Genotype, Phenotype and Ontogeny

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ABSTRACT

Building effective computational models of living systems requires both a sound conceptual basis and an accurate scheme of implementation. Basic biological principles must be clearly articulated to capture their essential features or logic but not reach beyond legitimate bounds. This paper analyzes the relationships among genotype, phenotype, and ontogeny, identifies pitfalls and gaps in these concepts, proposes to abandon the notion of simple mapping of genotype onto phenotype, and integrates them into a more complex, ontologically realistic model of development.

The gene concept in biology embodies two distinct roles, inheritance and development: genes carry traits from one generation to the next and genes encode proteins that specify traits. As vehicles of inheritance genes are entirely passive. However, expression of traits suggests a more active role without specifying what genes do or what they produce.

Resolving this ambiguity must focus on the cellular context in which genes operate during construction of the multicellular body. The phenotype (body) is a complex, highly ordered and temporally defined state, which requires energy to build and maintain. Genes provide templates for proteins necessary for harvesting energy, mediating exchange between a cell and its surroundings, repair, replication, and so forth. According to Harold [1], it is best to regard a cell "…as a spatially structured self-organizing system made of gene-specified elements". In other words, genes specify the elements but not the system in which the elements operate.

Ontogeny (Gk., *onto-*, existence, being + *-geny*, becoming) means literally "coming into being", constructing the multicellular body. As development progresses, the organism builds itself, and its phenotype (appearance, traits, organization, function, etc.) changes rapidly. The modular body plan enables cell-by-cell control of gene expression. Each cell operates in a microenvironment of other cells, the protein meshwork of the extracellular matrix (ECM), and tissue fluids. Cell clusters organize into pockets, cavities or layers as cells divide, grow, change shape, move about, and pull on the fabric of the ECM or adjacent cells to produce an environment suitable for subsequent steps of development. Microenvironments and control of gene expression are the basis for differentiation.

Some genes encode a single protein (or RNA molecule). Others encode several proteins (e.g., by alternative splicing), demonstrating that 1:1 mapping of genotype to phenotype is too simplistic. When extrapolated to a genome-wide perspective, genotype is clearly inadequate to specify phenotype, as recent cloning experiments confirm.

The relationship between genotype and phenotype in a developing embryo is complex, recursive, and linked to the environment (Figure 1). Phenotype is a higher ontological category than genotype because phenotype includes both the structure and function of components needed to carry out metabolism, communication, replication, repair and other higher order (emergent) properties. Not only is the genotype a representation of a subset of the phenotypic properties, embryonic functions include control of gene expression, which frequently involves signals from other cells, signals not genetically encoded but crucial to survival, growth, and development of the embryo.

GECCO'05, June 25–29, 2005, Washington, DC, USA.

Categories and Subject Descriptors

I.6.5 [**Simulation and Modeling**]: Model Development – *modeling methodologies.*

General Terms

Theory

Keywords

Genotype, Phenotype, Ontogeny

[1] Harold, F.M. *The Way of the Cell.* Oxford Univ. Press, Oxford, UK, 2001.

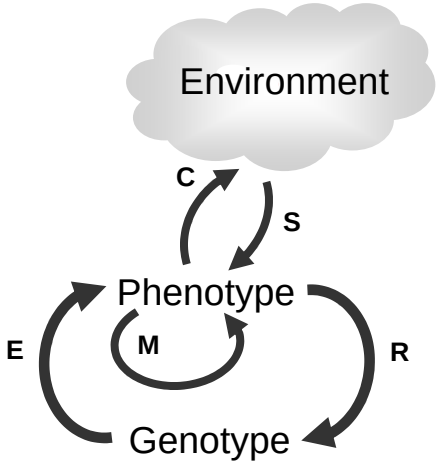

Figure 1. An integrated model for ontogeny.

Arrows: E, gene expression; M, Metabolism; C, cell signaling; S, sensory processes; R, gene regulation.