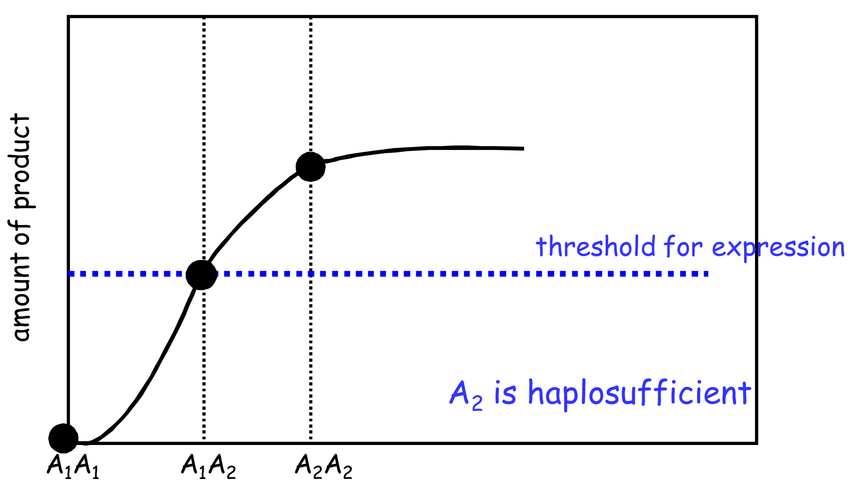
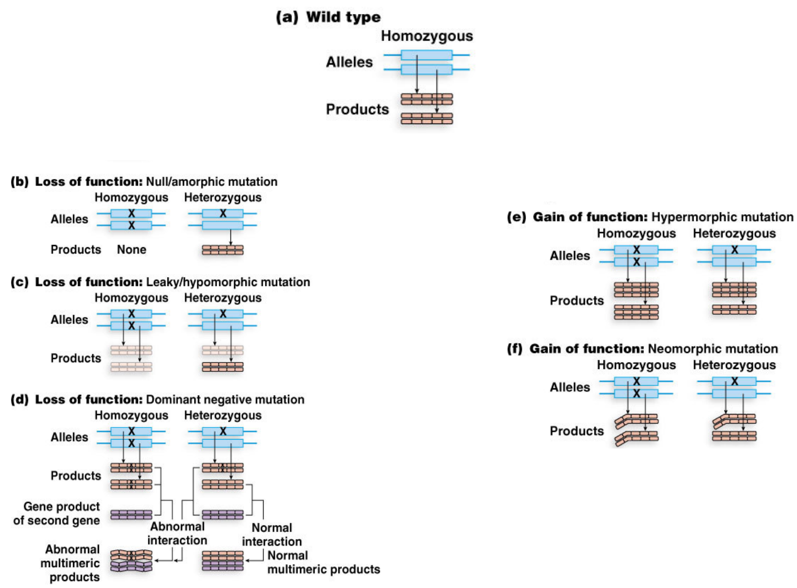
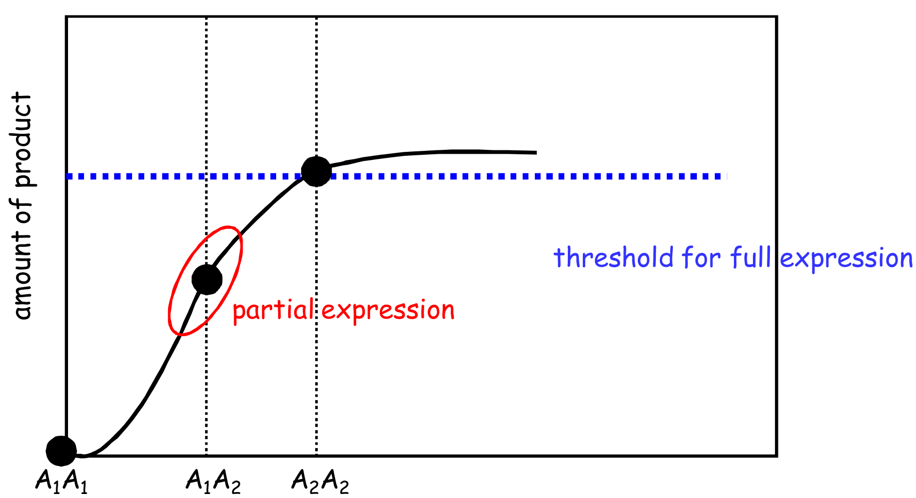
Genotype ⟶ Phenotype

