

From Genotype to Phenotype: Understanding the Introduction of Phenotypic Variations by Random Mutation



Background

In biology, the term *phenotype* refers to the observable traits of individuals that arise from the causal interaction of their genotype with the environment. "Observable" is meant in the scientific sense and so is not limited to traits that are directly visible. Any testable trait, whether physical or behavioral, is part of an organism's phenotype so long as it has a genetic basis.

For instance, a scientist may be interested in whether a strain of bacteria can metabolize some sugar, say glucose. One organism that can perform this testable function—*ability to metabolize glucose*—is positive for the trait (Glu+) and a second that cannot is negative for it (Glu-). Some difference between these organisms' genotypes determines which phenotypes they exhibit; perhaps a gene in the first produces an enzyme that makes glucose metabolism possible or some genetic sequence in the second changes the timing of a metabolic mechanism. One may, of course, think of these possibilities as phenotypic traits themselves—again, any observable trait with a genetic basis will count as part of the organism's phenotype.

Similarly, in Avida-ED, phenotypic variation depends upon genotypic variations. As in nature, different sequences of instructions in an Avidian's genome produce whatever different traits it can exhibit, from the performance (or non-performance) of the possible functions of its virtual metabolism to its gestation time, viability and so on.

Your goal in this exercise is to look at how changes in genetic sequence can lead to variation of phenotypic functions. At this point don't worry about the particular mechanisms by which sequences of genetic instructions produce specific functions. Focus just on how genotype and phenotype relate to each other in general.







Assignment Tasks

• **Preparation**. Familiarize yourself with some of an Avidian's observable traits. While you have a population running in the Population Viewer you can select any single organism in the virtual Petri dish and see some information about what traits its genotype produces by looking in its Stats box. In the screenshot above, for instance, the selected magenta-colored organism in the dish (the organism's cell is outlined in green) with the genome named *aadeb* is displayed. The metabolic rate and gestation time for that genome are two phenotypic traits that are listed, but the main traits that are of interest in this exercise are the functions in the table. In this case, the organism is negative for all of these functions except for one (orn+). The box to the right of this shows the prevalence of organisms with these functions in the population as a whole. Pressing the respective label highlights (with a blue cell outline) all the organisms that are positive for that trait. (In the screenshot, the *nan* button is pressed.)

You should already be familiar with how to examine the genome of an Avidian by dragging it into the Organism Viewer. In the Organism Viewer you will see another phenotype table that displays the functions that the organism can perform. Additionally, the x axis below the genome display shows when in the execution of the genetic sequence each function is completed (e.g. function 3— Oro—at instruction cycle 26, in the figure below). Finally, you can observe some of the computational mechanism itself by looking at what is happening in the registers, stacks and input/output buffers while the genetic program is executing.

• **Demo**: Use the Population Viewer to see new phenotypes arising and spreading in the population.

- (i) Flip to settings. Set the per site mutation rate to 5%. Drag in the @ancestor from the freezer to the *Ancestral Organism(s)* box. Set repeatability mode to Experimental. Flip back to viewer.
- (ii) Click *Play* and watch the population until a variety of phenotypes have evolved. (It could take 1000 or more updates before two or three functions arise.) Pause the run.
- (iii) In the Population Viewer, explore the phenotypic traits of the organisms in your population using the methods described above.
- (iv) Save organisms (one each) with the following two phenotypic functions (nan+, orn+) to the Freezer. Be sure to give them identifiable names.







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• **Observe:** In the Organism Viewer, record the requested data for each. Note: To record the final five instructions, start by finding the instruction at the cycle number when the function completes and then list it and the four instructions prior to it in the (clockwise) order they occur. In the previous figure, the function Oro completes at cycle 26 (the i instruction) in the following sequence:

wzcagcycpujmccqwtsncktlcccqqcycoubyijzso... In that case the "final five" instructions are oubyi.

- Control organism (@ancestor):

Phenotype: Nan-; Oro-Other Traits:

- Org #1 (Nan+@1000):

Phenotype: Nan+

Cycle# when Nan completes: Final five instructions: Other Traits:

- Org #2 (Oro+@1000)

Phenotype: Oro+

Cycle# when Oro completes: Final five instructions: Other Traits:

• **Predict**: What do you expect to see if you extend the run and perform the same tests 1000 updates later?

- Will the *timing* of the trait occurrences remain the same? Yes / No

- Why?

- Will the final four instructions be the same? Yes / No

- Why?

- Will the other traits all remain the same? Yes / No

- Why?



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• **Test**: Repeat steps (ii) and (iv) from the Demo two times, extending the duration of your original run by 1000 updates each time.

- Org #3 (Nan+@2000):

Phenotype: Nan+ Cycle# when Nan completes: Final five instructions: Other Traits:

- Org #4 (Oro+@2000)

Phenotype: Oro+

Cycle# when Oro completes: Final five instructions: Other Traits:

- Org #5 (Nan+@3000):

Phenotype: Nan+

Cycle# when Nan completes: Final five instructions: Other Traits:

- Org #6 (Oro+@3000)

Phenotype: Oro+

Cycle# when Oro completes: Final five instructions: Other Traits:





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Results:

- Were your predictions confirmed or disconfirmed? If disconfirmed, how would you now revise your expectation?

- When comparing the organisms drawn from different times, was the same phenotype always produced by the same genotype? What does that tell you about the relationship between genotype and phenotype?

• **Discussion**: What do your tests above reveal about how phenotypic variations arise in a population?

• **Supplemental Inquiry**: If you repeat the experiment again from the beginning, what patterns in the data do you expect to be the same and what do you expect to be different between the first and second replicates? Why?

