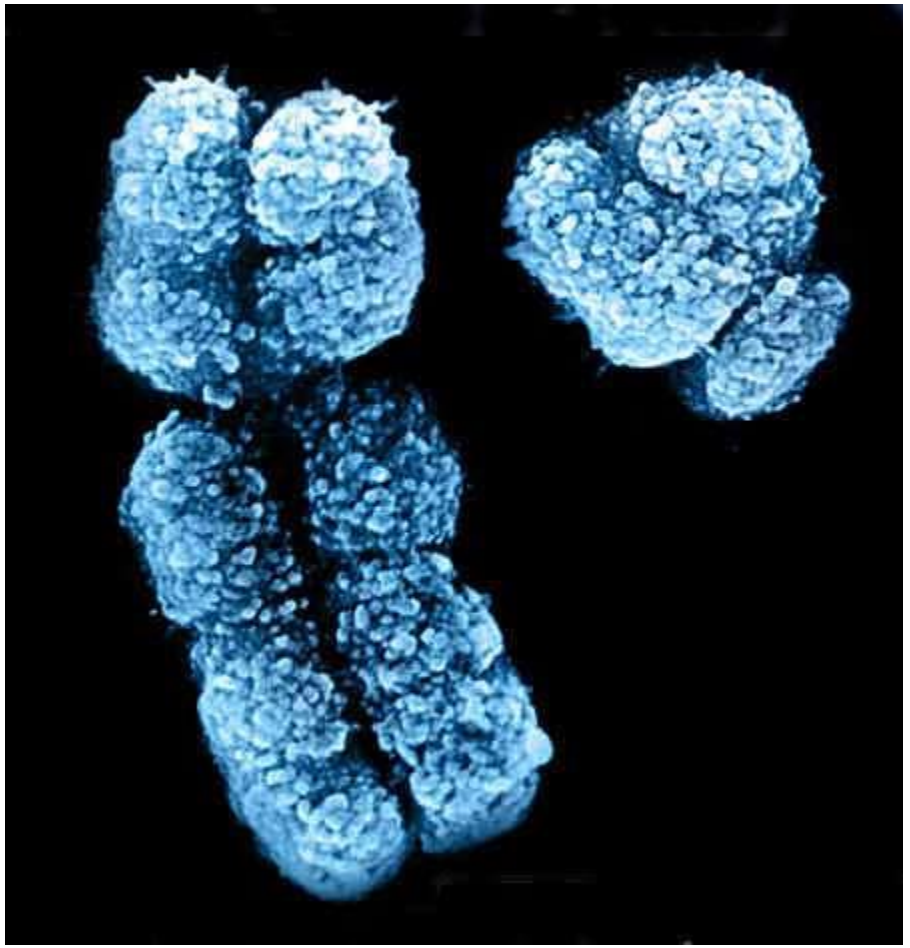
Today's class – continue: patterns of inheritance (non-Mendelian) and genetic crosses



Source: https://phys.org/news/2014-05-evolutionary-biologists-glimpse-early-stages.html

Alexandra Mykitiuk

Science Student Recognition Awards Committee (SSRA Committee).

We are hosting this night to celebrate and recognize UBC Science students for their initiative and actions that have made an impact on themselves and their communities.

Hey UBC Science Students!

SUS wants to celebrate and acknowledge YOUR amazing contributions to the UBC Science community and beyond! Whether you made a huge impact in a community initiative or your personal growth and development this academic year, you can nominate yourself for your great work! You also have the opportunity to nominate any peers or TAs for their excellency in the community as well.

The nomination form can be accessed in the SUS link tree or through the QR code above (next slide). Nominations are available until February 13, 2023 at 11:59 pm!

- If you have any further questions regarding the blurb, please feel free to reach out (Caleb Lowe, clowe02@student.ubc.ca).


Science Student Recognition Awards
NOMINATIONS OPEN
DUE: FEBRUARY 13, 2023
SCIENCE UNDERGRADUATE SOCIETY
UNIVERSITY OF BRITISH COLUMBIA

Midterm #1 (20%)– Genetics Unit (all)

One week from today – Tuesday, February 7th

- In class
- Start time of ~11:10 am (-224 class) or ~5:10 pm (-221)
- Duration – 50 minutes
- No lecture after class
- You may write in pen or pencil, but exams written in pencil or erasable pen or questions with whiteout will not be remarked.
- Bring: pen or pencils, calculator, study sheet (8.5” x 11” – both sides - any note, figures, etc. in your handwriting).
- Point form is okay – but any logical connections must be explicit
- Drawings okay – but must be annotated

- Exam not yet finalized. Currently, 3 major questions with subquestions, plus one bonus question
- Combination of multiple choice, select all, short answer, explanation questions
- Total marks – not yet known.

Review/Study Sessions

Ruby & Christie Review Session (new questions) – On Zoom.

- Day and Time TBA (poll on Canvas)
- will be recorded
- Zoom link will likely be the office hours link, but will confirm via announcement
- slides will also be shared

Brett's study session: this Friday, February 3, in BIOL1000, 5:00 pm – 7:00 pm

- bring questions, Brett will also be stepping you through a few "things"
- not recorded
- highly recommend attending – Brett is very clear.

I will likely have extra office hours this Sunday on Zoom (time TBA).
I will also be on Zoom on Monday night until midnight – start time TBA.

This Sunday's assignments

This Sunday @ 11:59 pm is the deadline for:

- Quiz 4 – Pedigrees & Probabilities (pedigrees – will be covered in Thursday's class)
- Worksheet #5 - Determining Inheritance Patterns – *Drosophila (can complete after today's class)*

OPTIONAL (no submission date):

- Worksheet #6 – Inheritance of Eye Colour and Fur Colour – Rats – only worksheet with non-dominant relationship between alleles *(can complete after today's class).*
- Pedigrees

Last class – autosomal modes of inheritance (patterns)

Autosomal dominant mode of inheritance (e.g. Peas: round seeds, yellow seeds, purple flowers, Huntington's Disease, Achondroplasia, neurofibromatosis type 1 – display a pattern of autosomal dominant inheritance)

- Means the gene for this trait is on an autosome
- An individual needs only one dominant allele (functional protein) to display the dominant phenotype; so, an individual with this phenotype could be homozygous dominant or heterozygous (e.g. TT or Tt)

Autosomal recessive mode of inheritance (e.g. Peas: wrinkled seeds, green seeds, white flowers, *cystic fibrosis, sickle cell anemia, Tay Sachs disease*)

- means the gene for this trait is on an autosome
- An individual must have two recessive alleles (non-functional or reduced function) to display the recessive phenotype (e.g. tt).

Some autosomal patterns

If two heterozygous individuals are crossed for a trait coded for by one gene, two alleles, with a dominant/recessive relationship, how many offspring phenotypes* would you expect to see and in what ratio?

*not including individual's sex

2 phenotypes, 3:1 phenotypic ratio – dominant:recessive

If you crossed a heterozygote for two traits with a homozygous recessive individual, the genes were on different chromosomes, dominant/recessive relationship between alleles, how many offspring phenotypes* would you expect and in what frequencies?

4 phenotypes, 25%, 25%, 25%, 25% phenotypic ratio

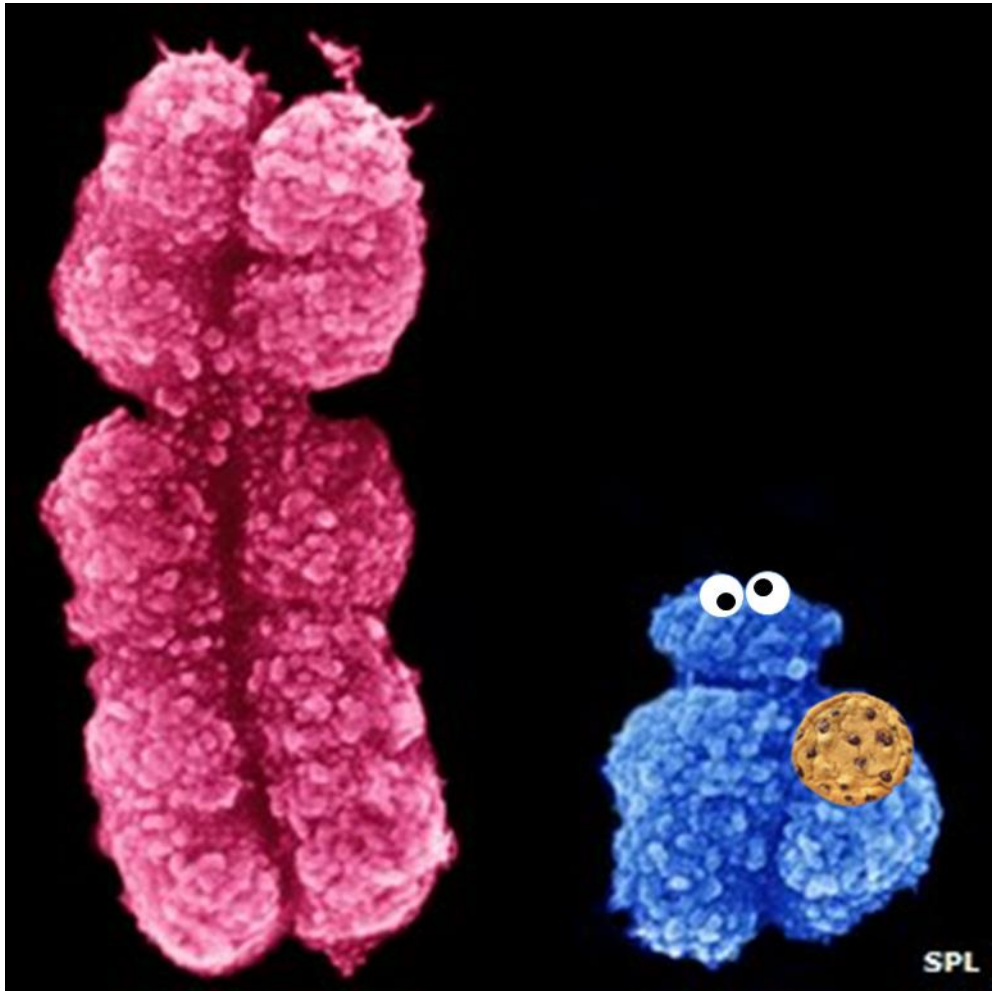
If you crossed a heterozygote for two genes with a homozygous recessive individual, the genes were closely linked on the same chromosome, dominant/recessive relationship between alleles, how many offspring phenotypes* would you expect and in what rat?

4 phenotypes, greater frequency of original P-generation phenotypes (non-recombinant) and fewer recombinant phenotypes

Questions?


As Pooh prepared to beam down
he suddenly realised he was
wearing a red shirt!