- interactions between alleles
  - dominance
  - multiple phenotypic effects of one gene
  - phenotypic variability in single genes
- interactions between genes







phenotype of A_1A_2 will be intermediate

early-flowering -- "day 0"
late-flowering --- 5 days later

P_0 early x late

A_1A_1 A_2A_2

F_1 intermediate (3.5 days later)

A_1A_2

F_2 $\frac{1}{4}$ early $\frac{1}{2}$ intermediate $\frac{1}{4}$ late

A_1A_1 A_1A_2 A_2A_2

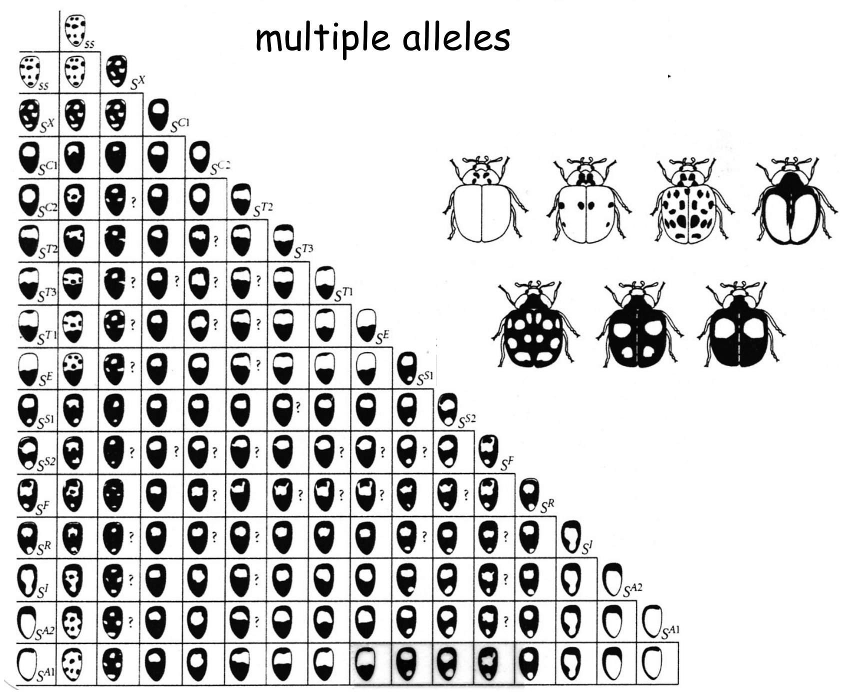
0 3.5 5

A_1A_1 early

A_1A_2 intermediate

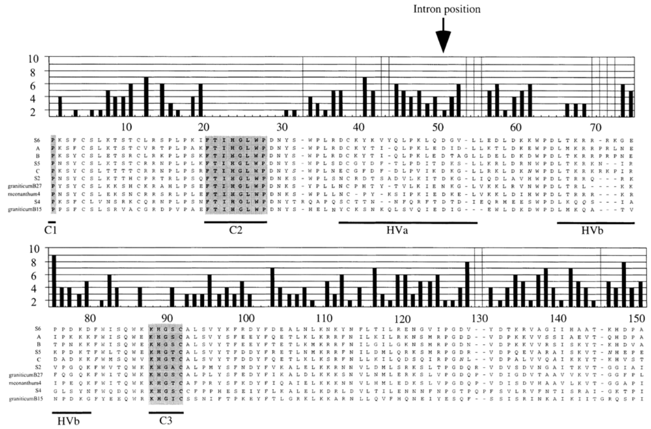
A_2A_2 late

multiple alleles



Self-incompatibility loci (*S* locus)

	cross		outcome
	pollen	ovule	(% fertility)
	s^1s^2	s^3s^4	100%
	s^1s^3	s^3s^4	50%
	s^3s^4	s^3s^4	0%



