

# Genetics

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## *Dominance and Relationships*

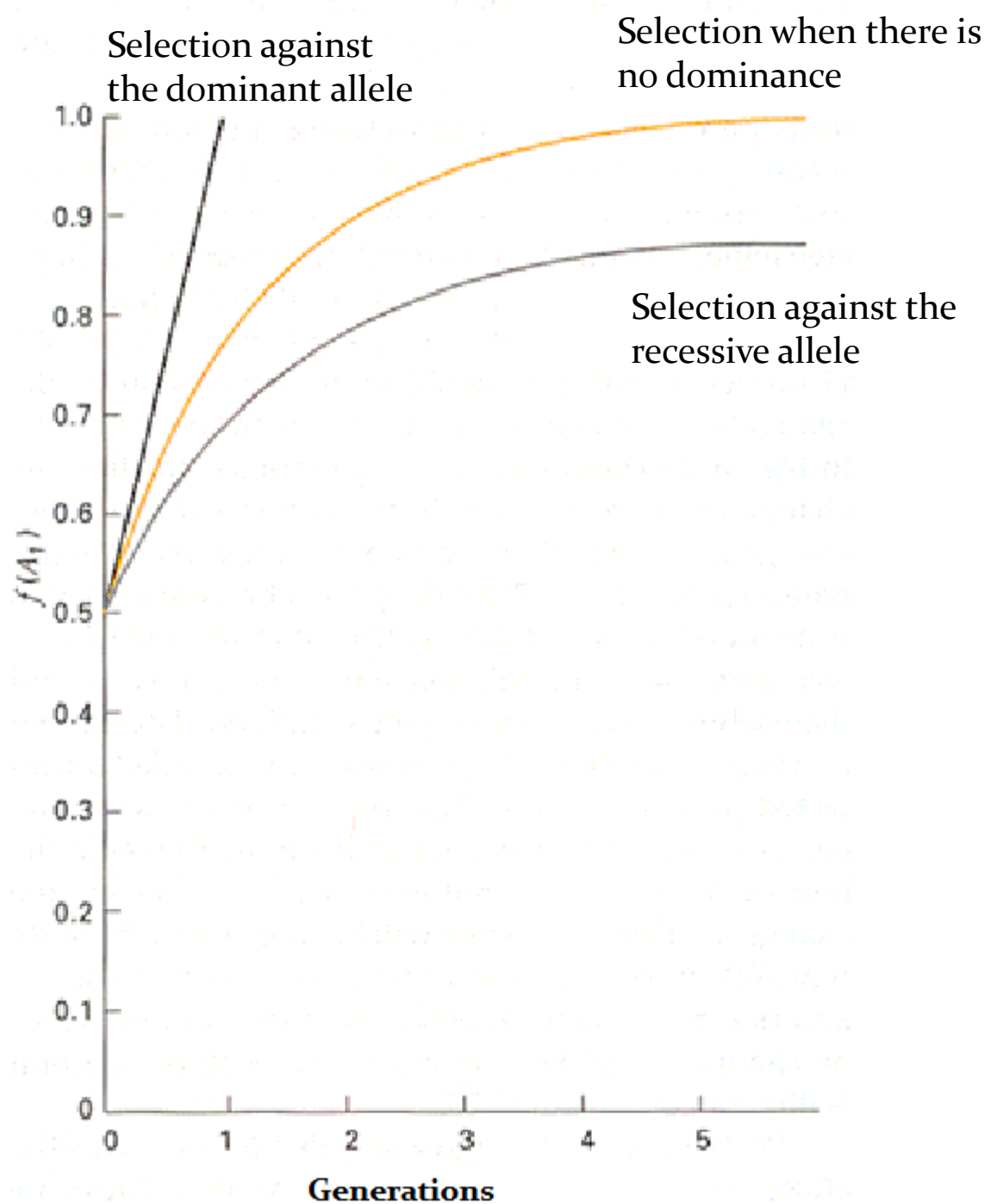
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# Selection Efficiency

- ❑ Dominance does not always mean higher fitness (survival, reproduction).
- ❑ Most of the dominant diseases have been eliminated.
- ❑ When the recessive allele is less fit, or when there is artificial selection against it, the tail of the curve can be quite long. Why?
- ❑ What kind of artificial selection people deal with?



# Mating Methods

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- Random mating
- Positive assortative mating
  - Based on genotypic similarities
  - Based on phenotypic similarities
- Negative assortative mating

# Random mating

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## □ Usually happens in plants

- Bees-insects or wind carry pollen, may cross pollinate to spread genes further afield
- GM (genetically modified) pollen drift and insect pollination may create for nearby non-GM or organic crops.
- No need to label products contaminated with less than 0.9% with GMOs in EU. Contamination could be from seed impurity, machine contamination when sowing or harvesting, cross fertilization and mixing during transport, storage and processing.

# Random mating

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- Classic farms have random mating
  - Bucks-rams are held within the herd, and mate with the ewes in heat (estrus)
- Random mating is the method in which the genetic variation is conserved the most.
  - Genetic diversification is generally seen as an adaptive strategy in unpredictable environments.

# Positive assortative mating

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- Mating of similar individuals
- Based on genotypic similarities
  - Inbreeding
- Based on phenotypic similarities
  - Part of the herd may be mated just to keep the phenotypic similarities.
  - Animals that show a breed's features well may be bred to take to shows and beauty pageants.

