Mechanisms responsible for Mendel's Postulates are generally conserved. However, expected patterns are sometimes masked, and exceptions for all postulates are common.

Postulate	Reason	Exception
1) Unit factors in pairs	**Diploids**	**Bacteria or polyploids**
2) Dominance	**Null alleles**	**Partial Dominance**
3) Segregation	**Anaphase I**	**Non-disjunction**
4) Genes are Independent	**Alignment of of Chromos at Metaphase I**	**Interactions: Epistasis Linkage**

Biochemical bases of phenotypic expression

Variation we see in a trait depends upon:

1) Interaction of alleles (dominance relationships)

2) Environment

3) Number of alleles in a population at a locus

4) Number of genes that affect a trait

5) Interaction of genes (epistasis)

These factors can mask the underlying mode of inheritance: Mendelian rules can be in effect, but segregation pattern is altered

1) Dominant **verses** recessive

Do we always see dominance of one allele over another?

Is the Dominant allele more frequent than the recessive?

Is the Dominant phenotype bigger than the recessive phenotype?

Is the Dominant allele more fit than the recessive?

Let's start looking at the underlying mechanisms that control allelic interactions and the exceptions to Mendel's rule that they create!

Looks like "blending inheritance" at first.

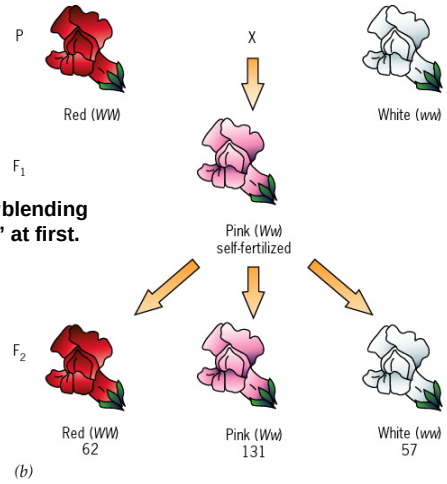
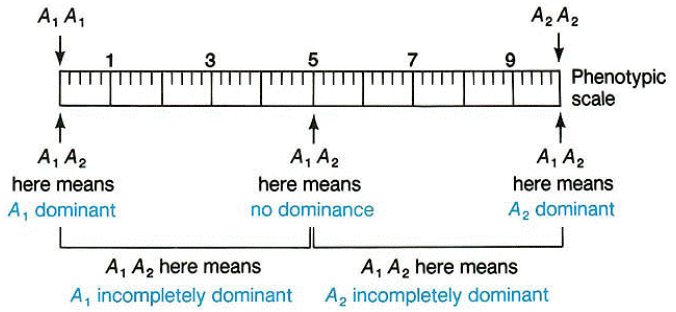


Fig. 3.10 Results of crosses between red and white varieties of snapdragons.

Fig 4.1 Genetic basis of flower color in snapdragons.

Dominance as a continuum.



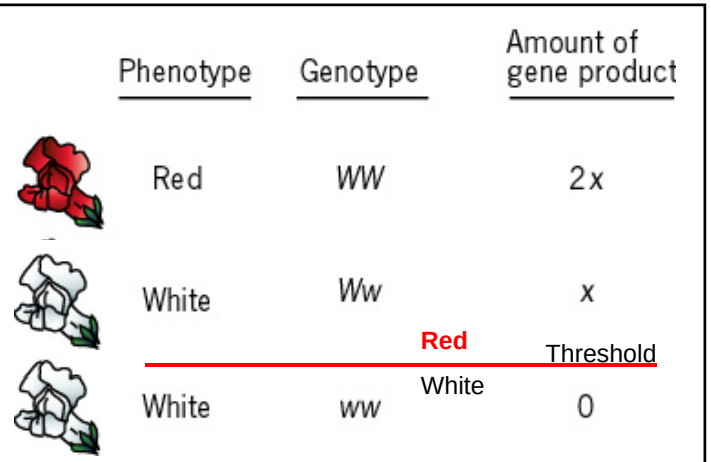


Fig 4.1 Genetic basis of flower color in snapdragons.