X-linked dominant and recessive modes of inheritance



Sex-linked* traits exhibit different patterns of inheritance

* Important: “Sex linkage” means something different than “physical linkage”

- The gene in question is located on one of the sex chromosomes (X or Y)
  - We would call these genes either X-linked or Y-linked

Patterns for X-linked traits?

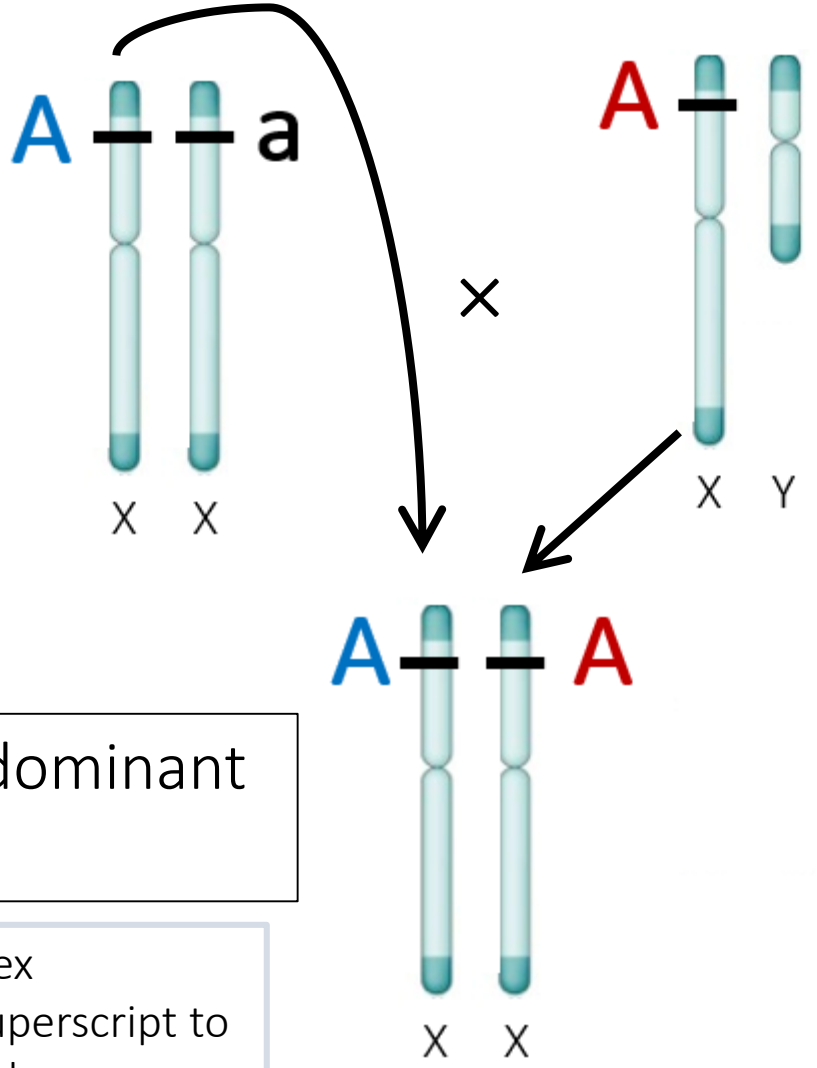
It depends on whether the dominant allele is on the X-chromosome of the mother or father

For simplicity, we will call XX individuals “female” and XY individuals “male”, but remember that sex phenotypes can be more complex and are not entirely determined by chromosomes.

X-linked trait inheritance: **Females (XX)**

mother

$X^A X^a$



father

$X^A Y$

Females inherit an X-chromosome from both parents

Genotype: Homozygous dominant
Phenotype: Dominant

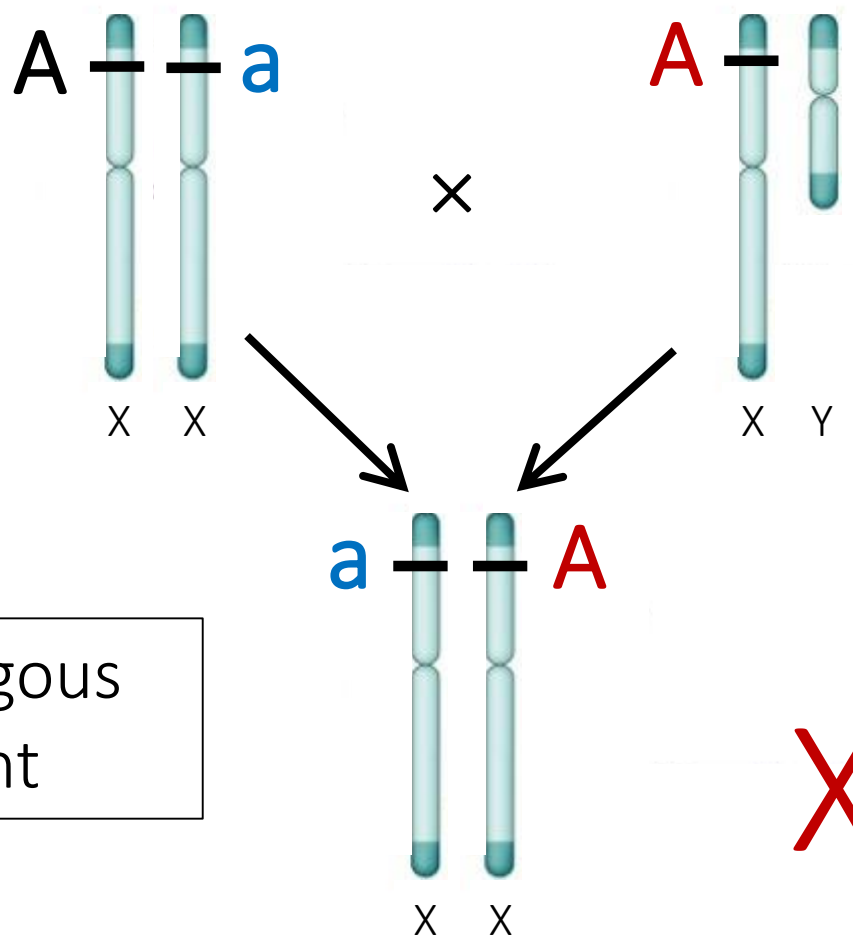
$X^A X^A$

Notation: we always write the sex chromosome and the gene in superscript to make clear the gene is sex-linked

X-linked trait inheritance: **Females**

Mother

$X^A X^a$



Father

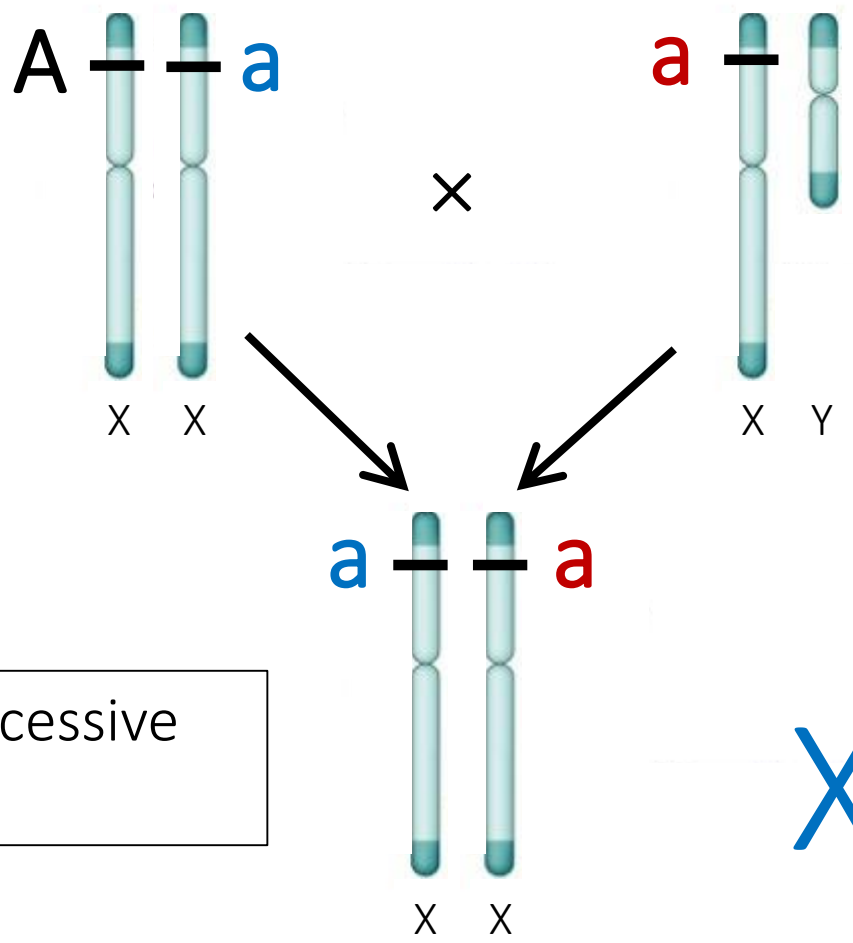
$X^A Y$

Genotype: Heterozygous
Phenotype: Dominant

$X^A X^a$

X-linked trait inheritance: **Females**

$X^A X^a$



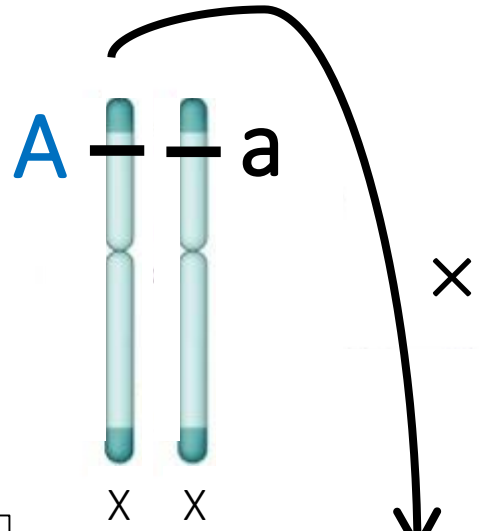
$X^a Y$

Genotype: Homozygous recessive
Phenotype: Recessive

$X^a X^a$

X-linked trait inheritance: **Males**

$X^A X^a$



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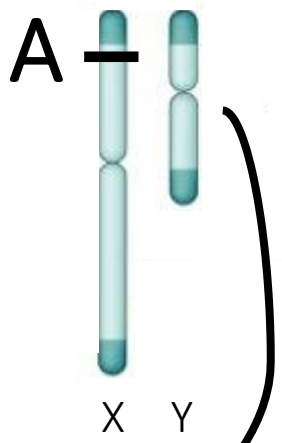
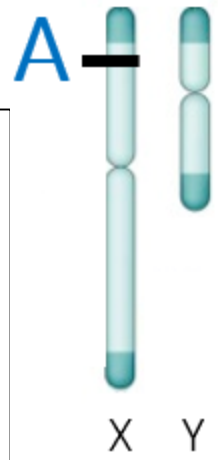


