Genetics Exercises
M1 Genotype-Phenotype
November 2021

1. The term « gene » can have several meanings (Table 1).

Table 1. Definitions of the Term "Gene"

A) Genes IX (Lewin, 2006, p. 845 and 852, Glossary) A gene is the segment of DNA specifying a polypeptide chain; it includes regions preceding and following the coding region (leader and trailer), as well as intervening sequences (introns) between individual coding segments (exons).
B) Quantitative Genetics (Falconer & Mackay, 1996, pp. 1–2) A gene is a unit of inheritance that is transmitted from parents to offspring. Suppose for simplicity that we were concerned with a certain autosomal locus, A, and that two different alleles at this locus, A1 and A2. [...] Each A1A1 individual contains two A1 genes.

Choose which definition(s) have been implicitly used in the following sentences:
a. Many of the **genes** not targeted by our library encode olfactory receptors that are unlikely to be cell-essential. (Blomen et al., Science 2015)
b. These Polycomb-repressed domains harbour **genes** encoding key developmental transcription factors, whose misexpression can have detrimental consequences in differentiated cells. (Boettiger et al., Nature 2016)
c. There has not yet been sufficient time for the corresponding resistance **genes** to spread into environmental reservoirs. (Versluis et al., Scientific Reports 2015)
d. Parkinson Disease is generally considered a multifactorial disorder that arises owing to a combination of **genes** and environmental factors.(Hou et al., Nature Reviews Neurology 2015)
e. ARID1B and ARID2 participate in widespread cooperation to repress hundreds of **genes**. (Raab et al., PLoS Genetics 2015)
f. Simulations reveal that hybrid populations rapidly and frequently become isolated from parental species by fixing combinations of **genes** that hinder successful reproduction with parental species. (Schumer et al., PLoS Genetics 2015)
g. Higher **gene** flow in sex-related chromosomes than in autosomes during fungal divergence. (Hartmann et al., Mol Biol Evol. 2019)

2. Compare the two visions of the genes below. What do you think?
"Now they swarm in huge colonies, safe inside gigantic lumbering robots, sealed off from the outside world, communicating with it by tortuous indirect routes, manipulating it by remote control. They are in you and me; they created us, body and mind; and their preservation is the ultimate rationale for our existence." (Dawkins, 1976)
"Now they are trapped in huge colonies, locked inside highly intelligent beings, moulded by the outside world, communicating with it by complex processes, through which, blindly, as if by magic, function emerges. They are in you and me; we are the system that allows their code to be read; and their preservation is totally dependent on the joy we experience in reproducing ourselves. We are the ultimate rationale for their existence." (Noble, 1999)

3. What are the following numbers?
a. Number of telomeres in a cell in G1 phase if its karyotype is 2n=16
b. Number of telomeres in a cell in G2 phase if its karyotype is 2n=16
c. Size of the human genome in base pairs
d. Number of recombination event per chromosome
e. % identity between human and chimpanzee DNA
f. Number of genes in the human mitochondrial genome
g. Number of different amino acids in the genetic code table
h. Total number of possible codons
i. Average number of de novo mutations in a person (germline-mutations) compared to his parents

4. Draw a cross between two parents, each homozygote for different alleles (a1,a2 and b1,b2) at two loci, *a* and *b*. Which generation may present the first recombined alleles between the two loci?
What will be the possible genotypes of the second generation (F2) after a cross among F1s?
Calculate the frequency of the different genotypes of F2 individuals:
4.1 if *a* and *b* are not linked, and
4.2 if they are located 1 cM apart on the same linkage group.

5. How many alleles of a given gene can be found in a haploid individual? in a diploid individual? in a population of diploid organisms after a chemical mutagenesis? in a natural population of diploid organisms?

6. Two snapdragons, one red and one white, are crossed. Their progeny is pink. What do you conclude? How many genes are involved in the color difference between the two parents?

7. One albino pigeon is caught in the Luxembourg garden and another in Central Park. The albino phenotype is caused by a recessive allele. What can you do to determine whether the same gene is responsible for the albino phenotype of both pigeons?
Unfortunately both are males. What can you propose instead?

8. A mutant *Drosophila* strain has no eyes. An eyeless female is crossed with a male of a wild-type line and F1 flies all have eyes. The F1 males are then backcrossed to the eyeless mother. The F2 generation displays a total of 87 flies with no eyes and 92 flies with normal eyes. What can you conclude about the genetic basis for the loss of eye? What can you conclude about the number of genes necessary for eye formation?

9. A line of *Drosophila* flies without eyes obtained after mutagenesis is crossed to a wild-type line showing a mean of 108 (+/- 5) ommatidia per eye. The F1 generation displays a mean of 35 (+/- 18) ommatidia per eye. What can you conclude?
What can you expect in the F2 generation if a single locus is involved?

10. A strain of flies with no hairs on part of the anterior legs is isolated from the Orsay orchard. It is crossed to a wild strain that was isolated on the Place Monge market, which shows a stable mean of 10.8 (+/- 0.5) hairs. The F1 generation displays a mean of 3.5 (+/-1.8) hairs. What can you conclude?
What can you expect in the F2 generation?

11. A butterfly species exists in two forms, "normal" (N) and crenelated (C). Five butterfly pairs are mated:

	Parent phenotypes		F1	
cross	Males	Females	Males	Females
#1	N	N	100% N	100% N
#2	C	C	100% C	100% C
#3	C	N	50% N, 50% C	50% N, 50% C
#4	C	N	100% C	100% C
#5	N	C	100% C	100% N

Write the genotypes of the parents of each cross and the mode of inheritance and of phenotypic expression of the alleles. (NB: The chromosomal basis for sex determination is not the same in all organisms.)

12. In a black, diploid, beetle species, loss-of-function mutant lines in four genes, *A*, *B*, *C*, *D,* as well as double and triple mutant combinations are available. They show the following phenotypes:

A: black	*A; D*: albino	*A; B; C*: yellow
B: brown	*C; D*: yellow	*A; B; D*: albino
C: yellow	*A; C*: yellow	*A; C; D*: albino
D: black	*B; C*: yellow	*B; C; D:* yellow
	A; B: brown	

Draw the genetic and biochemical pathways for pigment synthesis.
Which pigment accumulates in:
- animals of genotype *B; D?*
- in the F1 progeny of *A* and *D* animals?

13. You integrated a visible GFP transgene at random in a wild-type Drosophila genome and you would like to know on which chromosome it is, using recessive morphological markers for the four chromosomes (three autosomes and the X chromosome). Describe the crosses you do to map the transgene. Recombination occurs in Drosophila females but not in males.