# Nonrandom mating

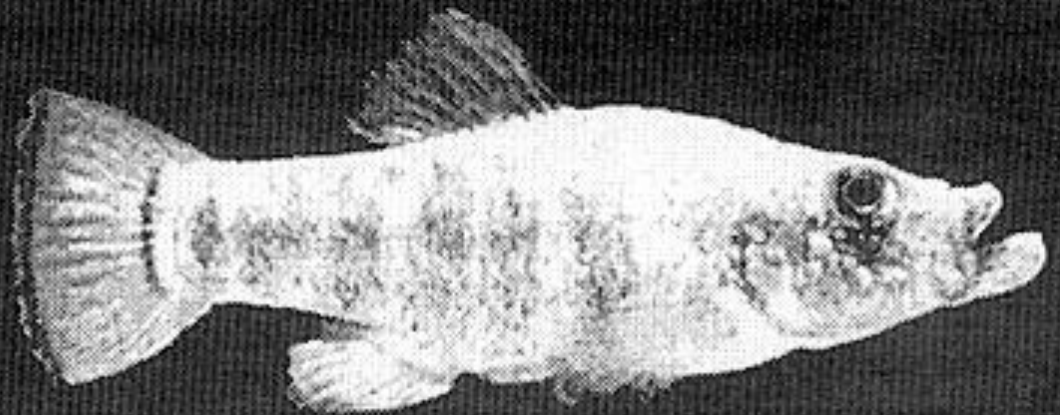
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- ❑ Positive assortative mating= Similar individuals mate more frequently than would be expected under a random mating pattern.
- ❑ Example: It is common for individuals of similar body size to mate with one another.
- ❑ This type of mating decreases the proportion of heterozygotes.

## A. Bahamian normal-form (Bn)



## B. Bahamian bulldog-form (BB)



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Tropic  
polymorphism  
causes strong  
assortative mating.  
Bn mate with their  
own, BB mate with  
their own

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# Nonrandom mating

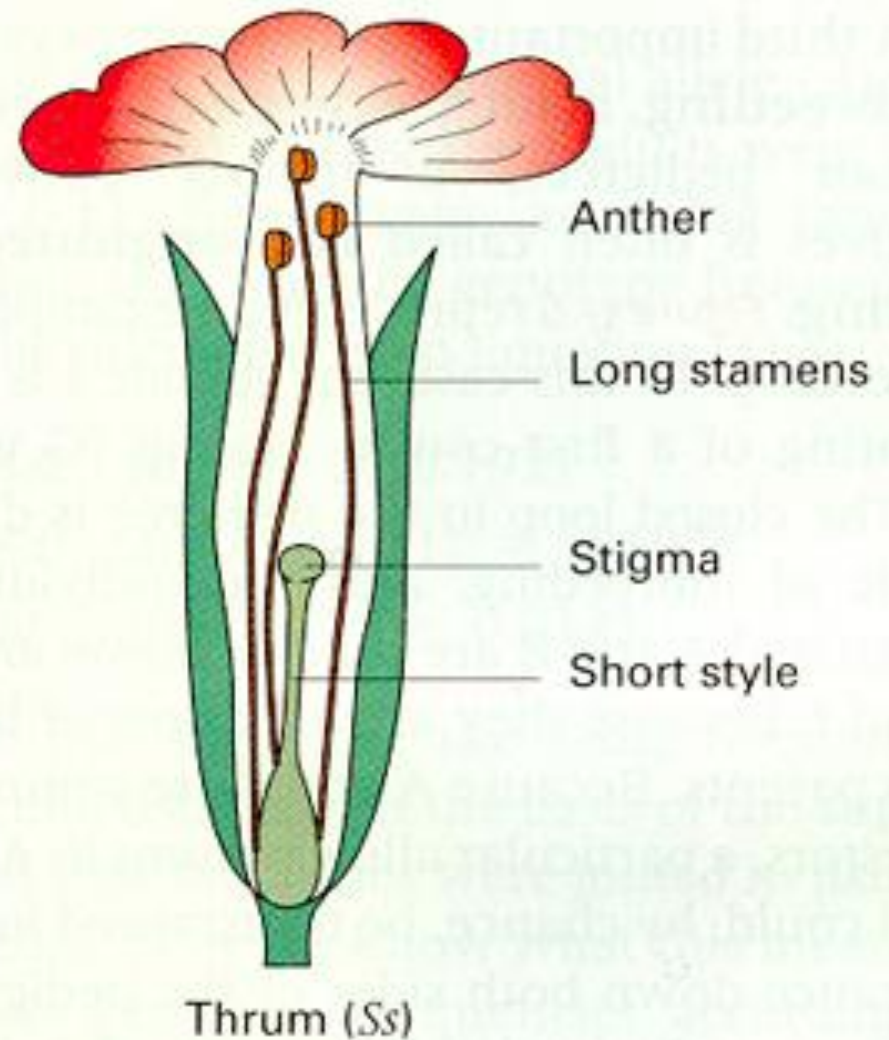
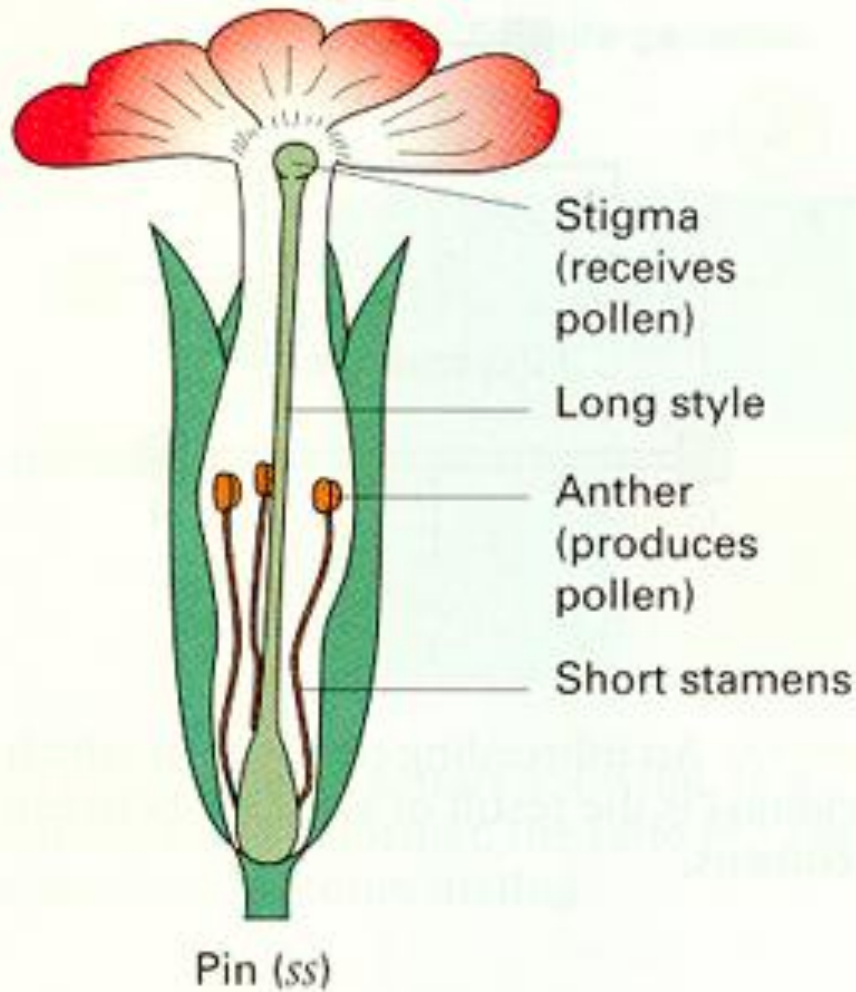
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- **Negative assortative mating**  
(disassortative mating) = individuals with diverse traits mate more frequently than would be expected under random mating
- Example: An experiment had subjects choose their preferred image with
  - an image modified to resemble the test subject
  - a non-resembling attractive face
  - a face more attractive than the resembling face, as determined by an outside group.