$X^A Y$

Males inherit X-chromosome from one parent and Y-chromosome from other parent

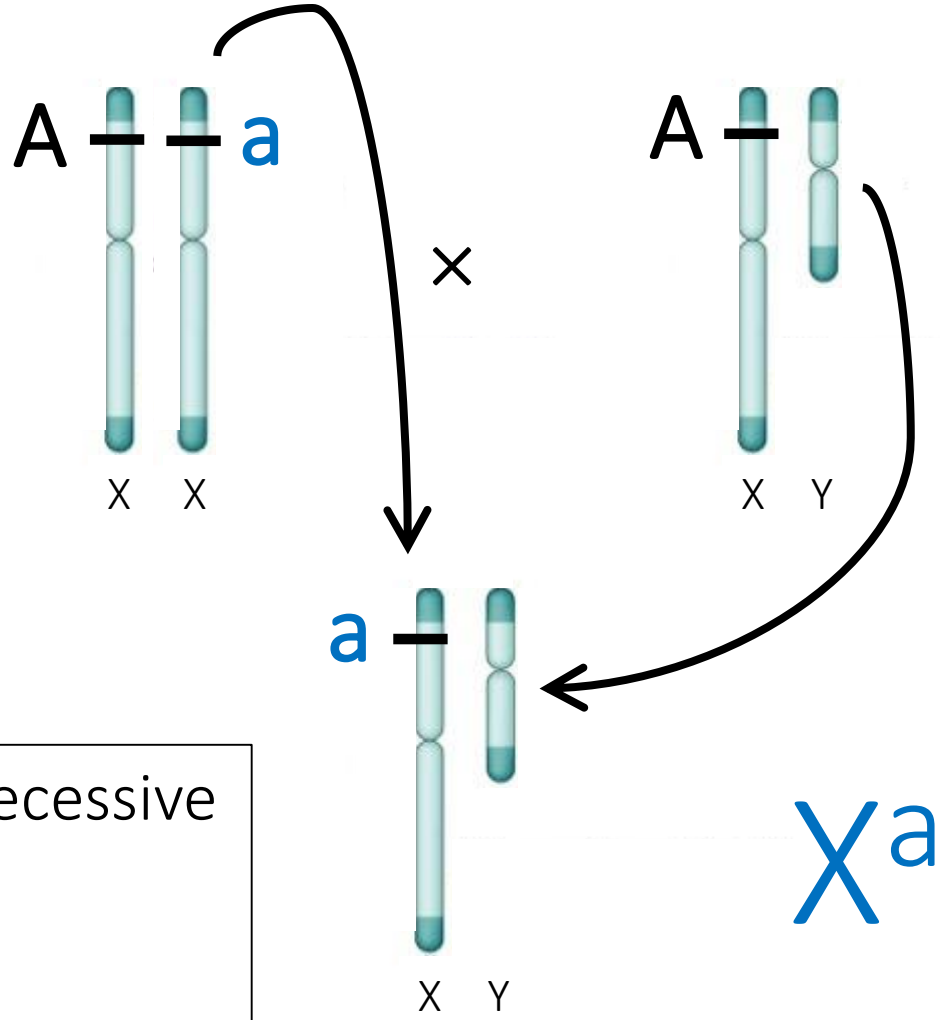


$X^A Y$

Genotype: Hemizygous* dominant
Phenotype: Dominant
FEMALE parent determines son's phenotype
*you do not need to know this term

X-linked trait inheritance: **Males**

$X^A X^a$



$X^A Y$

$X^a Y$

Genotype: Hemizygous* recessive
Phenotype: Recessive

*you do not need to know this term

X-linked trait inheritance: **Females**

Key takeaways

- A female inherits an X-chromosome from both parents.
  - If the male parent carries the dominant allele on his X-chromosome, the female offspring must also have the dominant phenotype because she will inherit this dominant allele.
  - A female can only have the recessive phenotype if she inherits the recessive allele from both parents.

X-linked trait inheritance: **Males**

Key takeaways

- The male parent never donates his X chromosome to male offspring, only his Y chromosome
- The female parent genotype determines the phenotype of her male offspring
  - If she donates a dominant allele, the male offspring phenotype will be dominant
  - **If she donates a recessive allele, the male offspring phenotype will be recessive**
- In other words, male offspring only need one recessive allele to have a recessive phenotype

Learning goals – X-linkage

- Determine whether a gene is on a sex chromosome (X only)
- Predict what gamete genotypes a parent can produce and in what proportions - for genes that are on an X-chromosome
- Predict offspring genotypes and phenotypes and in what proportions – for genes that are on an X-chromosome
- Be able to calculate the probability that two parents will have an offspring with a specific phenotype (one and two traits)

Patterns for X-linked inheritance

Look for sex bias (differences) in the phenotypes of the progeny (i.e. males and female differ)

- Could appear in F1 **OR** F2 generation

  - it depends upon whether mother or father is carrying the dominant allele.

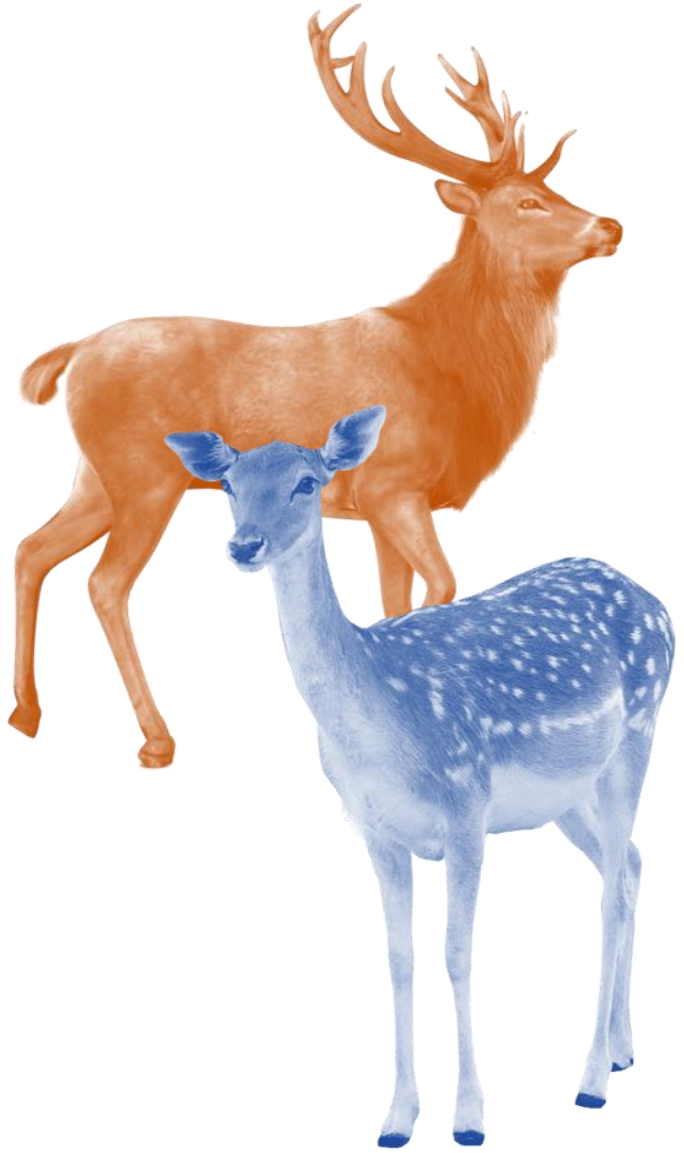
Example Let's pretend that coat colour in deer is an X-linked trait:

- Red coat ($\mathbf{X^R}$) is dominant to blue coat ($\mathbf{X^r}$)

X-linked - predicting genotypes and phenotypes

Example #1 - If we cross a blue female with a red male, if the trait is X-linked, what will the F1 genotypes and phenotypes be? Blue=recessive, Red=dominant

Ask yourself:
Who is the male donating his dominant allele to?
Who is the female donating her recessive allele to?



X-linked - predicting genotypes & phenotypes

If we cross a blue female with a red male, what will the F1 genotypes and phenotypes be?

	X^r	X^r
X^R	X^RX^r Red coat	X^RX^r Red coat
Y	X^rY Blue coat	X^rY Blue coat

In this example, **the male is carrying the dominant allele (and females the recessive a.) - You see a sex difference in the F1 gen.**

Male will donate dominant allele (red colour) to daughters; and y-chromosome to sons.
- So all daughters must have dominant phenotype

Mother will donate recessive allele to daughters and sons.
- So all sons must have recessive* phenotype

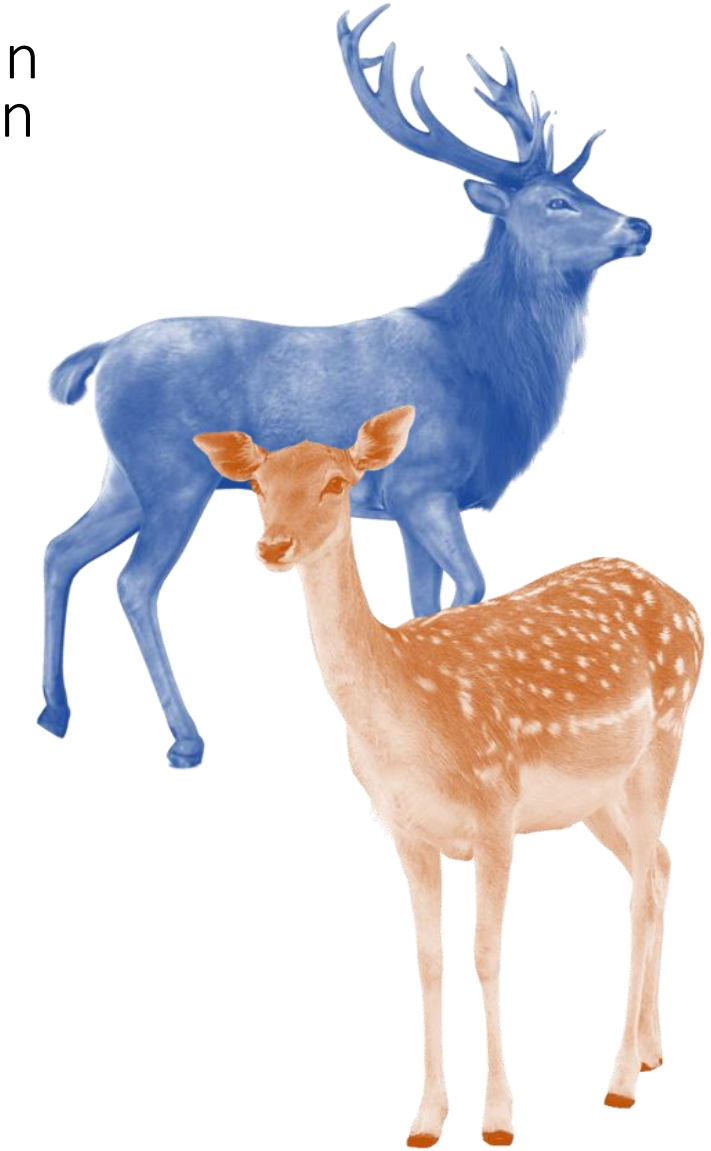
*note – pre-lecture slides mistakenly said dominant.

