Wilhelm Johannsen's Genotype-Phenotype Distinction [1]

By: Peirson, B. R. Erick Keywords: Heredity [2] Genetics [3] G x E interactions [4]

Wilhelm Johannsen first proposed the distinction between genotype and phenotype in the study of heredity while working in Denmark in 1909. The distinction is between the hereditary dispositions of organisms (their genotypes) and the ways in which those dispositions manifest themselves in the physical characteristics of those organisms (their phenotypes). This distinction was an outgrowth of Johannsen's experiments concerning heritable variation in plants, and it influenced his pure line theory of heredity. While the meaning and significance of the genotype-phenotype distinction has been a topic of debate—among Johannsen's contemporaries, later biological theorists, and historians of science—many consider the distinction to be one of the conceptual pillars of twentieth century genetics. Moreover, some have used it to characterize the relationships between studies of development, genetics, and evolution [5].

Johannsen introduced the concepts genotype and phenotype in 1909 in his textbook on heredity research, titled *Elemente der exakten Ereblichkeitslehre* (The Elements of an Exact Theory ofHeredity [6]), and he developed them more fully in a 1911 paper titled "The Genotype Conception of Heredity". The concepts of genotype and phenotype were an outgrowth of Johannsen's pure-line breeding experiments on barley (*Hordeum vulgare* [7]) and the common bean (*Phaseolus vulgaris* [8]).

Johannsen's pure-line experiments began around the time that he accepted a position as lecturer of botany and plant physiology at the Royal Veterinary and Agricultural College in Copenhagen, Denmark, in 1892. In these experiments, Johannsen raised barley and bean plants that had been self-fertilized, and he measured the physical dimensions of the seeds in every generation. He found that he could separate these pure lines into distinct groups based on the characteristics of the seeds that they produced. Using a combination of pedigree and statistical analyses, Johannsen demonstrated that the group to which a plant belonged was a stronger predictor of the characteristics of the seeds that it produced than were the characteristics of its mother plant. When he first reported the results of his experiments in 1903, Johannsen referred to this group identity as its type, but in 1905 he rebranded it with the term genotype. He contrasted the genotype of a group or organism with its phenotype, defining the latter as the individual qualities of those organisms.

These experiments took place within a broader attempt among researchers of heredity to characterize the kinds and nature of variation in organisms at around the turn of the twentieth century. At the time, biologists disagreed about how to understand the relationship between variation, heredity, and evolutionary change. Followers of Darwin contended that evolution [5] resulted from the action of natural selection [9] upon continuous heritable variation. Others argued that continuous variation was rarely heritable, or that even if it was, such variation had limits so that it could not possibly be the basis for long-term evolutionary changes. Advocates of the latter position, emboldened by the so-called rediscovery of Gregor Mendel's theory of inheritance, argued that evolution [5] must proceed by discontinuous leaps. For Johannsen, the genotype-phenotype distinction contributed to the latter position: he took the genotype to be especially immutable, and he took the power of natural selection [9] as limited to sorting out pre-existing genotypes within heterogeneous natural populations.

The genotype-phenotype distinction was part of Johannsen's campaign against what he called the transmissionconception [10] of heredity, according to which the characteristics of individual organisms are transmitted directly to their offspring. Examples of the transmission conception [10] of heredity include Charles Darwin [11]'s theory of pangenesis [12], in which the changing tissues and organs of an organism continually modify its germinal material, thereby impressing the characteristics of that organism on its eventual offspring. Johannsen's concept of genotype, by contrast, was ahistorical: the phenotypic characteristics of a mother and her offspring would arise under the influence of the same hereditary disposition, passed from generation to generation, and immune to the environmental circumstances in which it was expressed. Johannsen's genotype-phenotype distinction has some similarity to August Weismann's late nineteenthc century distinction between the germ and soma, in that both thought that the causal interactions between an organism's hereditary disposition and its physical characteristics was unidirectional. Although Johannsen acknowledged this affinity with Weismann's ideas, he was unwilling to engage in what he considered to be unjustified speculations about the material basis of the genotype, as he argued Weismann had done.

Johannsen's genotype-phenotype distinction explicitly proscribed a relationship between the study of development and the study of heredity. According to Johannsen, the genotype of an organism gives rise to the organism's phenotype through the process of development, under the influence of the environment. Johannsen felt comfortable equating the genotype with the notion of *Reaktionsnorm* (norm of reaction) proposed by Richard Woltereck in Germany in 1909, which, for a domain of different possible environments in which an organism could develop, referred to the full range of potential variations in an adult characteristic for that organism. Under the schema of the genotype-phenotype distinction, developmental biology was construed as the study of how genotypes give rise to phenotypes. This construal ran contrary to the views of many embryologists at the time who saw heredity as a process or production, the mechanisms of which were inseparable from those of development itself.

