150 years ago Gregor Mendel, based on his work with garden pea plants, developed a number of principles, now referred to as **Mendel’s Laws** of inheritance. According to the ‘Law of Segregation’, each allele of a parent will be passed into separate gametes during meiosis. The ‘Law of Independent Assortment’ states that in each gamete, alleles of one gene segregate independently of all other genes, thus allowing for new allele combinations through recombination.

According to the ‘Law of Dominance’, each gene has two alleles, one inherited from each parent. Alleles are dominant, co-dominant or recessive in their expression. Dominant alleles mask the expression of recessive alleles, while co-dominant alleles are expressed simultaneously. Based on Mendel’s Laws it is possible to make predictions about the probability that a particular allele is passed to an offspring, and to predict the phenotypic expression of an allele in the next generation. Moreover, the genotype of the parents might be predicted from the phenotypes of the offspring.

Since the discoveries of Mendel, however, a number of other patterns of inheritance and

gene expression have been identified which deviate from the simple dominant/recessive scheme. For example, heterozygotes may show an intermediate between the homozygous phenotypes (incomplete dominance). Or rather than just one trait, a single gene may influence the seemingly unrelated expression of multiple phenotypic traits (pleiotropy). Often a trait is not influenced by a single gene, but by several or even many genes (polygeny). Note that a high number of important traits are influenced by many genes and the environment. If the trait of interest varies quantitatively, it is called a quantitative trait, and the respective genes are called **quantitative trait loci** (QTLs).

In the case of human beings, for example, traits such as susceptibility to certain diseases, body weight, body height, intelligence, and many more, are determined by quite a number of QTLs plus the environment. Sometimes one gene influences the phenotypic expression of another gene (epistasis); the phenotypic expression of an allele may also be dependent on the sex of the individual and might be directly tied to sex chromosomes (sex linkage). Organelles (mitochondria, chloroplasts) are usually transmitted by only the male or female lineage, which also leads to deviations from normal segregation patterns for all traits encoded on mitochondrial or chloroplast genomes.

2.2. Genetic diversity

2.2.1. Concepts, Importance

Today, biodiversity is too often considered being synonymous to **species diversity**. Historically, however, species diversity—developed in evolutionary biology and ecology—was only one of two different ideas that led to the development of the concept of biodiversity. **Genetic diversity**—developed within genetics of cultivated plants—was the other one. Therefore, genetic diversity is often mentioned with reference to agriculture and maintaining food security. No question that this is an important issue: genetic erosion of several crops has already occurred leading to the world's dependence for food on only a few species, so that just three crops—rice, maize, and wheat—account for more than 50% of the calories and the proteins that people derive from plants.

But genetic diversity is much more than that. Actually, it is the basis for all evolution. It provides the means for populations to adapt to their ever-changing environment. The more genetic diversity, the better the chance that at least some of the individuals within the population will have an allelic variant that is suited for the new environment, and will produce offspring and continue the population into subsequent generations. Populations with low genetic diversity can become so well adapted to local conditions that any environmental change may suffice to destroy them. Thus, for preserving biodiversity at all levels, genetic diversity is of great importance.

Genetic diversity originates as a result of recombination or mutation during the process of cell replication. Genetic diversity is an important aspect of biodiversity in all organisms. In the case of bacteria, however, biodiversity information is essentially unobtainable by any other means because the majority of bacteria cannot be cultured. Thus genetic diversity is the only way to comprehensively describe bacterial diversity in

any ecosystem. Molecular data have already revealed the vast scope of microbial diversity. Often sequences of 16S ribosomal RNA (rRNA)—or of the corresponding DNA—are determined to assess genetic diversity in bacteria. Unique rRNA sequences are termed **ribotypes**.

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Biographical Sketches

Günter Theißen is a Professor of Genetics at the Friedrich-Schiller-University of Jena, Germany. His general research interest is in understanding the mechanisms that brought about the complexity and diversity of life. The experimental work of his research group is focused on the role of MADS-box genes in the development and evolution of green plants, using a diversity of model systems that range from green algae via mosses, ferns and gymnosperms to flowering plants. Professor Theißen has received

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J. Albert C. Uy was born in Manila, Philippines on November 24, 1971, and moved to the United States of America in the 1980s. He studied at University of California at Berkeley, and received his AB in Integrative Biology in 1995. He then pursued graduate studies at the University of Maryland, working with Gerald Borgia on animal communication in bowerbirds of Australia and New Guinea. He received his Ph.D. from the Department of Biology in 2000. He was then awarded a National Science Foundation Postdoctoral Research Fellowship to work with John Endler at the University of California, Santa Barbara. In 2002 through 2004, he was at the Department of Biology, San Francisco State University as an Assistant Professor. He is now an Assistant Professor of Biology in the Department of Biology at Syracuse University New York, USA.. His current work aims to understand the role of animal communication in generating biodiversity, with special emphasis on tropical birds.

Uy has authored/coauthored several academic articles (e.g., *Evolution, Proceedings of the Royal Society of London, Nature*), as well as a popular article on bowerbirds of *Natural History* magazine. His work has been funded by the National Science Foundation, and the Committee for Research and Exploration, National Geographic.