# Nonrandom mating

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- The study found that
  - male subjects preferred the female faces that resembled their own (assortative mating),
  - females did not prefer the male faces that looked like their own (disassortative mating).
- Negative assortative mating, or disassortative mating results in a greater number of heterozygotes.



*Primula officinalis* flower Pin and Thrum forms.  
 Anatomy of these flowers causes negative assortative  
 mating: one sends pollen the other receives it.

# Negative assortative mating: Based on genetic dissimilarities

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- ❑ Genetically different individuals mate, those that are not relatives, or those that are different breeds.
- ❑ Purpose of these matings are to produce a phenotypically superior crossbred generation.
- ❑ Breeding values (additive) of these animals are not considered, instead gene combination values are used.

# Negative assortative mating

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- Recessive and lethal genes hide under heterozygosity
- They combine and come out as homozygosis increases.
  - Genetic variance may be low especially in low heritability traits.
  - Small genetic variance means genetically similar individuals.
  - Effects of inbreeding may be catastrophic for these traits. Why?

# Negative assortative mating

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- Genotypic variance of  $F_1$  generation is less than the parental generation.
- Crossbred individuals are more resistant to environment compared to inbred individuals because they have various genes.
- Heterosis is the tendency for hybrid individuals to exceed their purebred parents in vigor, growth rate, reproduction, survivability etc.

# Dominance, additive

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- If there is additive gene action:
  - $A=1, AA=1+1=2, Aa=1+0.5=1.5$
  - $a=0.5, aa=0.5+0.5=1$
- If complete dominance:
  - $AA=2$
  - $Aa=2$
  - $aa=1$

# Dominance, additive

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- If incomplete dominance:
  - $AA=2$
  - $Aa=1.7$ , or  $1.8$  etc.
  - $aa=1$
  
- If there is overdominance:
  - $AA=2$
  - $Aa=2.1$  or  $2.5$  etc.
  - $aa=1$



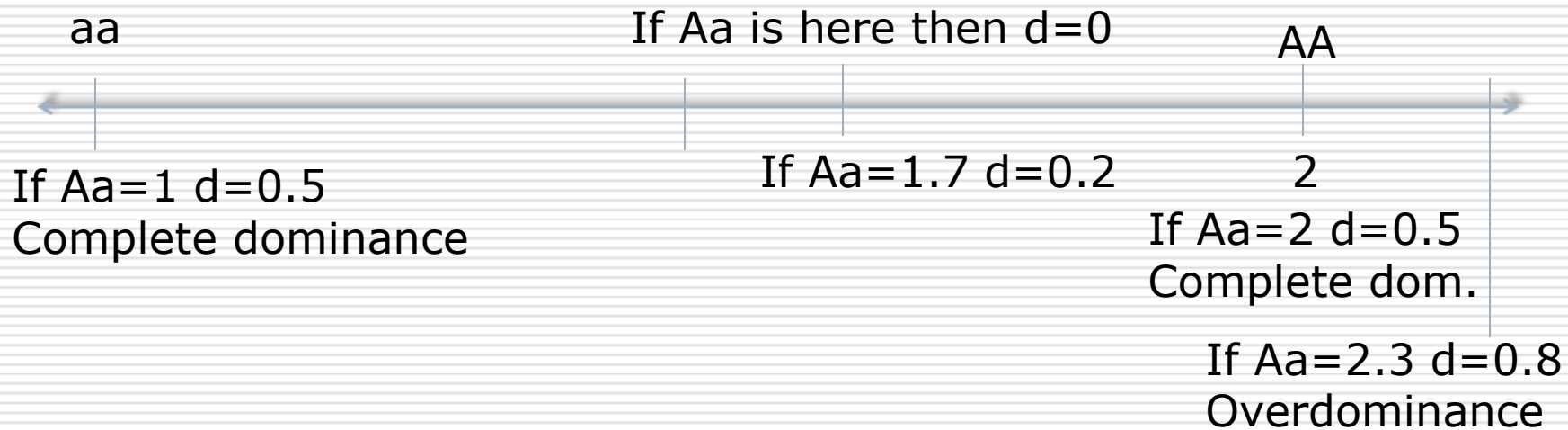
# Dominance, additive

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- If complete dominance for a:
  - $AA=2$
  - $Aa=1$
  - $aa=1$