multiple alleles; mixed dominance -- ABO blood groups

phenotype	genotype
O	ii
A	I^AI^A or I^Ai
B	I^BI^B or I^Bi
AB	I^AI^B

$$I^A = I^B > i$$

type A Rh^+ woman × type B Rh^+ man

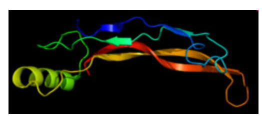
I^A/i; Rh/rh I^B/i; Rh/rh

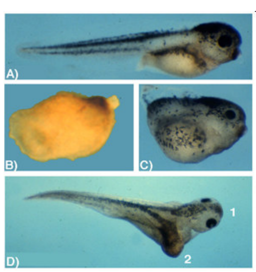
↓

A Rh^+ son ; B Rh^+ daughter; O Rh^- daughter

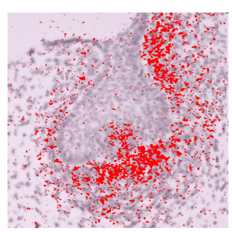
I^A/i; $Rh/-$ I^B/i; Rh/rh i/i; rh/rh

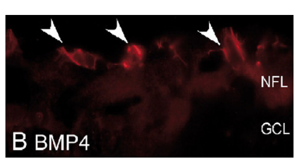
Pleiotropy:
the many faces of BMP-4



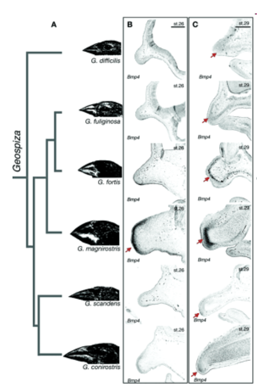


dorsal-ventral patterning in *Xenopus*

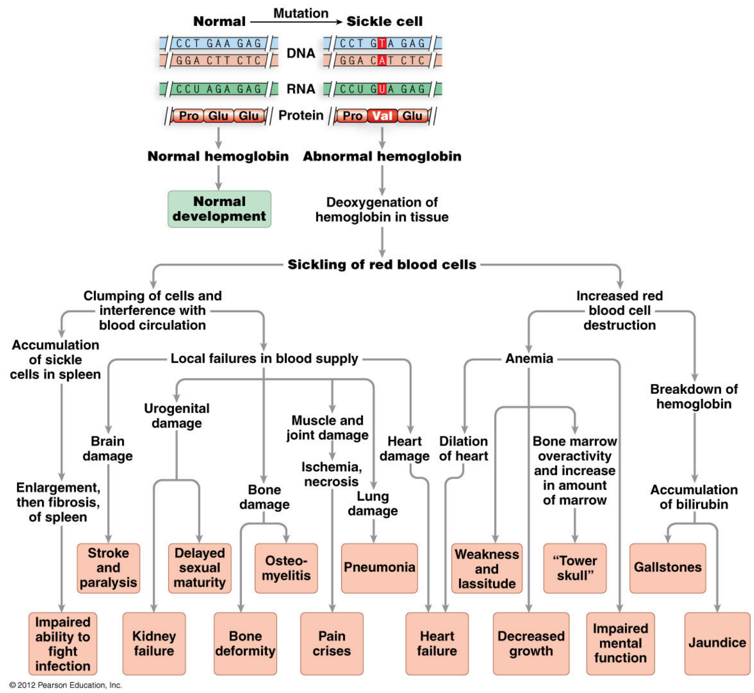

tooth development



retina development



bill shape differences



recessive lethal alleles, a special kind of pleiotropy

curly (Cy) ⟶ two effects on the phenotype
- morphology (dominant)
- viability (recessive)

Cy/Cy	Cy/+
dead	curly wings

Curly ♂ x Curly ♀

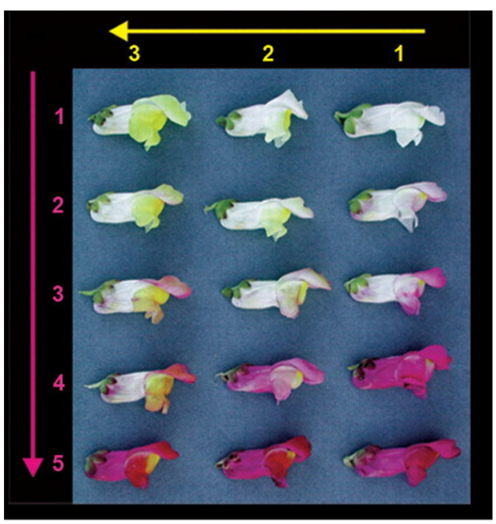
2/3 Curly 1/3 wild type

detection of complex dominance, multiple alleles, and recessive lethals:

departures from simple mendelian inheritance

- F_1 are different from parental phenotypes (incomplete dominance; >1 gene)
- F_2 ratio is similar to 3:1, but…
  - has >2 phenotypes (incomplete dominance, multiple alleles)
  - has something "missing" (recessive lethals)