"L" named after Karl Landsteiner on Chr 4

Genotype	Antigen Glycoprotein	Antibody made in blood
$L^M L^M$	M M / M M	Anti-N
$L^M L^N$	N M / M N	None
$L^N L^N$	N N / N N	Anti-M

Could get transfusion from NN or MM

The difference in these proteins is small but significant

M has Ser-1, Gly-5

N has Leu-1, Glu-5

Heterozygote expresses both alleles and is distinguishable from homozygotes, therefore Co-dominant

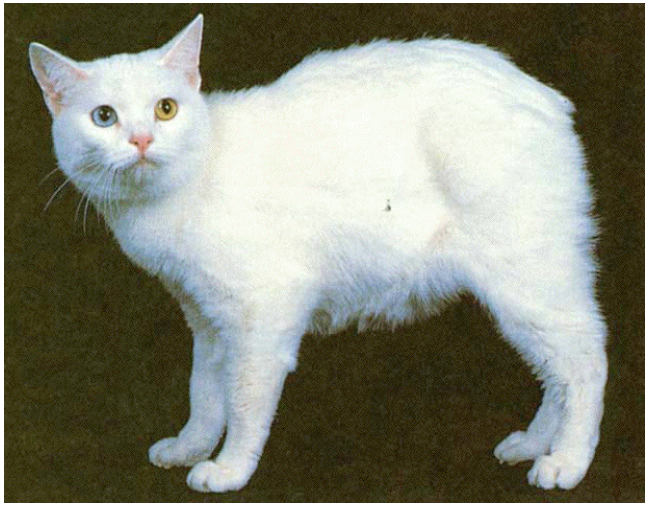
Table 26-1 Frequencies of Genotypes for Alleles at MN Blood Group Locus in Various Human Populations

Population	Genotype			Allele frequencies	
	MM	*MN*	*NN*	*p*(*M*)	*q*(*N*)
Eskimo	0.835	0.156	0.009	0.913	0.087
Australian	0.024	0.304	0.672	0.176	0.824
Egyptian	0.278	0.489	0.233	0.523	0.477
German	0.297	0.507	0.196	0.550	0.450
Chinese	0.332	0.486	0.182	0.575	0.425
Nigerian	0.301	0.495	0.204	0.548	0.452

SOURCE: W. C. Boyd, *Genetics and the Races of Man*. D. C. Heath, 1950.

(Griffiths et al. 1996)

More Allelic Interactions-- Manx



Manx Cats (kind of cute, let's breed them!!!)

Cross: Manx X Manx

1 Normal: 2 Manx

What is the genetics???

It segregates (produces different types of progeny).

Could it be test cross?

No, parents would be different.

Could it be Het x Het?

We expect 1:2:1 or 3:1 ratio!! Lets work with this.

M^LM Manx $M\ M^L$ X $M\ M^L$

¼ MM: ½ M^LM: ¼ ~~M^LM^L~~

M^LM^L Dies, M^L interferes with normal spinal development

Sickle Cell Anemia

Case of Recessive lethal in humans; also effect of environment on dominance:

- $\beta^A\ \beta^A$ Normal Never sickle
- $\beta^A\ \beta^s$ No anemia. Red cells sickle only under low O_2
- $\beta^s\ \beta^s$ Severe often fatal anemia. Cells always sickle

Found in high frequency in tropical (Malaria regions; Africa and Asia)

Tay-Sachs Disease

Mutation in hexosaminase A (HEXA) polypeptide on **Chr. 15**, that helps break down fatty acids in the nerve cells and generally lethal in childhood. Common in Ashkenazi Jewish, French Canadian and Cajun populations

Genotype	HexosaminaseA Production	Phenotype (Symptoms)
HexA/HexA	100%	Normal
HexA/HexA^T	50%	Normal
HexA^T/HexA^T	0%	Lethal

Is this disease "recessive" or "no dominance"?

Examples that don't show dominance (so we do not see 3:1 ratios in F2):

No dominance (partial or incomplete dominance)

Flower color in snapdragons

Co-dominance

MN blood group

Trait measurement changes our perspective of dominance

Manx

Sickle cell anemia

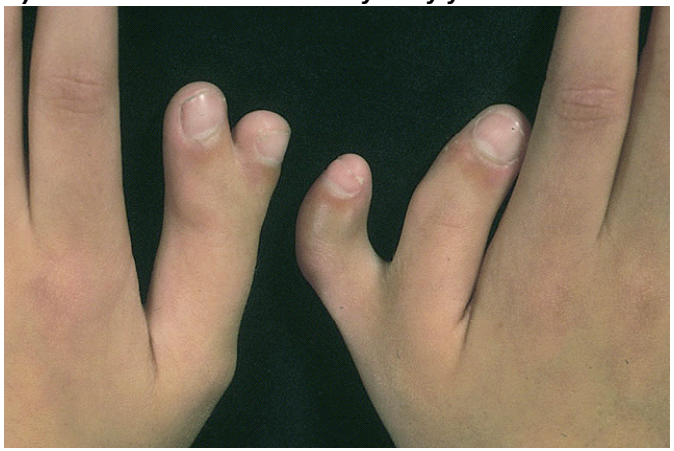
Tay-Sachs disease: deficiency in the production of hexosaminase A– Enzyme production of heterozygote is ½ that of normal homozygote

