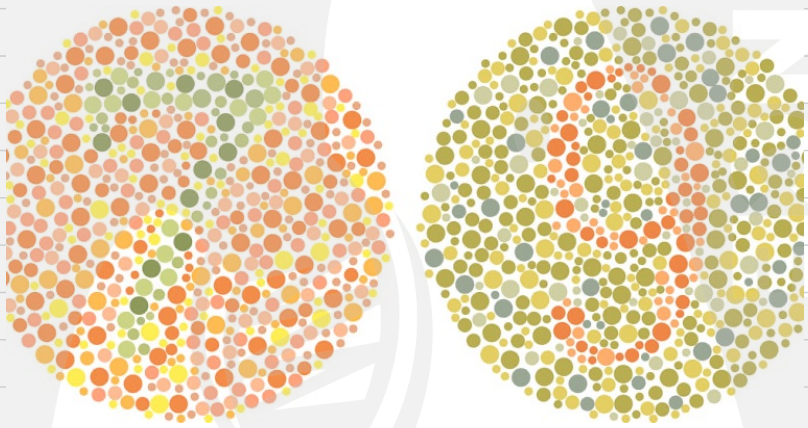
Inheritance

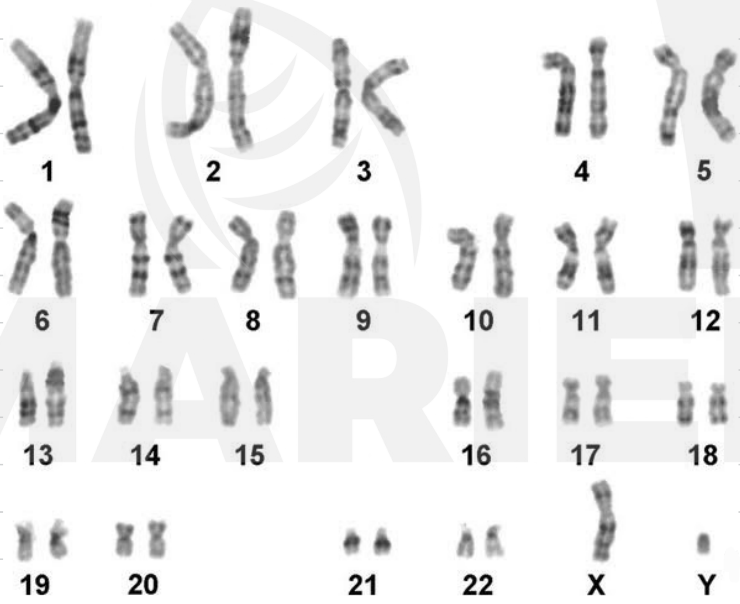
Learning outcomes

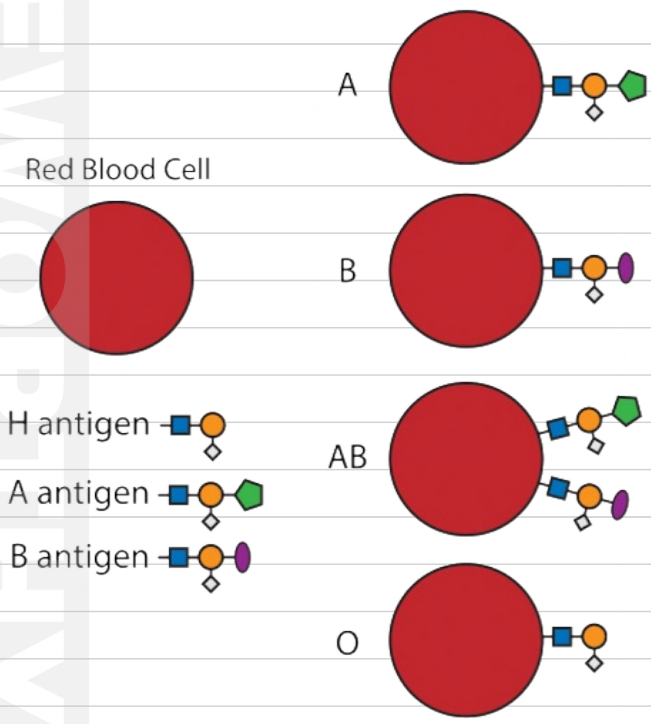
- [x] Understand how Mendel's pea plant experiments lead to his development of his laws of inheritance
- [x] Understand the 3 laws of inheritance
- [x] Deduce offspring genotype and phenotype probabilities from monohybrid crosses using Punnett squares
- [x] Understand the use of test-crosses in deducing unknown genotypes
- [x] Understand and deduce the inheritance, phenotypes, and genotypes of co-dominant traits
- [x] Understand and deduce the inheritance, phenotypes, and genotypes of sex-linked traits
- [x] Read, draw and analyze pedigrees to deduce type of inheritance and genotypes