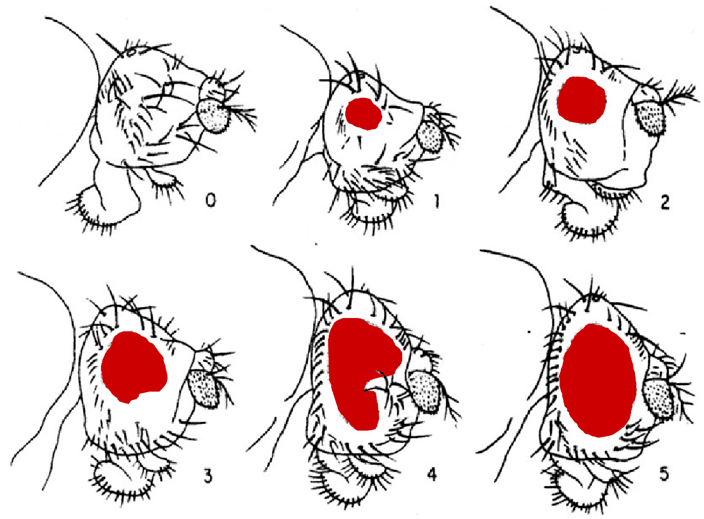
phenotypic variability in single genes



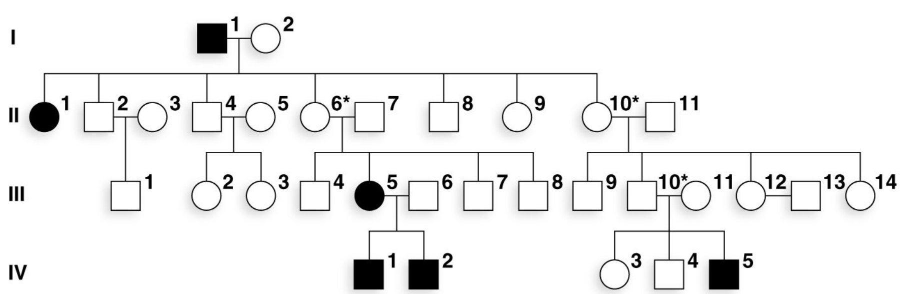
incomplete penetrance

variable expressivity

variable expressivity of Lobe eye in *Drosophila*



polydactyly—
autosomal dominant,
incomplete penetrance

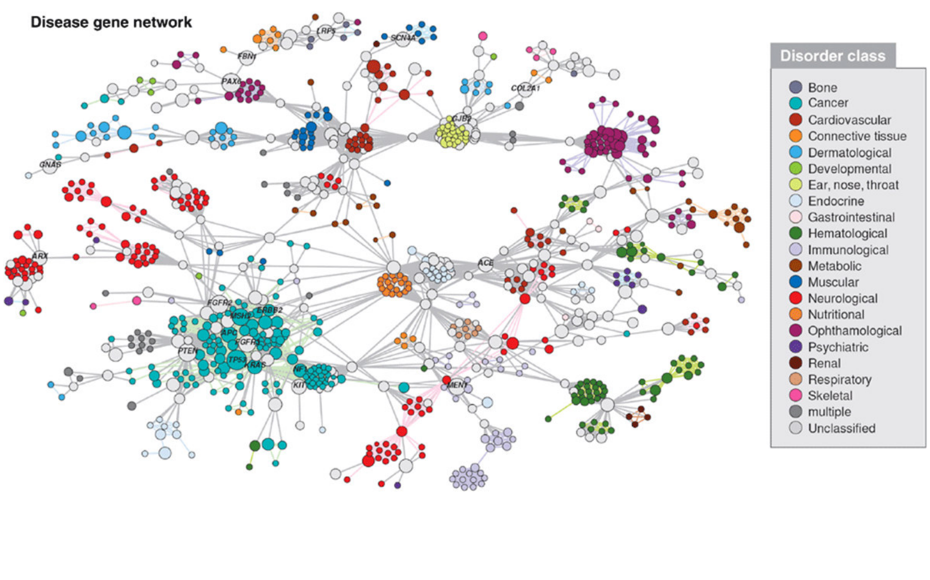


* Nonpenetrant individual

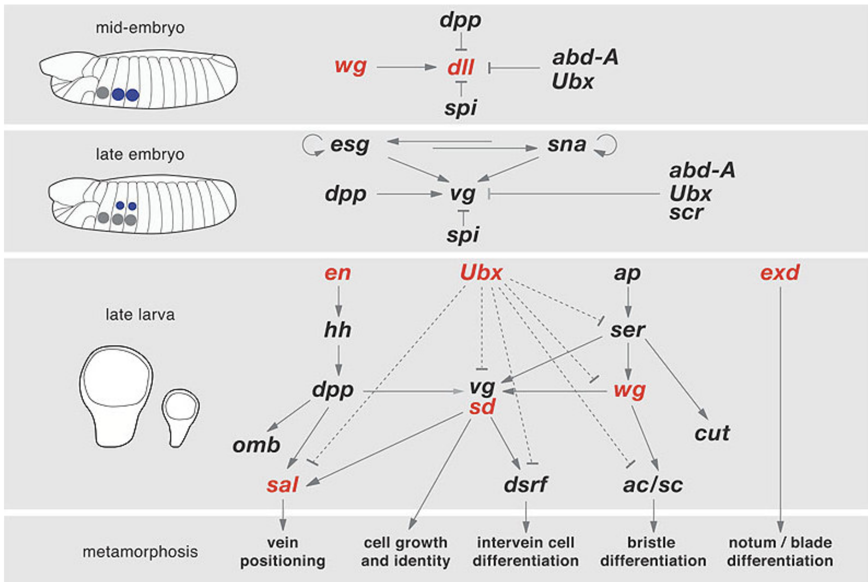
causes of variable expressivity and incomplete penetrance

- effects of other genes
- environmental factors
- genetic and environmental factors

interactions between loci



Goh et al 2007 The human disease network. PNAS the wing-patterning network in *Drosophila melanogaster*

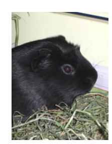


mammalian coat color pathway

pigment cell

{ pigment cell development }

spotting (S)
silvering (Si)
diminuation (Dm)
grizzling (Gr)



yellow (e)

tortoiseshell (eP)

black (E)

sootiness (so)

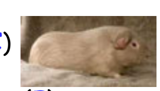
agouti (A)

{ yellow

pigment differentiation

black }

color genes (C)
dilution (P)
color modifiers (F)



B, b

pigment

{ yellow

brown

black }

physical environment

genotype → phenotype

other genes

interacting genes and proteins

interaction ⟶ "wild type"

- complementation

interaction ⟶ "alternate phenotype"