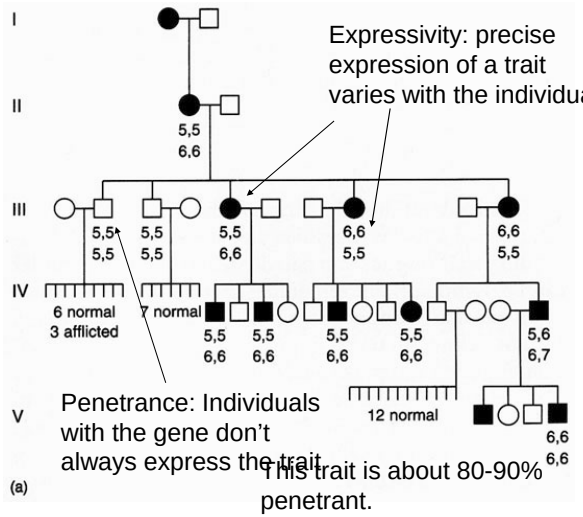
2) What part of the variation that we see is caused by genetics and what part is caused by environment?

Dean	and	Dan	Identical twins
6'3"		6'3"	
240lbs		190	
light hair thin		dark hair	
diabetes		no diabetes	
DNA sequencer		DNA sequencer	
PhD		MD	

Adult on-set diabetes 50% penetrance. This leads to other trait differences

2) Environmental Effects: Polydactyly



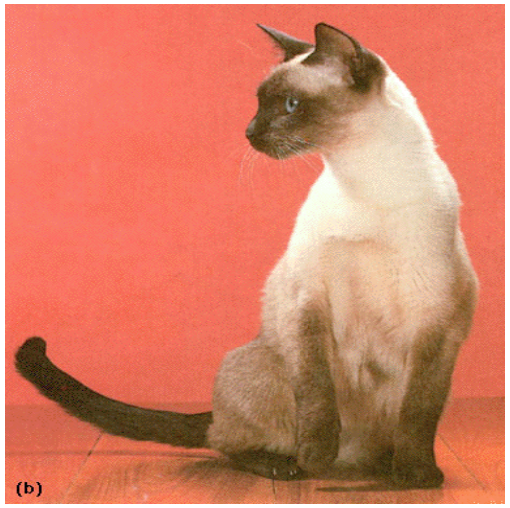


Expressivity: precise expression of a trait varies with the individual

Penetrance: Individuals with the gene don't always express the trait.

This trait is about 80-90% penetrant.

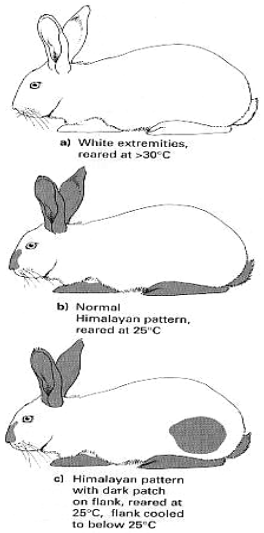
Siamese Cats



Himalayan Rabits

What's the cause of the cute color patterns?

Temperature sensitive enzyme that produces pigments.



Do you think polka-dotted bunny rabbits will be the next big pet?