# Dominance, additive

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# Negative assortative mating, Two theories

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- Two theories for the benefit of heterozygosity exist
  - Complete dominance
  - Overdominance

# Two theories, Dominance versus Overdominance

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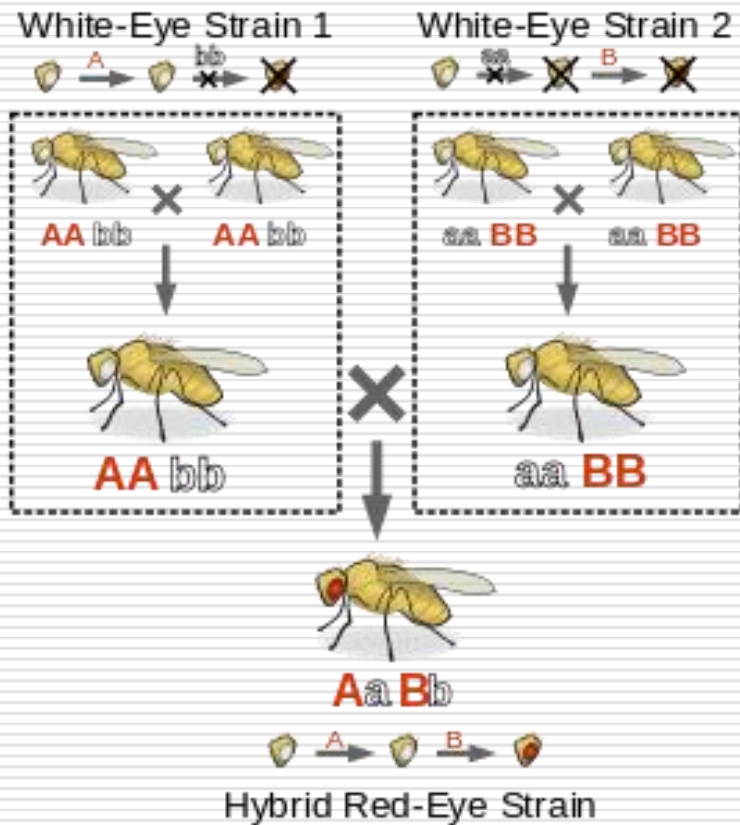
- Two competing genetic hypotheses, not necessarily mutually exclusive, have been developed to explain hybrid vigor.
- More recently, an epigenetic component of hybrid vigor has also been established.
  - Ni Z, Kim ED, Ha M et al. (January 2009). "Altered circadian rhythms regulate growth vigour in hybrids and allopolyploids". *Nature* 457 (7227): 327–31.
  - Baranwal VK, Mikkilineni V, Zehr UB, Tyagi AK, Kapoor S (November 2012). "Heterosis: emerging ideas about hybrid vigour". *J. Exp. Bot.* 63 (18): 6309–14.

# Dominance

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- The genetic **dominance hypothesis** attributes the superiority of hybrids to masking the expression of undesirable (deleterious) recessive alleles from one parent by dominant (usually wild-type) alleles from the other.
- It attributes the poor performance of inbred strains to the expression of homozygous deleterious recessive alleles.

# Complete Dominance: complementation



- ❑ If mutations are in different genes, complementation occurs.
- ❑ A complementation test example. Two strains of flies are white eyed because of two different autosomal recessive mutations which interrupt different steps in a single pigment-producing metabolic pathway.
- ❑ Flies from Strain 1 have complementary mutations to flies from Strain 2 because when they are crossed the offspring are able to complete the full metabolic pathway and thus have red eyes.
- ❑ Effects of hybrid vigor is seen in polygenic traits.

# Overdominance

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- The genetic overdominance hypothesis states that some combinations of alleles (by crossing two inbred strains) are advantageous when paired in a heterozygous individual.
- This hypothesis is used to explain the persistence of some alleles (such as the Sickle cell trait allele) that are harmful in homozygotes. PKU and Warfarin are two other examples.

# Overdominance

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- Under normal circumstances, such harmful alleles would be removed from a population through the process of natural selection.
- Like the dominance hypothesis, it attributes the poor performance of inbred strains to expression of such harmful recessive alleles.
- In any case, outcross matings provide the benefit of masking deleterious recessive alleles in progeny.
- This benefit has been proposed to be a major factor in the existence and maintenance of sexual reproduction among eukaryotes. It introduces shuffling of genes for increased genetic variance.



# Epigenetic heterosis

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- An epigenetic contribution to heterosis has been established in plants, and it has also been reported in animals.
- MicroRNAs (miRNAs), discovered in 1993 in *C. Elegans*, are a class of non-coding small RNAs which repress the translation of messenger RNAs (mRNAs) or cause degradation of mRNAs (RNA silencing).
- In hybrid plants, most miRNAs have non-additive expression (it might be higher or lower than the levels in the parents).
- This suggests that these small RNAs are involved in the growth, vigor and adaptation of hybrids.
- Produced from the introns, miRNAs turn off genes, helping different proteins made in different cells. The cells have the same genes, but they are different.

# Epigenetic heterosis

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- miRNAs sometimes cause histone modification and DNA methylation of promoter sites, which affects the expression of target genes (gene regulation).
- It was also shown that hybrid vigor in a hybrid of two *Arabidopsis* species was due to epigenetic control in the upstream regions of two genes, which caused major downstream alteration in chlorophyll and starch accumulation.
- The mechanism involves methylation of specific amino acids in histone H3, a protein closely associated with DNA, which can either activate or repress associated genes.

