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Some problems of biodiversity assessment at the genetic level: Genes and Genomes, or Genetics and Genomics?

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Abstract

Biodiversity may be approached at the genetic, species and ecosystem levels. Whole genomes are intelligent information-processing systems. We are progressing from a Constant Genome concept, subject to random, localized changes at a relatively constant mutation rate, to a Fluid Genome concept, subject to episodic, massive, and non-random reorganizations capable of producing new functional architectures. We discuss the demise of the gene concept, with particular reference to the Human Genome Project. We also question the undue reliance on the automatic sequencing of organic molecules for the understanding of the phenotype of organisms and of their genealogical relations. Under the molecular revolution the neo-Darwinian Genetic Theory of Evolution is inexorably giving way to a new, systemic, hierarchical, dynamic, organism and population-centered paradigm, Genomics or Phylogenomics. There is also much room for the expansion of microdiversity studies in the present to taxon and ecosystem-centered Macroevolutionary approaches in the full geological time dimension.
Key words: Intelligent Systems; Fluid Genome; Gene Concept; Human Genome Project; Macroevolutionary Theory; Microevolutionary Theory.

Resumo

ALGUNS PROBLEMAS NA AVALIAÇÃO DA BIODIVERSIDADE NA ESCALA GENÉTICA: GENES E GENOMAS, OU GENÉTICA E GENÔMICA? A biodiversidade pode ser abordada ao nível genético, específico e de ecossistemas. Genomas são sistemas inteligentes processadores de informação. Estamos progredindo da noção de um Genoma constante, sujeito a mudanças mutacionais localizadas e ao acaso, para um conceito fluído de Genoma, sujeito a reorganizações episódicas, extensas e não casuais, capaz de produzir novas arquiteturas funcionais. Discutimos o definhamento do conceito de gene, com referência particular ao Projeto Genoma Humano. Também questionamos a atual confiança excessiva no sequenciamento automático de moléculas orgânicas para o entendimento do fenótipo de organismos e das suas relações genealógicas. Sob a revolução molecular a Teoria Genética de Evolução Neodarwinista está inexoravelmente dando lugar a uma Teoria sistêmica, hierárquica, dinâmica, centrada no organismo e na população, a Genômica ou Filogenômica. Também sobra espaço para a expansão dos estudos de microdiversidade no presente para uma perspectiva Macroevolutiva, centradas nas espécies e ecossistemas, abrangendo a totalidade do tempo geológico.
Palavras-chave: Conceito de Gene; Genoma Fluido; Projeto Genoma Humano; Sistemas Inteligentes; Teoria Macroevolutiva; Teoria Microevolutiva.

Introduction

The term biodiversity was introduced by Wilson & Peter (1988) more than 20 years ago, and is now used to encompass a broad range of biotic scales, from genetic variation within species to biomes of the planet. It is frequently described in terms of numbers of genotypes, species, or ecosystems (Hooper et al., 2005).

Molecular genetic technologies have changed the way we describe and catalogue biological diversity (Mathews & Anker, 2009). Yet the interplay between genetic and morphological evolution is still elusive, while the origin and maintenance of biodiversity of ecological systems are still not fully understood (Vamosi et al., 2009). Thus understanding how biodiversity is generated has become a goal of basic science and a tool for the management and conservation of biological resources.

The most successful approaches at the genetic scale are mitochondrial genetic sequencing (Leung et al., 2009), phylogeography (Rocha et al., 2008; Avise, 2009), and DNA barcoding (Hebert et al., 2003a, 2003b; Sinniger et al., 2008), providing an impetus to the field of Phylogenomics (Philippe & Blanchette, 2006).

Notwithstanding these burgeoning programs if research, it remains unclear how well studies at the genome and population level fare across to the taxonomic and ecosystem scales. In this essay, we concentrate on problems with the basic units of biodiversity assessments.