- novel phenotypes
- suppression

eye color variation in Drosophila

P_0 brown × vermillion

↓

F_1 wild type

and,

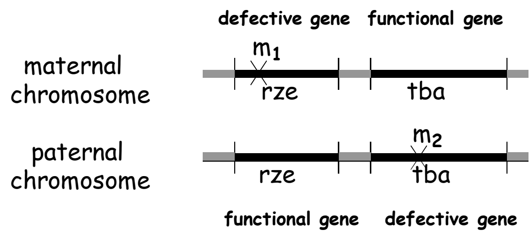
P_0 apricot ♀ × cherry ♂

↓

F_1 white ♀♀, apricot ♂♂

deciding one gene vs. two

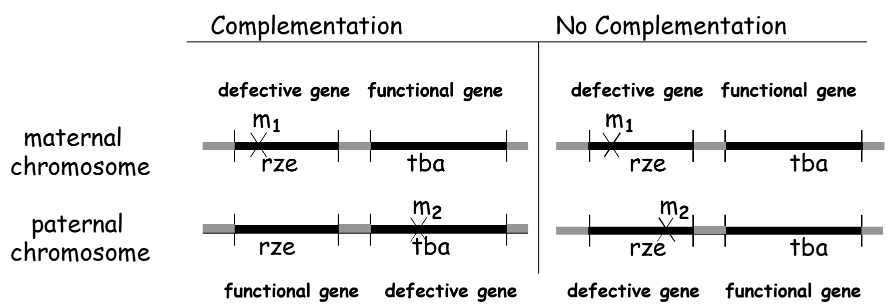
Complementation



deciding one gene vs. two

No Complementation

deciding one gene vs. two



two genes

one gene

interacting genes influence eye color in Drosophila

colorless precursor → ommochrome → pigment deposition

colorless precursor → drosopterin → pigment deposition

pigment deposition → eye color

P_0 brown × vermillion

b/b; +/+ +/+; v/v

F_1 wild type b/+; v/+

F_2 9 wild type: 3 brown: 3 vermillion: 1 white

+/-; +/- b/b; +/+ +/+; v/v b/b; v/v

interacting genes influence eye color in Drosophila

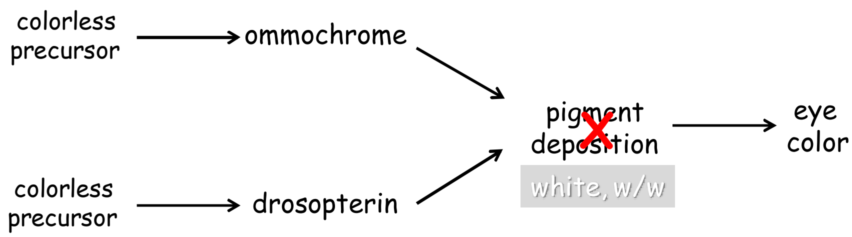
vermillion, *v/v*

colorless precursor → ommochrome → pigment deposition → eye color

colorless precursor → drosopterin → pigment deposition

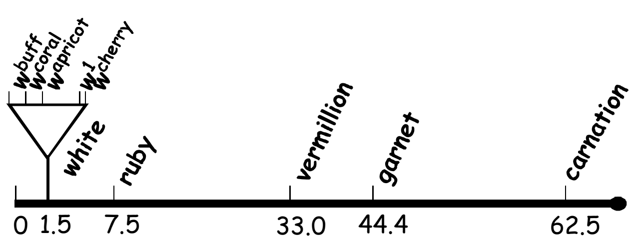
brown, *b/b*

interacting genes influence eye color in Drosophila

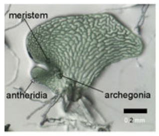


complex allelic interactions of *Drosophila* eye color

mutation	white	garnet	ruby	vermillion	cherry	coral	apricot	buff	carnation
white	-	+	+	+	-	-	-	-	+
garnet		-	+	+	+	+	+	+	+
ruby			-	+	+	+	+	+	+
vermillion				-	+	+	+	+	+
cherry					-	-	-	-	+
coral						-	-	-	+
apricot							-	-	+
buff								-	+
carnation									-