# Introns

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- ❑ Transcription, the conversion of DNA to RNA, is one of the most fundamental processes in cell biology.
- ❑ However, only about 3% of our total DNA encodes genes to be transcribed.
- ❑ RNA polymerase II, the enzyme that transcribes DNA to RNA, relies on a large set of proteins known as transcription factors to recognize the coding sequences and to transcribe the correct genes, in the correct cell type, at the correct time.

# Complete dominance

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- AA=2
- Aa=2
- aa=1
- 4 loci, recessive genotypes contribute 1 unit to phenotype, dominants 2

AA bb CC dd      x    aa BB cc DD  
2 + 1 + 2 + 1 = 6      1 + 2 + 1 + 2 = 6

F<sub>1</sub>:                    AaBbCcDd  
                          2+2+2+2=8

# OverDominance

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- 4 loci, recessive genotypes contribute 1 unit to phenotype,
- heterozygous genotypes 2 unit,
- homozygous dominants 1.5 unit.

aa bb CC DD    x    AA BB cc dd  
1+1+1.5+1.5=5    1.5+1.5+ 1 + 1=5

F<sub>1</sub>:                    AaBbCcDd  
                          2+2+2+2=8

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*Inbreeding*

***Positive Assortative Mating***

# Inbreeding

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- **Inbreeding** is positive assortative mating because individuals mate with relatives and these relatives are genetically similar to them.
- Inbreeding is measured using the coefficient  $F$ .
- $F$  is the amount of decrease in heterozygosity. This decrease occurs in the heterozygosity expected under HW equilibrium ( $2pq$ ).

# Important Point:

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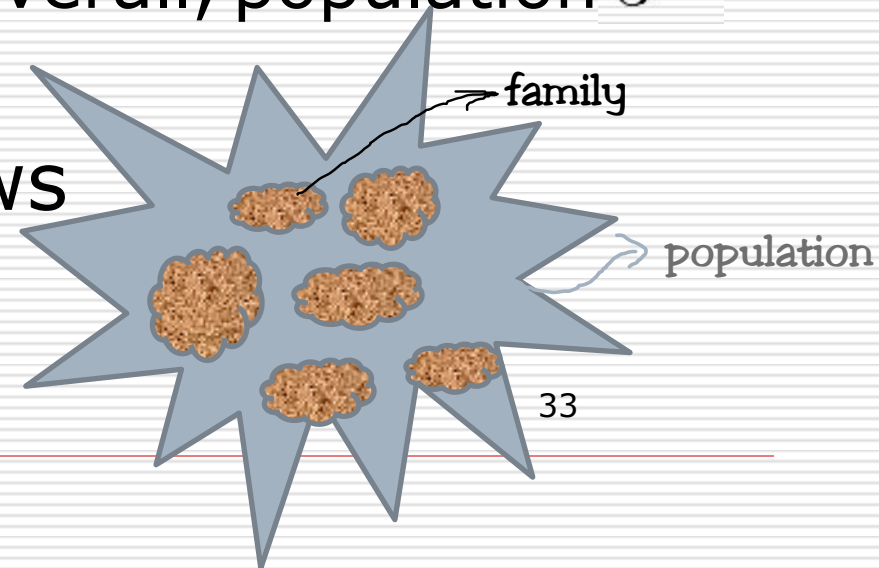
- Inbreeding can decrease heterozygosity but does not decrease genetic variation by itself.
- Selection is added to inbreeding if  $\sigma^2$  is to be decreased.
- Select and breed some family groups, and send the other families away



# Inbreeding and Selection

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- $\sigma^2$  decreases because family members resemble each other.
- Families are like subpopulations:
  - With inbreeding only, within group  $\sigma^2$  decreases but the overall, population  $\sigma^2$  stays the same.
- Lions and three cows



# Coefficient of inbreeding & Coefficient of relationship

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- ❑ We look like each other, Why?
- ❑ We have common ancestors 😊
- ❑ Because of their common ancestors, two animals or plants carry common genes: *identical by descent*
- ❑ As relatives are closer, they carry more common genes. Brothers carry more idb genes compared to cousins.

# Coefficient of inbreeding & Coefficient of relationship

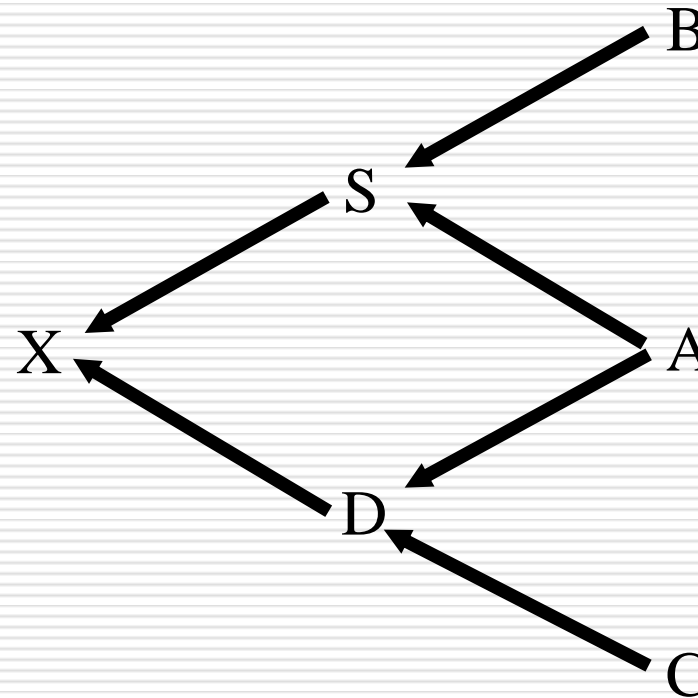
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- Each individual inherit half of their genes from their mother, and the other half ( $1/2$ ) from their father.
- This is called ***Coefficient of Relationship***= Ratio of the genes identical by descent in two individuals.