Example #1 – if we crossed F1 deer

If we cross the F1s, what phenotype frequencies do we see in the F2 generation? Is there a phenotypic difference between males and females? Blue=recessive, Red=dominant

No – so if P-male is carrying dominant X-linked allele, sex difference shows up in F1 not F2

	X^R	X^r
X^r	X^RX^r Red coat	X^rX^r Blue coat
Y	X^RY Red coat	X^rY Blue coat

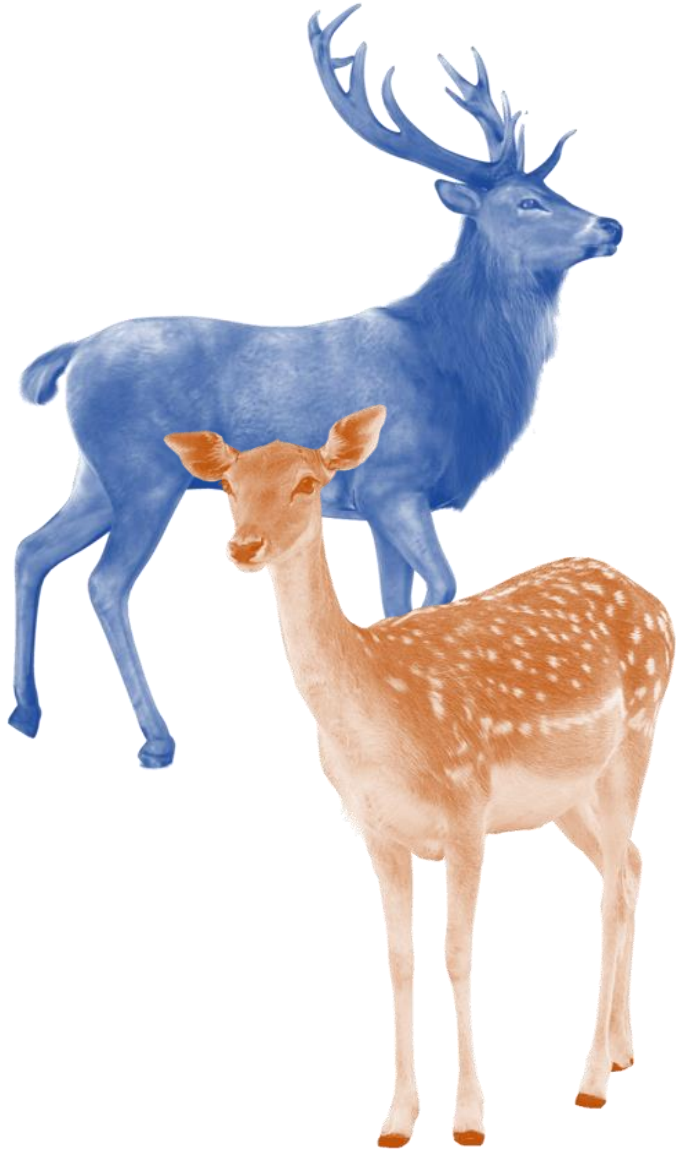


2nd deer example, X-linked trait

What happens in a reciprocal cross (blue male crossed to homozygous red female)? Blue=recessive, Red=dominant

Ask yourself:
Who is the female donating her dominant allele to?
Who is the male donating his recessive allele to?

	X^R	X^R
X^r		
Y		



2nd deer example- X-linked trait

What happens in a reciprocal cross (blue male crossed to homozygous red female)?

Now, there is no difference in the phenotypes of the F1 male and female deer!

	X^R	X^R
X^r	X^RX^r Red coat	X^RX^r Red coat
Y	X^RY Red coat	X^RY Red coat

Father will donate recessive allele to daughters.

But, mothers will donate dominant allele to both daughters and sons.

Therefore, the F1 individuals do not differ in phenotype, regardless of sex.

If this was the only cross we performed, we wouldn't know for sure if the gene was X-linked or autosomal.

Why F1 can tell you the relationship between alleles and not necessarily if gene is on autosome of X-chromosome

F2 generation, X-linked inheritance

Now, let's cross the male and female F1s (from the last question) and see what happens.

	X^R	X^r
X^R	X^RX^R Red coat	X^RX^r Red coat
Y	X^RY Red coat	X^rY Blue coat

- **We see a difference in the phenotypes of the F2 males and F2 females!!!**
- All F2 females are red.
- Half of the F2 males are red and half are blue.
- Sex difference = evidence of X-linkage.
- When mother carries dominant allele, sex difference shows up in F2 generation.
- <u>Do not make the mistake of thinking this is a 3:1 phenotypic relationship</u>

Question - probability

What is the probability that this cross (heterozygous female x male carrying dominant allele) will result in a baby deer that is male and red?

A. 0%
B. 25%
C. 37.5%
D. 50%
E. Not sure

	X^R	X^r
X^R	X^RX^R Red coat	X^RX^r Red coat
Y	X^RY Red coat	X^rY Blue coat

Question - probability

What is the probability that this cross will result in a baby deer that is male and red?

A. 0%

B. 25%

C. 37.5%

D. 50%

E. Not sure

For an autosomal trait, you would have to calculate the probability of having a "male" offspring, and the probably of having a red coat separately.

But, the probabilities of these two outcomes are already included in the Punnett Square

	X^R (1/2)	X^r (1/2)
X^R (1/2)	X^RX^R Red coat	X^RX^r Red coat
Y (1/2)	X^RY Red coat	X^rY Blue coat

Question – probabilities #2

What is the probability that this cross will result in a baby deer that is a blue male or a red female?

A. 18.75%
B. 25%
C. 50%
D. 75%
E. Not sure

	X^R	X^r
X^R	X^RX^R Red coat	X^RX^r Red coat
Y	X^RY Red coat	X^rY Blue coat

Question – probabilities #2

What is the probability that this cross will result in a baby deer that is a blue male or a red female?

A. 18.75%
B. 25%
C. 50%
D. 75%
E. Not sure

The question specifies "OR", so we should add the probabilities:

Probability of blue male = 0.25
Probability of red female = 0.50
Probability of either blue male or red female = 0.25 + 0.50 = 0.75

	X^R (1/2)	X^r (1/2)
X^R (1/2)	X^RX^R Red coat	X^RX^r Red coat
Y	X^RY Red coat	X^rY Blue coat

3-minute break

Additional notes on dominant/recessive trait relationships

1. Just because an allele is dominant in one relationship does not mean that it will be dominant in the presence of another allele. Not every locus is controlled by only 2 alleles!
   - E.g. A1 > A2, but A3 > A1
2. The dominant allele is not necessarily the most abundant allele in a population (or the dominant trait is not necessarily the most common)
   - Some diseases have a pattern of autosomal dominant inheritance
     - E.g. Huntington's disease, Marfan's syndrome
     - My numerous moles – autosomal dominant ☹

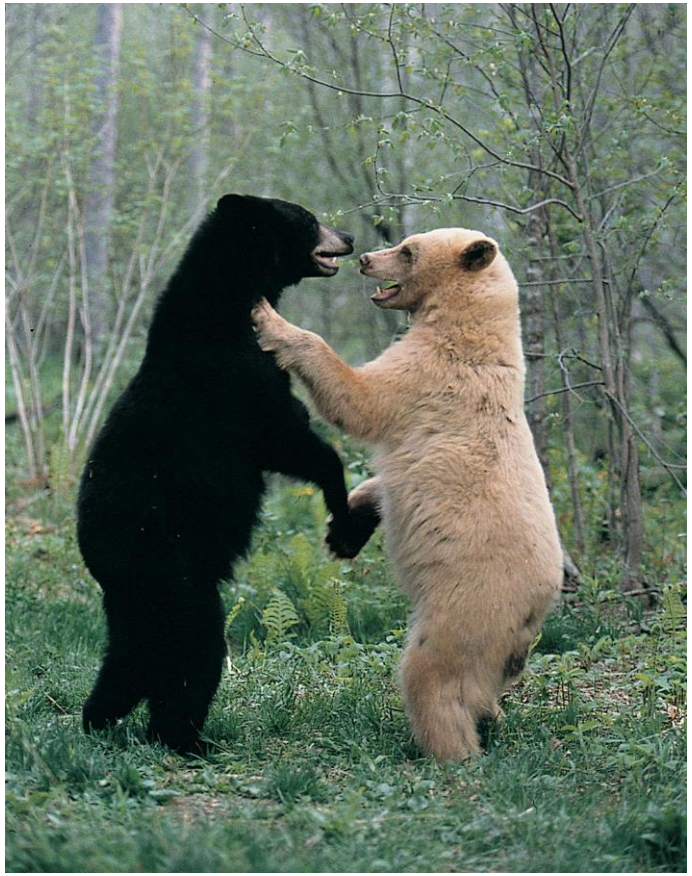
Learning goals – Non-dominance

- Determine whether two alleles have a dominant/recessive relationship or a non-dominant relationship
- Predict offspring genotypes and phenotypes and in what proportions – for a gene when alleles have a non-dominant relationship.

Dominant/recessive relationships & phenotypes

Under dominant/recessive relationships, a gene with 2 alleles can produce **2 possible phenotypes**:

# Genotypes	# Phenotypes
① *ff*	① White
② *Ff*	② Black
③ *FF*	



Non-dominant relationship between alleles

If neither allele is dominant, a gene with 2 alleles can produce 3 different phenotypes:

Important – for alleles with a non-dominant relationship, please due not use upper and lower case letters.