gene interaction and sex-determination in a fern



Hermaphrodite has:
- multicellular meristem
- archegonia
- few antheridia

Male has:
- no meristem
- no archegonia
- many antheridia

pheromone, antheridiogen (ACE)
-made by hermaphrodites
-young gametophytes become male

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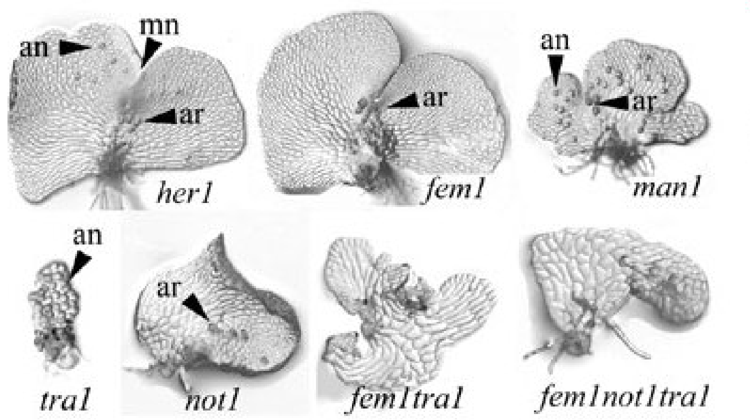
several mutations that affect sex-determination



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Compare the phenotypes of single, and different combinations of double or triple mutants

Summary of single-, double-, and triple-mutant phenotypes

Genotype	Phenotype + A_{CE}	Phenotype − A_{CE}	Additional references
Wild type	Male	Hermaphrodite	
feminization1 (*fem1*)	Female	Female	Banks (1994)
notchless1 (*not1*)[a]	Mostly female	Mostly female	
transformer (*tra*)	Male	Male	Banks (1994, 1997b)
hermaphroditic (*her*)	Hermaphrodite	Hermaphrodite	Warne *et al.* (1988); Eberle and Banks (1996)
many antheridia1 (*man1*)	Male	Many antheridia	Banks (1997b)
fem1 tra	Intersex	Intersex	Eberle and Banks (1996)
not1 tra	Male	Male	
fem1 man1[b]	Intersex-female	Female	Banks (1997b)
not1 man1	Male	Many antheridia	
her man1[c]	Many antheridia	Many antheridia	Banks (1997b)
her tra	Male	Male	Eberle and Banks (1996)
tra man1	Male	Male	Banks (1997b)
fem1 her	Female	Female	Eberle and Banks (1996)
not1 her	Female	Female	
fem1 not1	Female	Female	
fem1 man1 tra	Intersex	Intersex	Banks (1997b)
fem1 not1 tra	Asexual	Asexual	