# Arrow Diagram

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Each arrow is  $\frac{1}{2}$  because  $\frac{1}{2}$  genes are transmitted from each parent



S and D are half  
sibs=half siblings

Coefficient of relationship in some relatives.  
More common genes in closer relatives

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Identical twins, clones	1
Full sibs	$\frac{1}{2}$ (0.50)
Parent-offspring	$\frac{1}{2}$ (0.50)
Half sibs	$\frac{1}{4}$ (0.25)
Grandparent-grandchild	$\frac{1}{4}$ (0.25)
Uncle/aunt	$\frac{1}{4}$ (0.25)
Cousins	$\frac{1}{8}$ (0.125)
Half cousins (from half sibs)	$\frac{1}{16}$ (0.06)

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# Coefficient of inbreeding & Coefficient of relationship

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- The inbreeding coefficient is defined as the probability of homozygosity in a locus due to common ancestors.
- Thus the inbreeding coefficient for an animal ( $F$ ) is equal to half of the relationship coefficient between the parents.
- The relationship coefficient ( $R$ ) is between two individuals.

# Inbreeding depression

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- ❑ Depression?
- ❑ Inbreeding depression is the reduced biological fitness in a given population as a result of inbreeding - that is breeding of related individuals.
- ❑ Decrease in biological fitness lead to changes in reproduction performance and ability to survive.

# Inbreeding depression

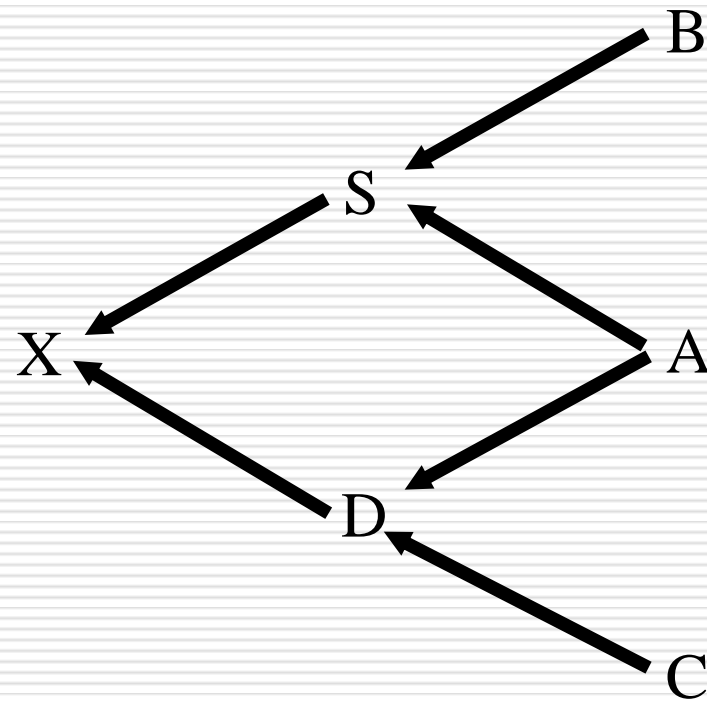
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- Inbreeding depression in natural population is often the result of a population bottleneck.
- The higher the genetic variation within a breeding population, the less likely it is to suffer from inbreeding depression.
- Traits with low heritability (reproduction and survival) suffer worse from depression



# Arrow Diagram

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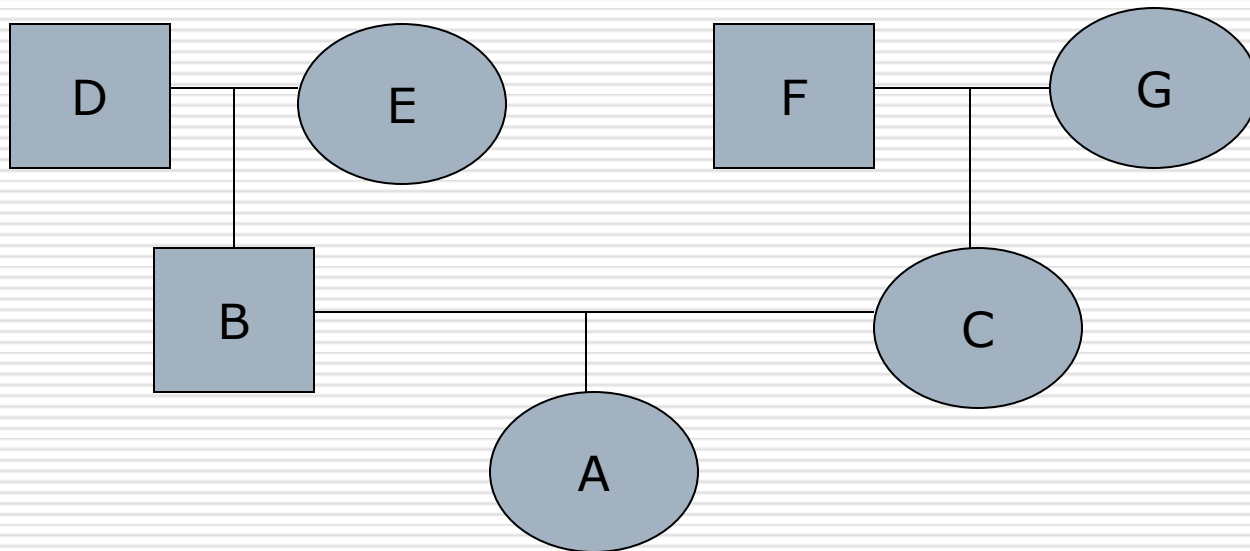


$$R_{SD} = \frac{1}{4} \text{ (half sibs)}. F_X = \frac{(\frac{1}{4})}{2} = \frac{1}{8}$$

# Genetic positive assortative mating: Inbreeding

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- Inbreeding is mating individuals that have stronger relationships than the other individuals in the population.