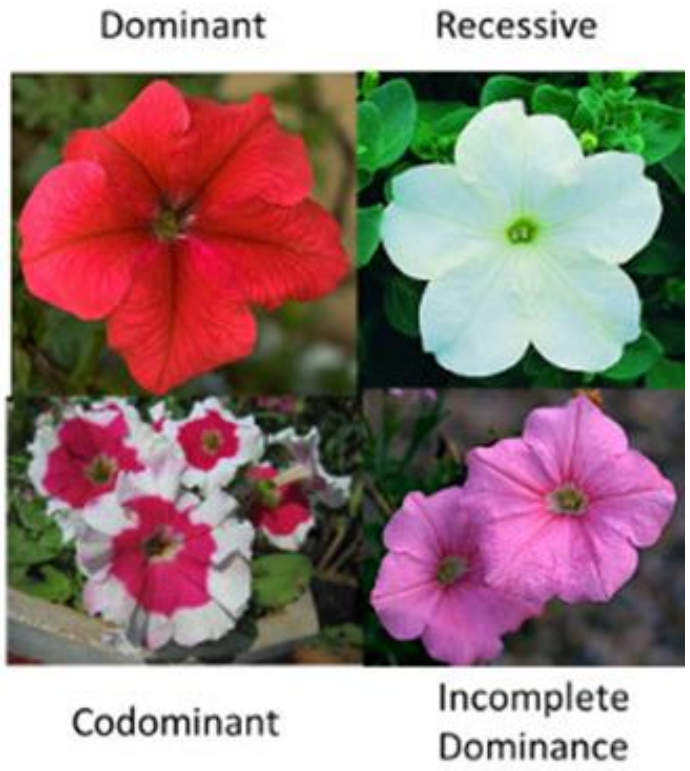
# Genotypes		# Phenotypes
① *ff*	⇨	① White
② *Ff*	⇨	② Grey
③ *FF*	⇨	③ Black

The heterozygote expresses aspects of both alleles (neither masks the other)

Non-dominant alleles can show **co-dominance*** or **incomplete dominance***

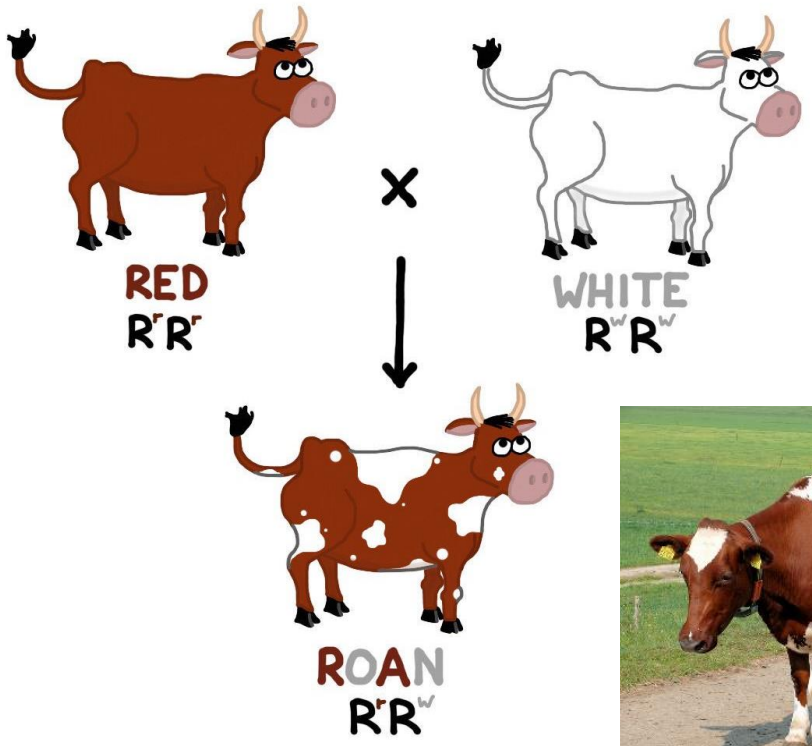
Under **co-dominance**, both traits appear in the heterozygote.

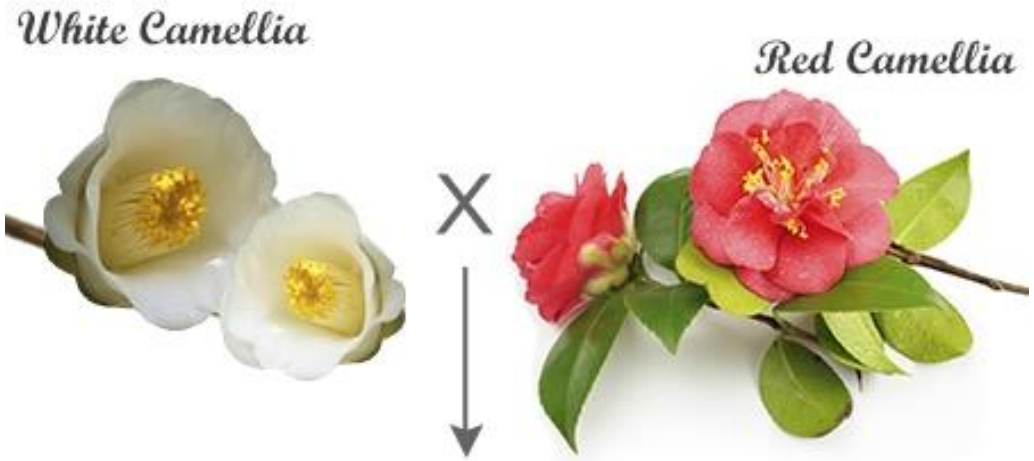
Under **incomplete dominance**, traits blend together.