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History

Early in the 21st century we should be completing an important paradigm shift from Classical Genetics to Molecular Systemic Biology. We will refer to this new paradigm as Genomics, or Phylogenomics, in order to contrast the molecular revolution with the orthodox Mendelian Genetics, developed over a century ago and then combined with Darwinism to form the 'Modern Evolutionary Synthesis" in the first half of the 20th century.

It all started in the 1950s with Barbara McClintock's pioneering studies of mutation and chromosome rearrangements in maize (e.g., McClintock, 1956). Her Nobel Prize winning efforts successfully linked genetic events with developmental changes in plants.

McClintock's tremendous foresight has been that cells are capable of coordinating and engineering their own genomes. Such cellular activities may be regularly programmed, or can be activated in response to environmental crisis. In other words, whole genomes within the cells become information-processing systems and act as true intelligent systems (Shapiro, 1991). The convergence between biology and information science now offers the potential for scientific investigation of possible intelligent cellular action in evolution. We may effectively attribute cells with properties of self-awareness and decision-making. And the existence of these capacities at the cellular level as well as the organism level should no longer be considered vague or mystical. Cellular cognition now has an experimental molecular basis (Shapiro, 1999).

These pioneer insights are being experimentally corroborated in all living domains of life in the flood of molecular work of the 90s (see E. Rosenberg, 1999).

Molecular Genetics

Molecular genetics is having a major impact on our views about evolution. We are shifting from an atomistic, static, mechanistic, Neo-Darwinian view of evolution, to a holistic, dynamic, systemic, hierarchical theory a much expanded Darwinian evolution.

However, the traditional view of a chemically constant Genome which evolves gradually by recombination and occasional point mutations formulated during the neo-Darwinian Modern Synthesis still has such a strong hold today on workers in molecular biology, that the implications of treating genetics and development as two sides of a coin are still confounding the majority of workers in the field. Many questions are being raised by molecular genetic studies that do not fit the classical concept of small differences in discrete genetic coding units. It is this reductionist Molecular Biology that is dying or gradually disappearing (Morange, 2008).

Phylogenomics

The Genome represents an integrated functional system, not just a collection of autonomous units called "genes". Instead of the conventional view of random mutations and independent selection at each locus, the new concept of the Genome offers much more than the sum of its basic parts. The dynamic and plastic Genome controls the rapid development of phenomena requiring the action of many "genes" simultaneously. Rather than providing just a bead of nucleotides along a chromosomal string, the Genome is hierarchically layered into several functional levels. This means that beyond the primary sequence of protein coding nucleotides, we find that the Genome is organized Lego-like into a hierarchy of modular assemblages of regulatory and coding motifs, in which many loci share in the functioning of the same higher order motifs. Genome-wide networks in the cell thus function dynamically to control developmental phenomena in the organism, providing us at last with a functional bridge between the cellular and the organism levels of organization of living matter. Cells also present a truly astonishing array of repair systems that serve to remove accidental and stochastic sources of mutation. These surprising mechanisms of homeostasis permit cells to protect themselves precisely against the kinds of accidental genetic change that, according to conventional evolutionary theory, should represent the basic sources of evolutionary variability. Our current knowledge of genetic change is thus fundamentally at variance with neo-Darwinist postulates. From the notion of a Constant Genome, subject only to random, localized changes at a more or less constant mutation rate, we are progressing to a Fluid Genome concept, subject to episodic, massive and non-random reorganizations capable of producing new functional architectures.

As just two examples of some of these higher order levels within the Genome, let us mention (1) the role of the tertiary structure of proteins in uncovering evolutionary relationships (Robertus, 1998; Babu et al., 2008), with the implication that amino acid sequences can often become quite flexible because many sequences can fold into similar structures, and (2) the gene action and activation roles of homeobox systems for coordinating multilocus gene expression along the body axis in living beings during development (Robert, 2001).

The demise of the gene concept represents a particularly interesting and illustrative consequence of the paradigm shift from Classical Genetics to Genomics, or Phylogenomics.