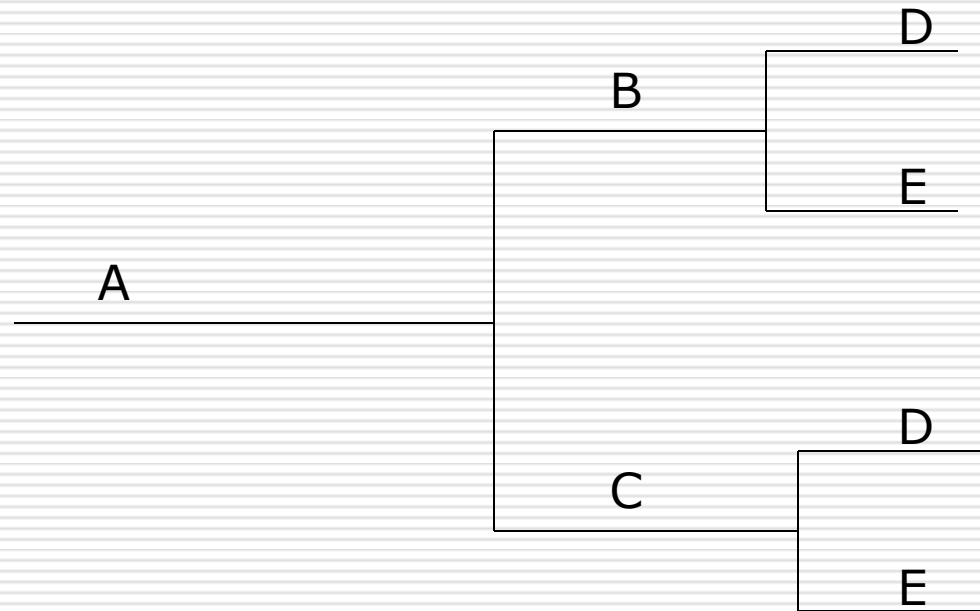
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B are C not relatives, inbreeding (F) is zero

# Coefficient of inbreeding & Coefficient of relationship

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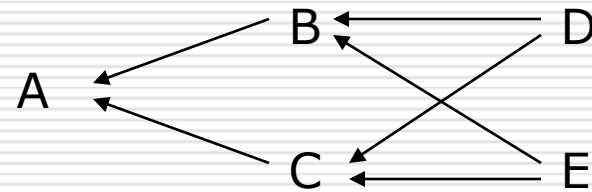
Fathers are represented with letters at the top and moms are represented with the letters at the bottom.



# Coefficient of inbreeding & Coefficient of relationship

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Coefficient of relationship is the percentage of idb genes carried by two individuals due to common ancestors.



Coefficient of relationship can be calculated by following paths over common ancestors.

$$R = \sum_{i=1}^n \left( \frac{1}{2} \right)^s$$

$n$  = # of paths over common ancestors

$s$  = # of arrows from the individual to common ancestor and then to the other individual

# Inbreeding Coefficient

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- Inbreeding coefficient is the probability that two alleles in a locus is identical by descent.
- These two genes (alleles) are copies of a gene replicated from a common ancestor.

# Inbreeding Coefficient

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- Half of the coefficient of relationship is the inbreeding coefficient of the offspring of these two individuals.
  - The common ancestor must have a zero inbreeding coefficient

$$F_x = \frac{1}{2} R_{SD} = \sum_{i=1}^n \left( \frac{1}{2} \right)^{p_1 + p_2 + 1}$$

1 is added to add a 1/2 for F

- S dad, D mom,  $p_1$  # of arrows from one parent to the common ancestor,  $p_2$  # of arrows from the other parent to the same common ancestor, we add it all up.

# Inbreeding Coefficient

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- If the common ancestor is inbred, the formula has an addition:

$$F_x = \sum_{i=1}^n \left[ \left( \frac{1}{2} \right)^{p_1 + p_2 + 1} (1 + F_A) \right]$$

# Coefficient of inbreeding & Coefficient of relationship

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- Why use inbreeding since it is so bad?
- May want to mate a good male with more females,
  - Even a superior individual may be cloned
- Many breeds are developed using inbreeding
- Assortative mating may be required for sympatric speciation, meaning the evolution of a new species without geographic isolation.



# Inbreeding or not inbreeding?

## 1. Positive effects

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- Inbreeding may fix some gene combinations and make sure it is transmitted to the next generations.
- Purebreds may be wanted for hybrids
- Lines within purebreds may be desired for hybrids
- Inbred strains (lines) may be desired for scientific studies
  - the usual procedure is mating of brother-sister pairs for a minimum of 20 generations

# Inbreeding or not inbreeding?

## 2. Negative effects

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- ❑ Inbreeding causes genes with deleterious effects to come out and decreases general vigor, causing inbreeding depression.
- ❑ Low  $h^2$  traits such as reproduction and survivability are affected.
- ❑ Inbreeding is usually avoided due to its bad effects

# Hybrids

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- Wild mice warfarin resistance
  - Vitamin K coagulates blood
  - Warfarin prevents blood coagulation (clotting) and it is used as a rat/rodent poison
- Two alleles, **K** and **k**
- **kk** mice get sick due to Vitamin K deficiency
- **KK** mice die if given Warfarin
- **Kk** mice are resistant to Warfarin and they don't have Vitamin K deficiency

# Heterosis=Reverse inbreeding

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- Heterosis and its applications are why chicken, corn etc. are cheaper.
  - White meat was more expensive/guest food when I was a kid.
- Hybrid vigor is an offspring performing better than the average of its parents. It is the improved or increased function of any biological quality in a hybrid offspring.
- An offspring exhibits heterosis if its traits are enhanced as a result of mixing the genetic contributions of its parents.
- Hybrid animals usually have better reproductive capabilities compared to purebreds. Malaria- sickle cell anemia

# Heterosis 2

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- Why hybrid animals—for example chicken—is not used for producing chicks, but the farmers purchase a new crop of chicks each time??
- Why progeny from hybrid parents do not perform as well as its hybrid parents?
- How can these genetic companies sell animals/seeds continuously?