*Not testable

Examples of **co-dominance**

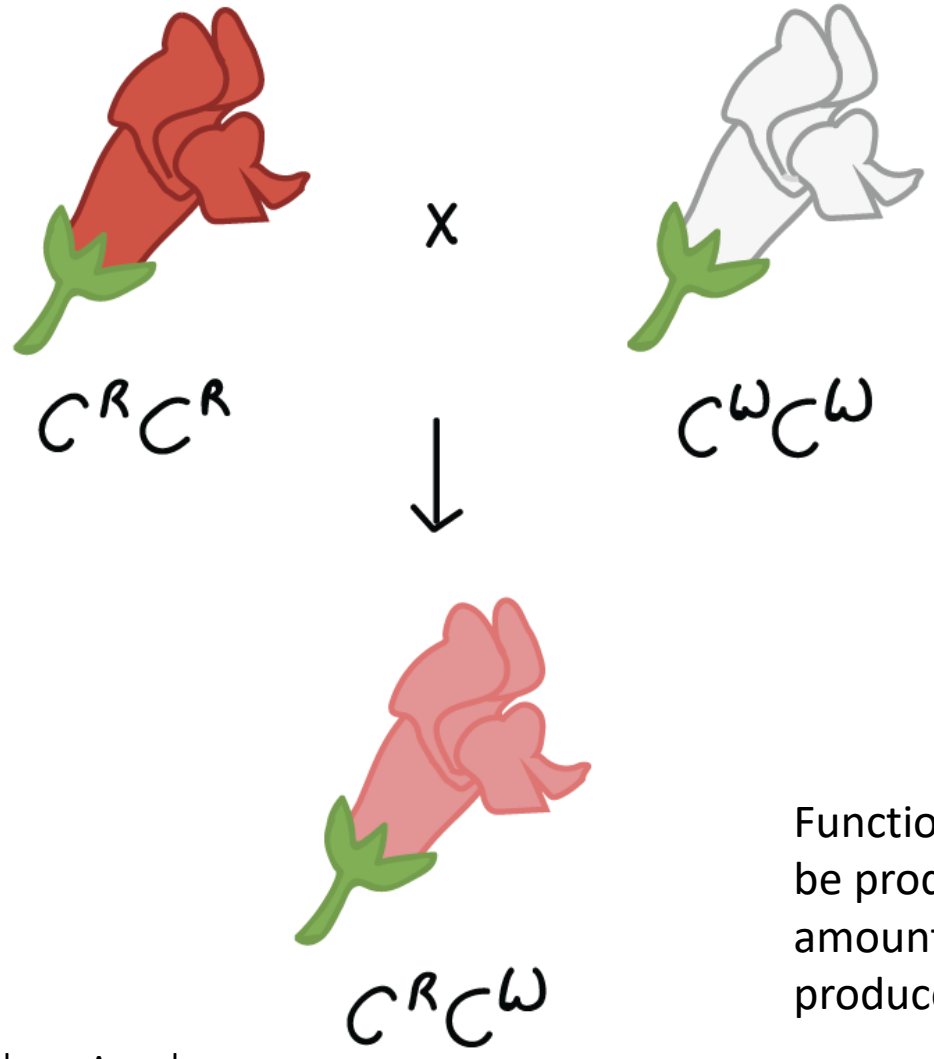


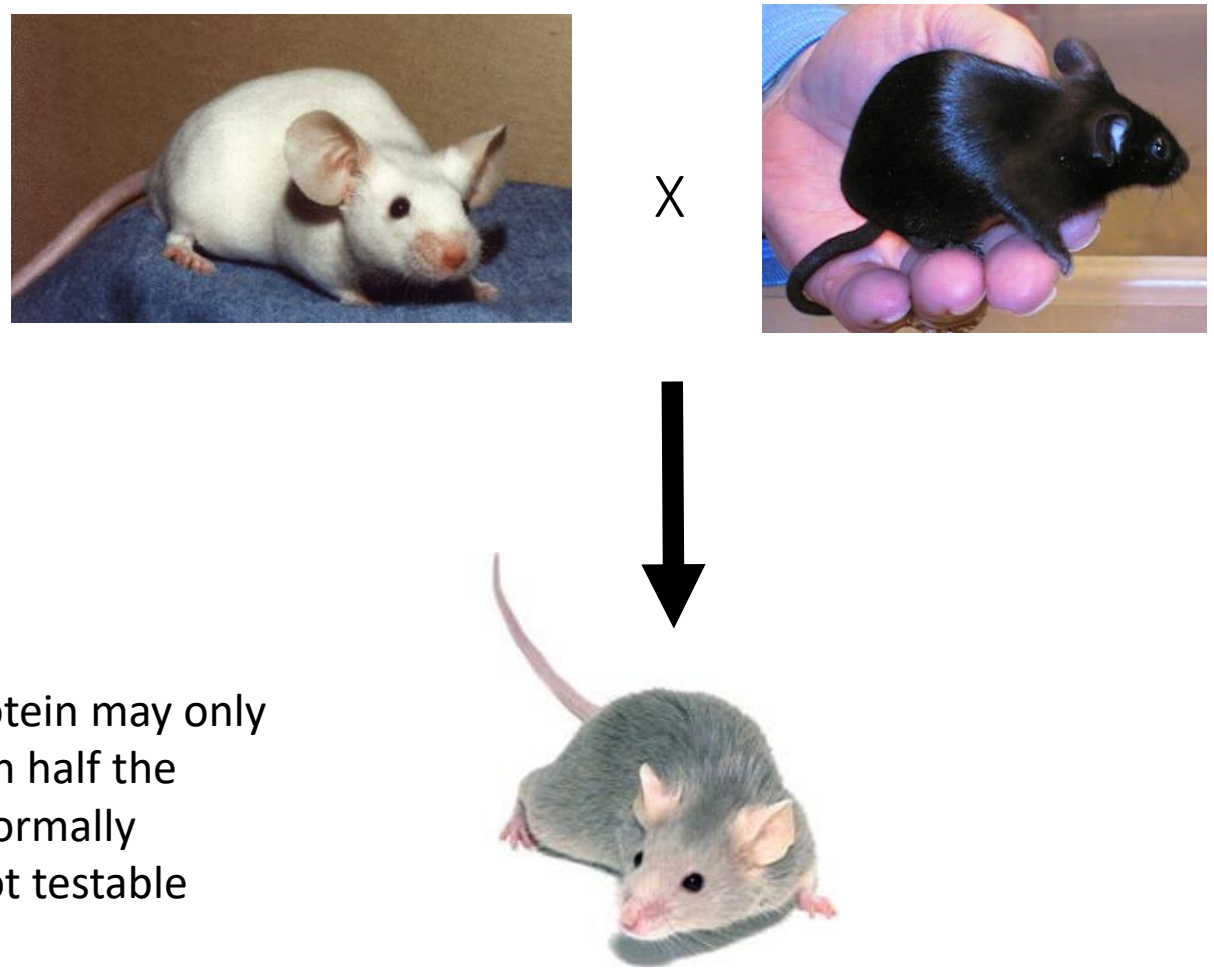


Different cells have different phenotypes – not testable

Sources: iBiologia, BiologyWise

Examples of **incomplete dominance**



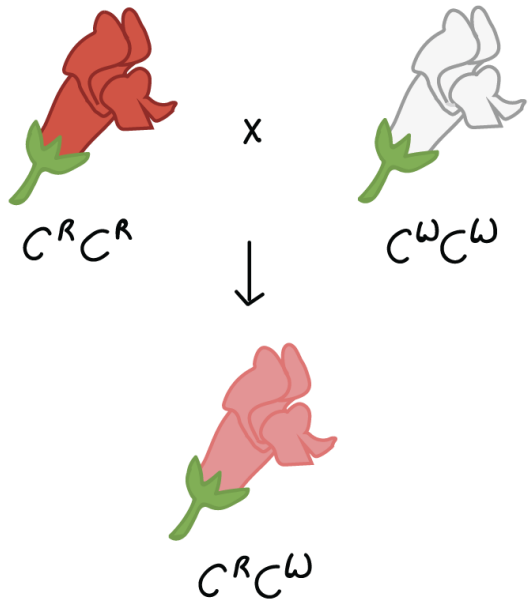


Functional protein may only be produced in half the amount it is normally produced – not testable

Source: Khan Academy

Non-dominance - Patterns

If two alleles have a non-dominant relationship, and you cross two homozygous parents with different genotypes, the offspring have a different (3rd) phenotype.



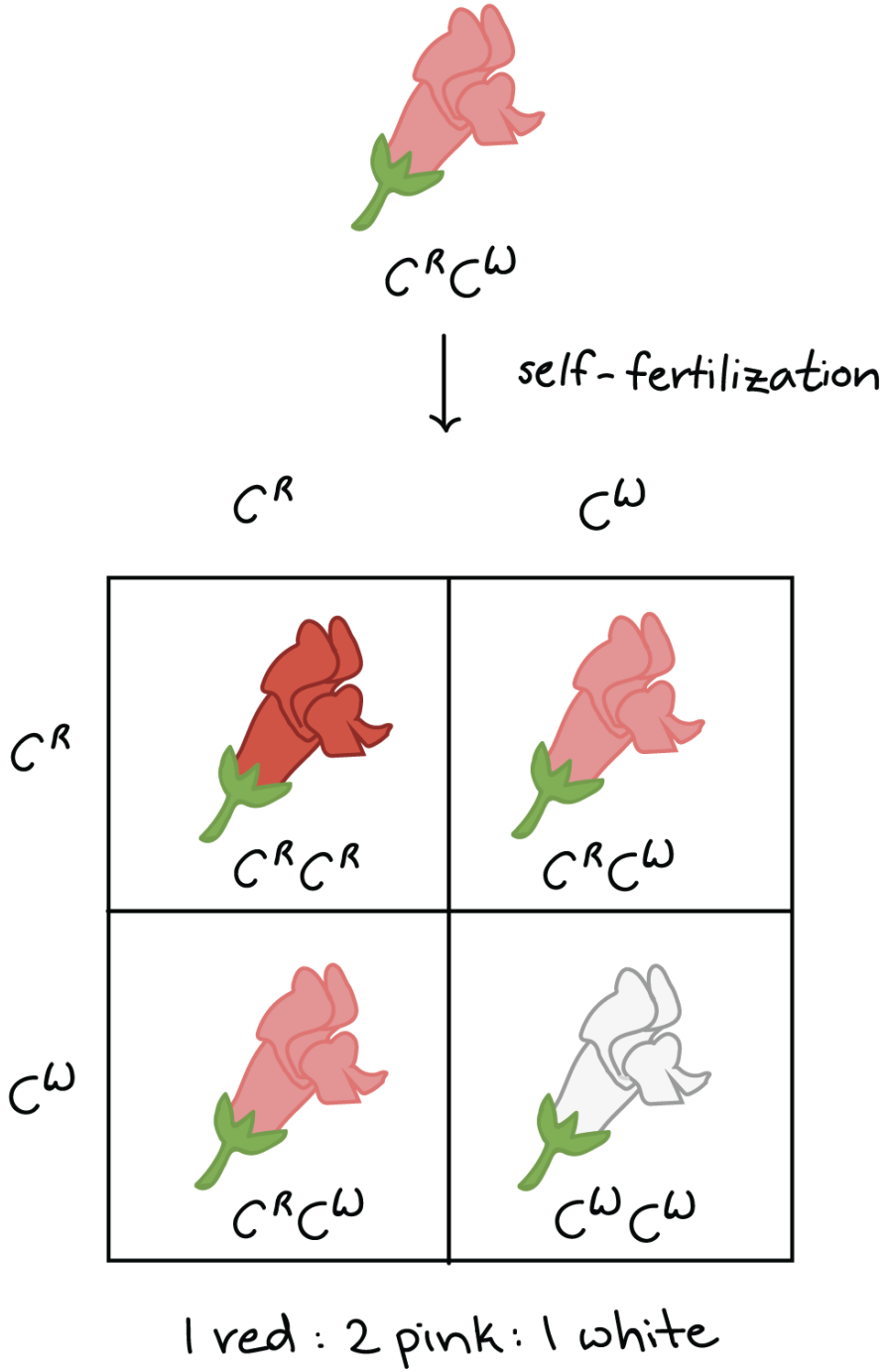
Source: Khan Academy

Non-dominance

If you cross 2 heterozygotes, and the alleles have a non-dominant relationship:

Creates **3 phenotypes** with a **1:2:1** phenotypic ratio

Have we seen a 1:2:1 phenotypic ratio before?



Source: Khan Academy

Question: Patterns of non-dominant inheritance

Are the horse parents likely to be homozygotes or heterozygotes?

A. Homozygotes
B. Heterozygotes
C. Insufficient information
D. Not sure

P_1

×

F_1

Answer

	A1	A2
A1	A1A1	A1A2
A2	A1A2	A2A2

Are the horse parents likely to be homozygotes or heterozygotes?

A. Homozygotes

B. Heterozygotes

C. Insufficient information

D. Not sure

P_1

×

F_1

<u>Key: 3 phenotypes in offspring</u>.

If both parents were homozygotes (e.g. A1A1, A2A2, how many phenotypes would you expect in the offspring)?

Question

If a litter of kittens resulting from the mating of two short-tailed cats contains 3 kittens without tails, two kittens with long tails and six kittens with short tails, what would be the simplest way of explaining the inheritance of tail length in these cats? Indicate genotypes in your answer.

Feel free to discuss

Answer

	T1	T2
T1	T1T1	T1T2
T2	T1T2	T2T2

If a litter of kittens resulting from the mating of two short-tailed cats contains 3 kittens without tails, 2 kittens with long tails and 6 kittens with short tails, what would be the simplest way of explaining the inheritance of tail length in these cats?

Two alleles (e.g. T1 and T2) with a non-dominant relationship

3 different genotypes: no-tail, short-tail, long-tail

T1T1 T1T2 T2T2

3 different phenotypes: ~ 1 : 2 : 1

The key here is that 3 different phenotypes were present for 2 alleles in a 1:2:1 ratio.

Worksheet #6 (now optional) – Inheritance of eye and fur colour in rats

1 . As part of your grad studies, you are investigating the inheritance of fur colour (blue vs. golden) and eye colour (green, red or yellow) in a rare species of rat. The table below shows the results of several different genetic crosses between male and females . Assume that these traits are influenced by two different genes, and that for each gene there exist two alleles.

Table 1.

Cross #	Parents (P)	Offspring (F1)
1	Green eyes, golden fur x red eyes, blue fur	34 yellow eyes, golden fur
2	Red eyes, golden fur x red eyes, blue fur	15 red eyes, golden fur 16 red eyes, blue fur
3	Green eyes, golden fur x green eyes, golden fur	28 green eyes, golden fur 9 green eyes, blue fur
4	Red eyes, blue fur x yellow eyes, blue fur	18 yellow eyes, blue fur 16 red eyes, blue fur

Learning goals - summary

- Determine whether a gene is on an autosome or a sex chromosome (X only)
- Determine whether two alleles have a dominant/recessive relationship or a non-dominant relationship
- Predict what gamete genotypes a parent can produce and in what proportions - for genes that are unlinked or linked, or on an autosome or sex chromosome
- Predict offspring genotypes and phenotypes and in what proportions – for genes that are unlinked or linked, on an autosome or sex chromosome, and whether alleles have a dominant/recessive relationship or a non-dominant relationship
- Be able to calculate the probability that two parents will have an offspring with a specific phenotype (one and two traits)

Genetic Crosses & Pedigrees

- Biologists can use their knowledge about parental genotypes and/or phenotypes & offspring genotypic and phenotype ratios to determine the most likely mode of inheritance for a trait.
- This requires doing genetic crosses, i.e., mating numerous individuals and looking at the phenotypes of the offspring to establish a hypothesis.
- Remainder of today's class – how to use genetic crosses to determine mode of inheritance.
- Thursday's class – Using pedigrees to determine mode of inheritance in humans.
- Both very testable

Genetic Crosses - Learning goals

When provided with information about the outcome of genetic crosses, be able to:

- make inferences* regarding the inheritance of the trait/phenotype, i.e.:
    - Autosomal dominant
    - Autosomal recessive
    - X-linked Dominant
    - X-linked Recessive
    - Non-dominant (autosomal genes only)

*we could ask you to identify the most likely mode of inheritance OR we could suggest a mode of inheritance and ask you to assess the claim.