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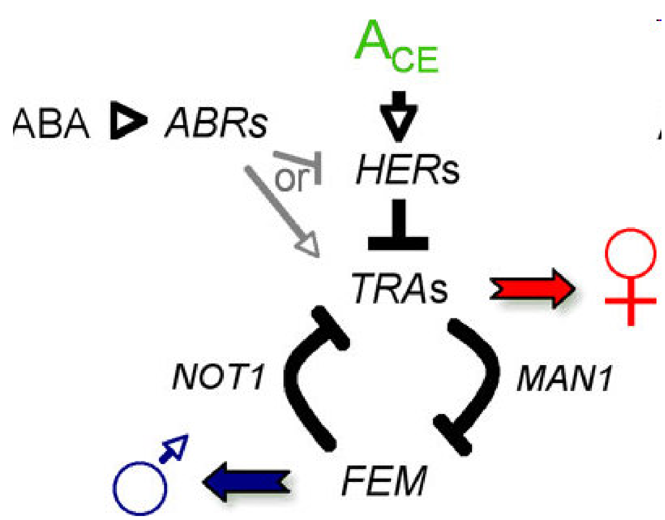
Compare the phenotypes of single, and different combinations of double or triple mutants



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epistatic modification of ABO phenotypes

"A" phenotype x "O" phenotype

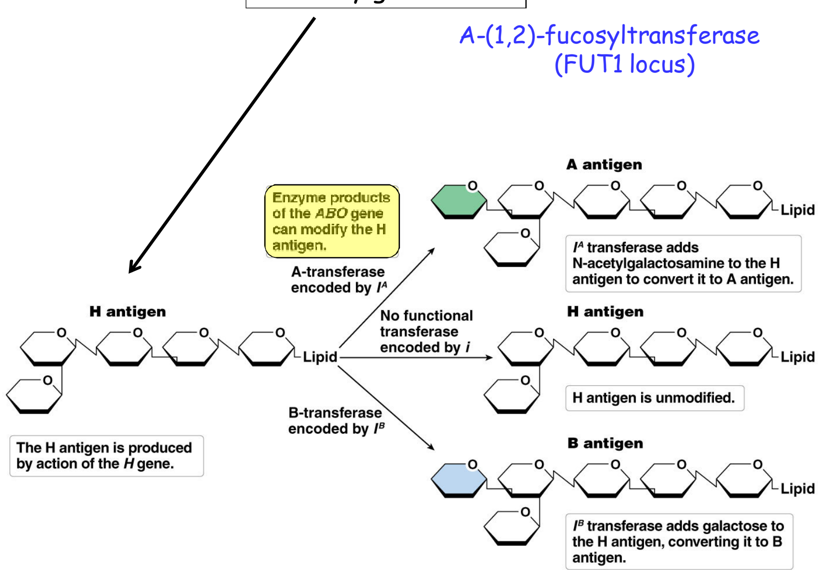
expect: all "A" phenotype offspring, if "A" parent is I^AI^A

or: mix of "A" and "O" offspring, if "A" parent is I^Ai

instead: an "AB" phenotype offspring

N-acetylglucosamine

A-(1,2)-fucosyltransferase
(FUT1 locus)



epistatic modification of ABO phenotypes

"A" phenotype x "O" phenotype

I^AI^A Hh I^BI^B hh

↓

"AB" phenotype

I^AI^B Hh

incomplete dominance occurs when the product of one allele is insufficient to produce the expected phenotype

with incomplete dominance, heterozygotes have a phenotype intermediate to both homozygotes

typically, more than two alleles segregate at a locus; multiple alleles can be detected when crosses between heterozygoted yield non-standard F_2 ratios

pleiotropic genes affect more than one aspect of the phenotype; it can be detected by non-standard ratios

complementation tests can determine if two mutations are in the same gene or different genes

two locus interactions can be detected by the presence of novel phenotypes in the F_1 or F_2 or an F_2 ratio that resembles one for two traits rather than one trait