The proper application of the terms genotype and phenotype was a subject of some dispute following its introduction. The zoologist Herbert Spencer Jennings [13] in the US, for example, interpreted the notions of genotype and phenotype as non-contrasting, arguing in a 1911 letter to the journal *Science* that while the abstract term genotype referred to the particular hereditary constitution of an organism, the phenotype more concretely referred to a group of individuals having the same physical characteristics. George Shull, also in the US, allegedly consulted Johannsen on the matter, and Shull responded to Jennings in 1912. Shull contended that the terms were equally abstract and opposed, the phenotype referring not to a group of physically similar individuals itself, but the shared characteristics that were the basis for delimiting the group. The relationship between genotype and phenotype—in terms of both its mechanistic and conceptual content—has been a recurring point of contention within evolutionary biology.

As the debate over the role of continuous and discontinuous variation inevolution [5] wore on, and as both experimental and theoretical advances led biologists to converge around the notion of the gene, the language of genotypes and phenotypes gained broad acceptance. Historian of science Jan Sapp has argued that the genotype-phenotype distinction served to alienate embryologists from mainstream heredity research, enabling a gulf between developmental biology and what would become the population genetic account of evolutionary change at the heart of the Modern Evolutionary Synthesis. Later debates about the units of selection, and about the relation between microevolutionary and macroevolutionary processes, were largely framed in terms of the genotype-phenotype distinction. Some theorists and philosophers have characterized the modern field of evolutionary developmental biology [14] as filling in the so-called black box between genotype and phenotype.

Sources

1. Allen, Garland E. "Heredity under an Embryological Paradigm: The Case of Genetics and Embryology."*Biological Bulletin* [15] 168 (1985): 107–21.
2. Churchill, Ferederick B. "William Johannsen and the Genotype Concept."*Journal of the History of Biology*7 (1974): 5–30.
3. Dunn, Loren C. "Johannsen, Wilhelm Ludvig." *Complete Dictionary of Scientific Biography.*7: 113–5.
4. Hall, Brian K. "Unlocking the Black Box between Genotype and Phenotype: Cell Condensations as Morphogenetic (Modular) Units." *Biology and Philosophy* 18 (2003): 219–47.
5. Jennings, Herbert Spencer. "'Genotype' and 'Pure Line'."*Science* 34 (1911): 841–2.
6. Johannsen, Wilhelm Lugvig. "Om Frøhviden og dens Udvikling hos Byg." [About the endosperm and its development in barley]. *Allgemeine Zeitschrift fur Bierbrauerei und Malzfabrik* 12 (1884): 625–750.
7. Johannsen, Wilhelm Ludvig. *Über Erblichkeit in Populationen und reinen Linien. Eine Beitrag zur Beleuchtung schwebender Selektionsfragen* [On heredity in pure lines and populations. A contribution to pending questions of selection]. Jena: Gustav Fischer, 1903.
8. Johannsen, Wilhelm Ludvig. *Elemente der Exakten Erblichkeitslehre* [The elements of an exact theory of heredity]. Jena: Gustav Fischer, 1909. http://dx.doi.org/10.5962/bhl.title.1060 [16] (Accessed December 7, 2012).
9. Johannsen, Wilhelm Ludvig. "The Genotype Conception ofHeredity [6]." *The American Naturalist* 45 (1911): 129–59.
10. Johannsen, Wilhelm Ludvig. *Falske Analogier med henblikk paa Lighed, Slaegtskab, Arv, Tradition og Udvikling*[False analogies with respect to similarity, kinship, tradition and development]. Copenhagen: J. H. Schultz, 1914.
11. Kim, Kyung-man. "On the Reception of Johannsen's Pure Line Theory: Toward a Sociology of Scientific Validity."*Social Studies Of Science* 21 (1991): 649–79.
12. Provine, William B. *The Origins of Theoretical Population Genetics*. Chicago: The University of Chicago Press [17], 2001.
13. Rheinberger, Hans-Joerg. "Heredity and its Entities around 1900."*Studies In History and Philosophy of Science Part A*39 (2008): 370–4.
14. Roll-Hansen, Nils. "The Crucial Experiment [18] of Wilhelm Johannsen." *Biology and Philosophy* 4 (1989): 303–29.
15. Roll-Hansen, Nils. "Sources of Wilhelm Johannsen's Genotype Theory."*Journal of the History of Biology*42 (2009): 457–93.
16. Sapp, Jan. "The Struggle for Authority in the Field ofHeredity [6] , 1900–1932: New Perspectives on the Rise of Genetics." *Journal of the History of Biology*16 (1983): 311–42.
17. Shull, George Harrison. 1912. "'Phenotype' and 'clone'."*Science* 35 (1912): 182–3. http://www.biodiversitylibrary.org/item/61279#page/349/mode/1up [19] (Accessed December 7, 2012).
18. Snorrason, Erling B. "Hansen, Emil Christian."*Complete Dictionary of Scientific Biography.*6: 99–101.
19. Wanscher, Johan Henrik. "An Analysis of Wilhelm Johannsen's Genetical Term 'Genotype' 1909–26."*Hereditas* 79 (1975): 1–4.
20. Yule, George. Udny. "Professor Johannsen's Experiments inHeredity [6]: A Review." *New Phytologist* 2 (1903): 235–42.

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Subject

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