- be able to use evidence (quantified, if possible) and reasoning/logic to support your claim about the mode of inheritance.

If asked to determine most likely mode of inheritance for a trait - my approach: 6 steps

*ALWAYS look at each trait separately (because the different traits may have different modes of inheritance = potentially messy data)

- Step #1 – Carefully read the scenario and scan the data*

  - looking for key information, e.g.

    Are parents true-breeding parents or not (scenario)?

    Do you notice any patterns in the F1 and F2 generation?

    - Do males and females have different phenotypes in the F1 or F2 generation, suggesting X-linked trait?
    - What are the phenotypic ratios, e.g. 3:1? 1:1? 1:2:1?

My recommended approach

- Step 2: After scanning data for one trait, you should have a hunch or a starting hypothesis for mode of inheritance.

- Step 3: Define your genes/alleles/genotypes based on this hypothesis.

- Step 4: Use a Punnett Square to make PREDICTIONS about the EXPECTED genotype/phenotype frequencies for the F1 and F2 generation, <u>if that hypothesis is true</u>

- Step 5: Compare PREDICTED frequencies with OBSERVED frequencies – are they similar or not? QUANTIFY. Yes - go to Step 6. No – return to step 1.

- Step 6: Conclusion - explicitly state whether your results support your hypothesis about the mode of inheritance for the trait.

Dragon example – scale colour

True breeding female dragons with red scales and long horns were crossed with true breeding male dragons with green scales and short horns. The results for scale colour were as follows:

Cross 1: Females, red scales x Males, green scales

F1s: All had red scales

Hmmm – F1 females (at least) carrying an allele for both red scale colour and green scale colour. What does this suggest about relationship between alleles?

Cross 2: F1 females from cross 1 x F1 males from cross 1

F2s:

	Red	Green
Males	39	11
Females	41	16

Hmmm – a 3:1 phenotypic ratio in F2 generation for both sexes. Where have we seen that ratio before?

Step 2 – hypothesis for mode of inheritance: scale colour

Question – What is your hypothesis about the mode of inheritance for scale colour.

A. Autosomal, with red scales dominant to green scales
B. Autosomal, with green scales dominant to red scales
C. X-linked, with red scales dominant to green scales
D. X-linked, with green scales dominant to red scales
E. Non dominant

Cross 1: Females, red scales x Males, green scales

F1s: All had red scales

Cross 2: F1 females from cross 1 x F1 males from cross 1

F2s (see table)

	Red	Green
Males	39	11
Females	41	16

Step 2 – hypothesis for mode of inheritance: scale colour

Poll question – What is your hypothesis about the mode of inheritance for scale colour.

A. Autosomal, with red scales dominant to green scales
B. Autosomal, with green scales dominant to red scales
C. X-linked, with red scales dominant to green scales
D. X-linked, with green scales dominant to red scales
E. Non dominant

F1 males and females all have the same phenotype (red scales).

Females are heterozygotes, and have red scales, indicating red>green.

Autosomal or X-linked? See F2

F2: 3:1 phenotypic ratio (red:green) (ratio that Mendel observed when gene was autosomal, dominant/recessive relationship)

Cross 1: Females, red scales x Males, green scales

F1s: All had red scales

Cross 2: F1 females from cross 1 x F1 males from cross 1

F2s (see table)

	Red	Green
Males	39	11
Females	41	16

Step 3 – define genes, alleles, genotypes

- Scale Colour: R

R= red
r =green

RR = red

Rr = red

rr = green

For the future (e.g., if you take a genetics course, and are writing by hand) avoid using letters that are similar in both capital and lower case, e.g. c, s, o…..

For X-linked – remember biological sex is part of a phenotype.

Step 4 – Calculate PREDICTED phenotype frequencies if hypothesis is true

Trait #1 – Scale colour: Autosomal, red scales dominant to green scales (R > r)

Cross parents: True breeding females, red scales (RR) x True breeding males, green scales (rr)

	r
R	Rr

F1s: Predicted genotype: Rr
Predicted phenotype: Red scales (all F1 dragons)

Cross F1s: F1 females (Rr) x F1 males (Rr)

	R	r
R	RR	Rr
r	Rr	rr

F2s: Predicted phenotype ratio: 3:1 phenotypic ratio, red to green scales

Step 5. Compare observed and predicted frequencies for hypothesis – scale colour

Cross	Generation	Predicted Phenotype freq.	Observed Phenotype freq.
RR x rr (P)	F1	Rr – all red scales	All red scales – both sexes
Rr x Rr (F1)	F2	RR, Rr, Rr, rr 3:1 phenotypic ratio – red scales:green scales	80:27 = 3:1 (red to green) Males: 39:11 = 3:1 (red to green) Females: 41:16 = 3:1 (red to green)

Predicted F2s (overall, not broken down by sex:

	R	r
R	RR	Rr
r	Rr	rr

Observed F2s (by sex)

	Red	Green
Males	39	11
Females	41	16

Step 5 – Continued (described in words - QUANTIFY)

If the mode of inheritance for scale colour was autosomal with red dominant to green, then we would predict that:

A cross between two pure breeding dragons (RR x rr) in the parental generation would produce a heterozygous F1 generation (Rr) that all had red scales, which is what we observed (can't quantify).

We would also predict that a cross between two F1 heterozygotes (Rr) would result in a 3:1 phenotypic ratio of red scales to green scales, which is what was observed. In the F2 generation, there were 80 offspring with red scales (39 males and 41 females) and 27 offspring had green scales (11 males and 16 females), which is approximately the 3:1 predicted ratio.

Important: genotypes specified, phenotypes quantified (if possible) for both generations, and an explicit comparison of expected (predicted) and observed results was included.

Step 6: Concluding statement (reasoning) - do data support hypothesis?

So… the observed phenotypic ratios for scale colour for both the F1 and F2 generation are consistent with the predicted results, which supports the hypothesis that scale colour in dragons is an autosomal trait, with red scales dominant to green scales.

We could also give you a hypothesis and ask you to assess that hypothesis.

…..as opposed to propose a hypothesis for mode of inheritance..

- In this case, do NOT try to prove what the mode of inheritance is (that is not the question).
- Rather come up with predicted phenotype ratios if the proposed hypothesis is true and compared to observed phenotypic frequencies.

So, you can skip steps 1 & 2, i.e. scan data to come up with a hunch about mode of inheritance. Start at step 3.

Dragon example – horn length

True breeding female dragons with long horns were crossed with true breeding male dragons with short horns. The results were as follows:

Cross 1: Females, long horns x Males, short horns

F1s: All (100%) had long horns

Cross 2: F1 females from cross 1 x F1 males from cross 1

F2s:

	Short	Long
Males	26	24
Females	0	47

Use the information above to **assess the hypothesis** that short horns in dragons is an example of an X-linked dominant trait.

Step 3A – Define genes, alleles, genotypes based on hypothesis that short horns are an X-linked dominant trait.

Horn Length: X^H = short horn and X^h = long horn

Step 3B – Define all possible genotypes and associated phenotypes that can be produced based on the proposed hypothesis

X^HX^H = female with short horns
X^HX^h = female with short horns
X^hX^h = female with long horns
X^HY = males with short horns
X^hY = males with long horns

Remember – an individual's sex is part of their phenotype

Step 4 – Predicted F1 & F2 phenotype frequencies if hypothesis is correct (short horn length = X-linked dominant phenotype)

Cross 1: Females -long horns X^hX^h x Males – short horns X^HY

	X^h
X^H	X^HX^h
Y	X^hY

F1 prediction:
-All F1 females – hybrids - have short horns
-All F1 males have long horns

Cross 2: F1 females from cross 1 x F1 males from cross 1

F2s:

	X^H	X^h
X^h	X^HX^h	X^hX^h
Y	X^HY	X^hY

F2 prediction:
- 1:1 ratio of females with short:long horns
- 1:1 ratio of males with short:long horns.

Step 5. Compare observed and predicted frequencies –horn length. Do they support claim about mode of inheritance?

Cross 1: Females, long horns x Males, short horns

F1s: All (100%) had long horns

Cross 2: F1 females from cross 1 x F1 males from cross 1

F2s:

	Short	Long
Males	26	24
Females	0	47

Cross	To create:	Predicted Phenotype freq.	Observed Phenotype freq.	Support?
X^hX^h x X^HY (P)	F1	X^HX^h - females all short horns X^hY – males all long horns	All long horns – both sexes	No
X^HX^h x X^hY (F1)	F2	X^HX^h, X^hX^h – females 1:1 phenotypic ratio short:long horns X^HY, X^hY – males 1:1 phenotypic ratio short:long horns	All females (47)- long horns; Males 26:24 or 1:1	No

Compare observed and predicted frequencies –horn length.

Cross	To create:	Predicted Phenotype freq.	Observed Phenotype freq.	Support?
X^hX^h x X^HY (P)	F1	X^HX^h - females all short horns X^hY – males all long horns	All long horns – both sexes	No
X^HX^h x X^hY (F1)	F2	X^HX^h, X^hX^h – females 1:1 phenotypic ratio short:long horns X^HY, X^hY – males 1:1 phenotypic ratio short:long horns	All females (47)- long horns; Males 26:24 or 1:1	No

No, the data does not support the hypothesis that short horns is an X-linked dominant trait.

If short horns was an X-linked dominant trait, we predicted that:

- all F1 females would have short horns because, if the hypothesis was true, they would inherit the dominant allele for short horns from their father (X^HY); however, all F1 females (and males) had long horns, suggesting that, in fact, long horns is dominant to short horns.

If the hypothesis that short horns is an X-linked dominant trait was correct, we also predicted that:

- there would be a 1:1 ratio of short horns:long horns for F2 females and F2 males because the F1 females would donate the dominant allele for short horns to 50% of her offspring. Although we observed this ratio in the F2 males (24:26 short:long), all F2 females (47) had long horns.

Note – unless an exam question specifies that you need to use all available evidence, you could just refer to the F1 data as evidence that short horns in dragons is not an X-lilnked dominant trait.

Step 6: Concluding statement - do data support hypothesis?

- The observed F1 and F2 phenotypic results do NOT match the predicted phenotypic results for short horns being an X-linked dominant trait. Therefore, short horns in dragons is not an X-linked dominant trait.