# Gender in Poultry

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- The male reproductive organs are located on the outside of the body and are relatively easy to see, even in newborns.
- In male birds, the reproductive organs are inside the body cavity.
  - This makes sexing newly hatched chicks difficult.

# Gender in Poultry

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- Cloaca can be squeezed gently, allowing the chicken sexer to see if the chick has a small "bump", which would indicate that the chick is a male.
  - This is called venting, and requires experience.
- Cloaca is the only opening for the intestinal, reproductive, and urinary tracts of all amphibians, birds and reptiles possess this orifice, from which they excrete both urine and feces.
  - Chicken/pigeon fertilizer is more valuable.
- Placental mammals possess two or three separate orifices for evacuation.

# ZW sex determination

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## AVIAN GENETICS - GENDER DETERMINATION

MAMMALS  
(e.g., humans, cows, etc.)

Male = XY  
Female = XX

Males determine the sex of the offspring



BIRDS  
(including all poultry species)

Male = ZZ  
Female = ZW

Females determine the sex of the offspring



# ZW sex determination

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- In the ZW system, the ovum determines the sex of the offspring, in contrast to the XY sex-determination system and the XO sex-determination system, wherein the sperm determines the sex.
- The letters Z and W are used to distinguish this system from the XY system.
- Males are the homogametic sex (ZZ), while females are the heterogametic sex (ZW).
- The Z chromosome is larger and has more genes, just like the X chromosome in the XY system.

# ZW sex determination

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- It is unknown whether the presence of the W chromosome induces female features or the duplication of the Z chromosome induces male ones;
- unlike mammals, no birds with a double W chromosome (ZWW) or a single Z (ZO) have been discovered.
- It appears possible that either condition could cause embryonic death, or that both chromosomes could be responsible for sex selection.
  - Smith CA, Roeszler KN, Hudson QJ, Sinclair AH (2007). "Avian sex determination: what, when and where?". *Cytogenet. Genome Res.* 117 (1-4): 165-73.

# Gender in Poultry

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- ❑ The Barred Plymouth Rock breed carries a gene for barring (B) that is carried on the Z chromosome.
- ❑ This gene produces a white bar on normally a black feather.
- ❑ The gene has incomplete dominance over the non-barring gene (b).
- ❑ In adults, the male, with two barring genes (BB), has feathers with broader white bars than those of the female, who has only one of the barring genes (B\_).
- ❑ As a result, females are overall lighter in color.

# Gender in Poultry

- ❑ Male offspring get a Z chromosome from each parent
- ❑ Females get a Z from their father and a W from their mother
- ❑ Z chromosomes never pass from mother to daughter
- ❑ W chromosomes always pass from mother to daughter

B = Barred      b = non-barred

		<i>FEMALE</i>	
		Z <sup>B</sup>	W
<i>MALE</i>	Z <sup>B</sup>	Z <sup>B</sup> Z <sup>B</sup>	Z <sup>B</sup> W
	Z <sup>b</sup>	Z <sup>b</sup> Z <sup>b</sup>	Z <sup>b</sup> W

Z<sup>B</sup>Z<sup>B</sup> = Barred male with broad white bars  
Z<sup>B</sup>W = Barred female with more narrow white bars

# Silver/Gold for sexing

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- Another characteristic that has been used in some commercial strains is the silver (S) and gold (s) color alleles. Gold males are mated to silver females.
- $Z^sZ^s \times Z^SW \rightarrow Z^sZ^S$  and  $Z^sW$
- The resulting males will be silver, females will be gold.
- The silver and gold genes have been used in both the egg and meat chicken (broiler) industries.
- Some of the strains of brown-shelled egg layers have the silver/gold sexing characteristics.
- The gold/silver and barred/non-barrred genes have been combined and used in some commercial brown-shell egg layers.

# Silver/Gold for sexing

S = silver      s = gold

		<b>FEMALE</b>	
		Z <sup>S</sup>	W
<b>MALE</b>	Z <sup>s</sup>	Z <sup>S</sup> Z <sup>s</sup>	Z <sup>s</sup> W
	Z <sup>s</sup>	Z <sup>S</sup> Z <sup>s</sup>	Z <sup>s</sup> W

Z<sup>S</sup>Z<sup>s</sup> = Silver male  
Z<sup>s</sup>W = Gold female

B = Barred      b = non-barred  
S = silver      s = gold

		<b>FEMALE</b> <i>Barred Plymouth Rock (silver, barred)</i>	
		Z <sup>SB</sup>	W
<b>MALE</b> <i>Rhode Island Red (gold, non-barred)</i>	Z <sup>sb</sup>	Z <sup>SB</sup> Z <sup>sb</sup>	Z <sup>sb</sup> W
	Z <sup>sb</sup>	Z <sup>SB</sup> Z <sup>sb</sup>	Z <sup>sb</sup> W

Z<sup>SB</sup>Z<sup>sb</sup> = Black and white barred male  
Z<sup>sb</sup>W = Black-red, non-barred female

# Gender in Poultry

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# Gender in Poultry

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