Human Genome Project

It is somewhat ironic that the Human Genome Project was announced worldwide as having become 97% mapped (Simpson et al., 2000), there appeared to be no general and universal genetic unit of the type suggested by classical genetics (Keller, 1998; Dietrich, 2000). Both the structural and functional concepts of the gene have become more confounded and more hopeless than ever (Griffiths & Neumann-Held, 1999). Biologists no longer believe in the existence of a non-ambiguous entity that can be called a "gene". Minimally, genes tend to be viewed more as "activated" by the organism rather than as actors controlling the organism, or more as processes rather than substances (Keller, 1998). The identification of alternative splicing and other forms of editing mechanisms operating on the level of protein synthesis and function, however, have also confounded attempts at a clear-cut functional definition of the gene. More recently, research on processes of methylation and gene imprinting even disturb accepted definitions of the genes as units of transmission (Keller, 1998). It is thus more reasonable today to speak of Genomes than of genes. The Human Genome Project has become the center of molecular biology in the last decade. The attempt to understand the role of "junk" DNA (the 95% or so of the human sequence that does not code for specific proteins or RNAs necessary for making proteins) might become one of the best reasons for continuing the Human Genome Project (Vicedo, 1998). Included in the "junk" DNA one finds introns, non-translated flanking regions associated with specific proteins, and a variety of repetitive sequences varying in length and location (Nowak, 1994). A. Rosenberg (1998) thinks that "junk" DNA is what makes the project pointless. Gould & Vrba (1998) suggest that repetitive sequences might be the genetic source of exaptations, i. e., organism characteristics that are co-opted rather than adapted. Some biologists (Caporale, 1984) have looked to "junk" DNA as the source of a "higher level genetic code". "Junk" DNA is thus forcing a re-conceptualization of the Genome. The focus of our attention must clearly be directed to access the evolutionary importance of changes in non-coding components of the Genome. The Human Genome Project will make it possible to experimentally address the conflict between reductionist and holistic accounts of biology in a fashion much clearer than has been possible previously (Grinnell, 2000).

It is becoming more and more evident that the structural sequence of nucleotides and individual amino acids along the DNA molecule is much less important than previously imagined for the coding of proteins and for the understanding of the final physical phenotype of organisms and for the establishment of their genealogical relationships. The important explanation for the complexity of species and ecosystems must be sought in the complicated process of expression of the Genome and in the form that proteins are expressed, synthesized and modified.

Consequently, much more important than methods that simply sequence the DNA molecule, is the search for new methods that correlate these sequences with the production of specific proteins, transitory and contingent molecules (Gayon, 1998) and, ultimately, phenotypic characteristics. Instead of thinking of a collection of individual isolated genes whose information is utilized in an automatic/mechanical fashion, we must now think about integrated, multigenic systems that can be turned on and off in a coordinated fashion according to the needs of the organism (Shapiro, 1991).

Conclusion

As more and more experimental molecular facts appear in rapid succession, the neo-Darwinian Genetic Theory of Evolution is being replaced by a new paradigm. The technological innovations brought fourth by the molecular revolution are playing their part in the development of a new, systemic, hierarchical, dynamic, organism and population-centered Microevolutionary Theory.

There still remains much room for the expansion of the organism centered views of genetic and population microdiversity in the present to the taxon and ecosystem levels of macrodiversity in both the present and the geological past (Peterson et al., 2007).

After the intense focus on genetics in the last century, we may hope that Darwin´s bicentenary year, 2009, will mark not only the rediscovery of Darwin (Stürzenbaum et al., 2009), but also the renaissance of evolutionary morphology (Budd & Olson, 2007), and the burgeoning of community ecology grounded by phylogenetic structure (Hardy, 2008). Our understanding of biodiversity and evolutionary changes should also be enhanced by looking beyond mutations and population genetics to consider the mechanisms, constraints and biases of development (Blumberg, 2009).

A full revolution from Microevolution to Macroevolution still lies ahead in the future.

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