Worksheet #5 - *Drosophila*

Wild-type *Drosophila melanogaster* have dark red eyes and smooth bristles (these are the "hairs" that they have on their back). Mutant *D. melanogaster* have *scarlet* (bright red) eyes and *singed* bristles, which look like they have been burned.

A geneticist performed a cross between pure-breeding females with dark red eyes and *singed* bristled and pure-breeding males with *scarlet eyes* and smooth bristles. The results were as follows:

Cross 1: female, dark-red eyes, *singed* bristles **X** male, *scarlet* eyes, smooth bristles

Progeny (F1): 82 females, dark red colour and smooth bristles (one is used for cross 2)
79 males, dark red colour and *singed* bristles (one is used for cross 2)

Cross 2: F1 female from cross 1 **X** F1 male from cross 1

Progeny (F2):

	Dark red eyes, smooth bristles	Dark red eyes, *singed* bristles	*scarlet* eyes, smooth bristles	*scarlet* eyes, *singed* bristles
Females	29	31	10	8
Males	27	30	9	11

(a) Based on these data, what is the mode of inheritance of the phenotypes *scarlet* eyes and *singed* bristles, respectively? Show your work/steps.

Next class

- Using pedigree to determine mode of inheritance

The slides that follow are some extra crosses questions for practice (with answers)

Optional at home practice question #1 – chipmunks

- Parents: a female chipmunk with erect ears and a straight tail is crossed with a male chipmunk with floppy ears and a bent tail.
- F1s: all the F1 chipmunks have erect ears and straight tails.
- F2s: the F1 chipmunks are crossed and the following F2 data are obtained:

  61 female chipmunks with erect ears and straight tails

  19 female chipmunks with floppy ears and straight tails

  30 male chipmunks with erect ears and straight tails

  31 male chipmunks with erect ears and bent tails

  9 male chipmunks with floppy ears and straight tails

  11 male chipmunks with floppy ears and bent tails

sometimes the data do not come in a table

How are the traits of erect and floppy ears and straight and bent tails inherited? Show all of your work.

Summarize chipmunk data

P ♀ - erect ears &straight tail x ♂ floppy ears & bent tail

F1 All chipmunks have erect ears and straight tails.

F2

	Erect Ears, Straight Tails	Floppy Ears, Straight Tails	Erect Ears, Bent Tails	Floppy Ears, Bent Tail
Females	61	19	0	0
Males	30	9	31	11

What is the mode of inheritance for ear shape?

Step 1: Scan data

Step 2: Hypothesis for mode of inheritance – one trait (recommend start with ears – organization of my slides)

Step 3: Define genes/alleles/genotypes for that trait.

Step 4: Use a Punnett Square to make predictions about the expected genotype/phenotype frequencies for the F1 and F2 generation, if that hypothesis is true

Step 5: Compare predicted frequencies with observed frequencies – Do they match or not. Do they support the hypothesis.

Repeat for 2nd trait

Step 6: Conclusion - explicitly State whether your results support your hypothesis about the mode of inheritance for the trait.

Step 1 – Scan data for ear shape

P ♀ - erect ears x ♂ floppy ears

F1 All chipmunks have erect ears

F2

	Erect Ears, Straight Tails	Floppy Ears, Straight Tails	Erect Ears, Bent Tails	Floppy Ears, Bent Tail
Females	61	19	0	0
Males	30	9	31	11

Patterns?

source: Canadian Geographic

Step 2: Hypothesis about ear shape

Poll Question:

A. Autosomal – erect ears dominant to floppy ears
B. Autosomal – floppy ears dominant to erect ears
C. X-linked – erect ears dominant to floppy ears
D. X-linked – floppy ears dominant to erect ears
E. Uncertain.

Step 2: Hypothesis about ear shape

Poll Question:

A. Autosomal – erect ears dominant to floppy ears
B. Autosomal – floppy ears dominant to erect ears
C. X-linked – erect ears dominant to floppy ears
D. X-linked – floppy ears dominant to erect ears
E. Uncertain.

Step 3 - Define genes/alleles/genotypes

- E = ear shape
- E (or E1) = erect ears
- e (or E2) = floppy ears
- EE = erect ears
- Ee = erect ears
- ee = droopy ears

Step 4 – Predicted frequencies – ear shape

- Parents EE x ee
- F1 – all heterozygotes (Erect ears)

	E
e	Ee

- F2 - 3:1 phenotypic ratio – Erect ears:Floppy ears

	E	e
E	EE	Ee
e	Ee	ee

Step 5. Compare observed and predicted frequencies – ear shape

Cross	Generation	Predicted Phenotype freq.	Observed Phenotype freq.
EE x ee (P)	F1	Ee – all erect ears	All erect ears – both sexes (can't quantify)
Ee x Ee (F1)	F2	3:1 – erect ears:floppy ears	122:39 = ~3:1 (erect to floppy) Females: 61:19 = 3:1 Males: 61:20 = 3:1

F2s:

	Erect Ears	Floppy Ears
Females	61	19
Males	30 + 31 = 61	9 + 11 = 20

Do the observed data match the predicted data for ear shape?

Step 6 – Conclusion about mode of inheritance for ear shape

- The observed phenotypic ratios for ear shape are consistent with the predicted phenotypic ratios, supporting the hypothesis that ear shape is autosomal with erect ears dominant to floppy ears.

Tail shape

P ♀ - erect ears & straight tail x ♂ floppy ears & bent tail

F1 All chipmunks have erect ears and straight tails.

F2

	Erect Ears, Straight Tails	Floppy Ears, Straight Tails	Erect Ears, Bent Tails	Floppy Ears, Bent Tail
Females	61	19	0	0
Males	30	9	31	11

Use the information above to assess the hypothesis straight tails is an autosomal dominant trait?

Step 3: Define gene, alleles and genotypes

- A = straight tails
- a = bent tails
- AA = female or male with a straight tail
- Aa = female or male with a straight tail
- Aa = female or male with a bent tail

Step 4 – predicted frequencies (based on proposed hypothesis) – tail shape

- Parents AA x aa
- F1 – predict all F1s are heterozygotes with straight tails

	A
a	Aa

- F2 - predict a 3:1 phenotypic ratio of straight tails to bent tails for both males and females.

	A	a
A	AA	Aa
a	Aa	aa

Step 5 – compare predicted and observed

Cross	To create generation	Predicted	Observed	Support hypothesis?
AA x aa (P)	F1	Aa – all females and males have straight tails	All F1 males and females have straight tails (can't quantify)	
Aa x Aa (F1)	F2	AA, Aa, Aa, aa 3:1 phenotypic ratio of straight tails to bent tails for both males and females	Females: 80 straight tails, 0 bent tails Males: 39 straight tails, 42 bent tails (~1:1 ratio).	No

Observed F2 phenotypic ratios are not consistent with predicted ratios if straight tails is an autosomal dominant trait. If the hypothesis was true, we expected to see a 3:1 phenotypic ratio of straight tails to bent tails for both males and females. However, in the F2 generation all female mice (n=80) had straight tails and there was approximately a 1:1 ratio of straight tails (n=39) to bent tails (=42) in the F2 males.

Step 6 – Conclusion about hypothesized mode of inheritance

- The observed data not support the hypothesis that straight tails are an autosomal dominant mode of inheritance in mice.

Optional at home practice question #2 - hamsters

A friend has two pet hamsters, a male and a female, that are kept in the same cage. The male hamster has medium length white fur that is approximately 5 cm in length. The female hamster has medium length golden fur that it is also approximately 5 cm in length. One February day, the female hamster gave birth to 18 pups. The phenotypes of her offspring are as follows:

Short fur (< 2cm)	Medium length fur (~5 cm)	Long fur (10 cm)
2 females golden fur 2 males golden fur	5 females golden fur 4 males golden fur	3 females golden fur 2 males golden fur

What is the likely mode of inheritance for fur length in hamsters?

Hamster data

P ♀ - white, medium length fur x ♂ golden medium length fur

F1

Short fur (< 2cm)	Medium length fur (~5 cm)	Long fur (10 cm)
2 females golden fur 2 males golden fur	5 females golden fur 4 males golden fur	3 females golden fur 2 males golden fur

What is the most likely mode of inheritance for fur length in hamsters?

Step 1: Scan data

Step 2: Hypothesis for mode of inheritance

Step 3: Define genes/alleles/genotypes for that trait.

Step 4: Use a Punnett Square to make predictions about the expected genotype/phenotype frequencies for the F1 generation, if that hypothesis is true

Step 5: Compare predicted frequencies with observed frequencies.

Step 6: Conclusion - explicitly state whether your results support your hypothesis about the mode of inheritance for the trait.

ANSWERS ON THE NEXT SLIDES

Step 1 & 2

3 phenotypes (short, medium and long fur)
1:2:1 phenotypic ratio in F1s

Short fur (< 2cm)	Medium length fur (~5 cm)	Long fur (10 cm)
2 females golden fur 2 males golden fur	5 females golden fur 4 males golden fur	3 females golden fur 2 males golden fur

1:2:1 Phenotypic ratio is the pattern that is observed when two heterozygotes are crossed and the alleles have a non-dominant relationship.
So, likely mode of inheritance for fur length is autosomal, non-dominance

Step 3 – define genes, alleles, genotypes

- F = fur length
- F1 = short fur
- F2 = long fur
- F1F1 = short fur (2 cm)
- F2F2 = long fur (10 cm)
- F1F2 = medium length fur (5 cm)

Other notion is possible, e.g. F^S, F^L – but do not use upper/lower case for notation, e.g. F, f as that implies a dominant/recessive relationship

Step 4. Predicted phenotypic ratios if hypothesis is true

- Male and female guinea pigs are heterozygotes (F1F2)

	F1	F2
F1	F1F1 (short)	F1F2 (medium)
F2	F1F2 (medium)	F2F2 (long)

Predict a 1:2:1 phenotypic ratio of short:medium:long furred hamsters

Step 5 - Compare predicted and observed frequencies

- Male and female guinea pigs are heterozygotes (F1F2).
- Predict: 1:2:1 phenotypic ratio - short(1):medium(2):long(1)

	F1	F2
F1	F1F1 (short)	F1F2 (medium)
F2	F1F2 (medium)	F2F2 (long)

- Observed: 1:2:1 phenotypic ratio – short(4):medium(9):long(5)

Short fur (< 2cm)	Medium length fur (~5 cm)	Long fur (10 cm)
2 females golden fur 2 males golden fur	5 females golden fur 4 males golden fur	3 females golden fur 2 males golden fur

Step 6 - conclusion

- Observed phenotypic ratios match the predicted phenotypic ratios with 3 phenotypes in a 1:2:1 ratio, which supports the hypothesis of fur length having an autosomal non-dominance mode of inheritance.

- Can you be certain about the mode of inheritance for fur colour from this data?