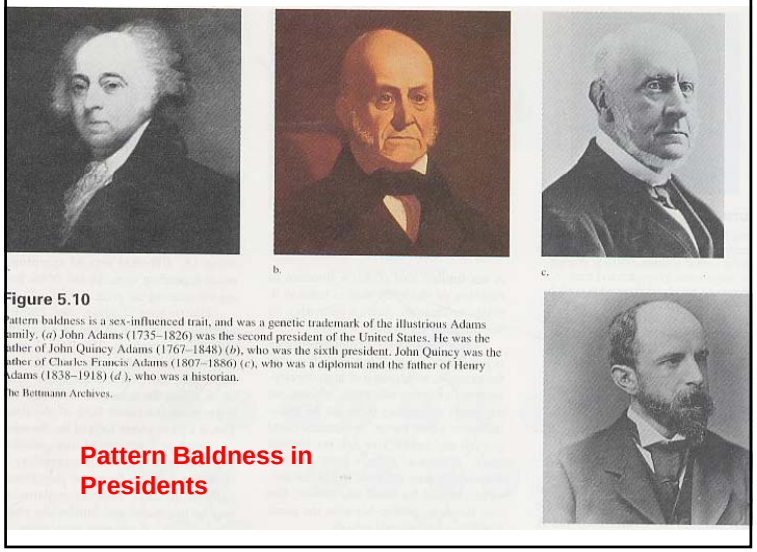


Figure 5.10

Pattern baldness is a sex-influenced trait, and was a genetic trademark of the illustrious Adams family. (*a*) John Adams (1735–1826) was the second president of the United States. He was the father of John Quincy Adams (1767–1848) (*b*), who was the sixth president. John Quincy was the father of Charles Francis Adams (1807–1886) (*c*), who was a diplomat and the father of Henry Adams (1838–1918) (*d*), who was a historian.

The Bettmann Archives.

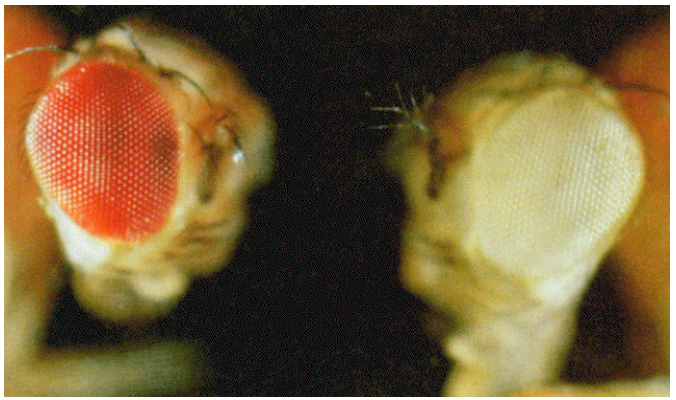
Pattern Baldness in Presidents

Genetics of Sex Influenced Genes

Genotype	Male Phenotype	Female Phenotype
Pb Pb	Bald	Bald
Pb pb	Bald	Not Bald
pb pb	Not Bald	Not Bald

Pb is dominant in males but recessive in females!!!

3) Multiple Alleles



Some alleles present at the *white* eye locus of *Drosophila melanogaster*

Allele	Name	Eye Color Phenotype
w^+	white-wildtype	brick red
w	white	pure white
w^a	white-apricot	yellow-orange
w^{bf}	white-buff	light-buff
w^{bl}	white-blood	yellow-ruby
w^{cf}	white-coffee	deep ruby
w^e	white-eosin	yellow-pink
w^{mo}	white-mottled	light orange mottled
w^{sat}	white-satsuma	deep ruby
w^t	white tinged	light pink

4) Multiple Genes combine to give color and patterns

Orange and Black Pigments

O_B_

Black only

oo B_

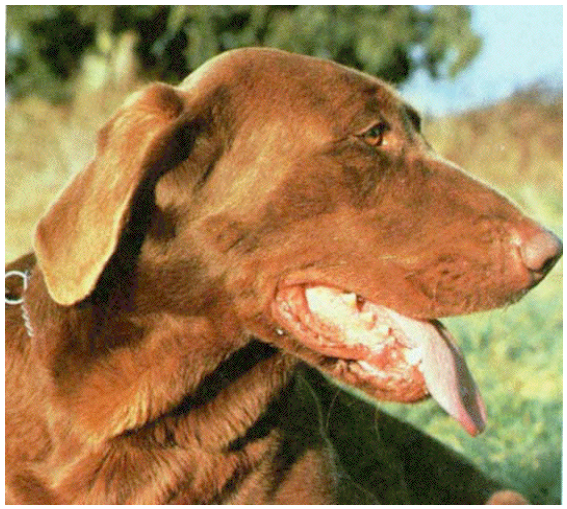
Orange only

O_ bb

Neither orange or black

oo bb

5) Multiple genes can interact. Gene interaction is called epistasis.



B_ Black

bb chocolate

E_ Deposition of color

ee No color

Bb Ee x Bb Ee

Black Black

This is an F1 x F1 cross. What would the phenotypic ratio in the F2 population be?

A) 3:1

B) 1:1:1:1

C) 9:3:3:1

D) 9:4:3

E) 12:3:1