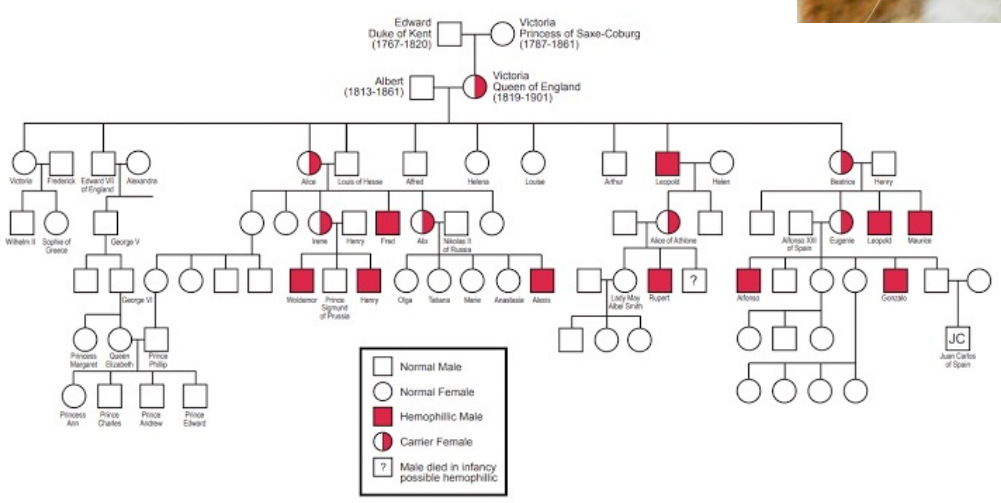
Key Terms

- phenotype
- true breeding line
- diploid
  homologous chromosome
- genotype
- homozygous
- heterozygous
- autosomes
  sex chromosome
- haploid
- Punnett square
- Co-dominance
- Sex-linkage
- carrier
- pedigree









Mendelian Genetics

Gregor Mendel was interested in gardening, particularly pea plants. He observed that a pea plant had several distinct characteristics where for each, they only had one trait or another, never a combination of both.

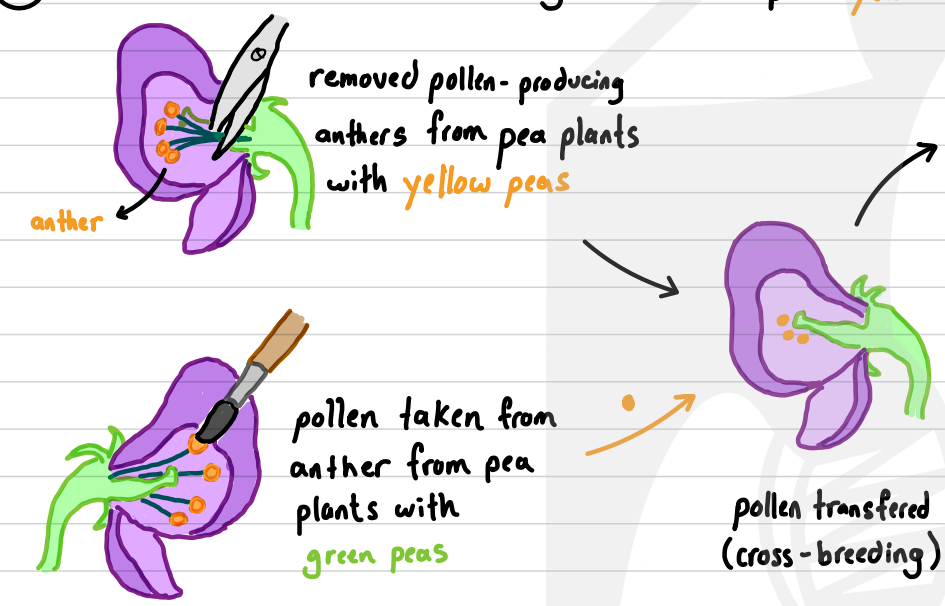
Mendel observed several **phenotypes**: expressed/observable characteristic

flower colour	flower position	seed colour	seed shape	pod shape	pod colour	stem length
purple	terminal	yellow	round	inflated	green	tall
white	axial	green	wrinkled	constricted	yellow	short

Cross-breeding experiments - for each trait

① Created **true breeding lines**: individuals when crossed only produce offspring that express one trait

② Crossed the two true breeding lines - example: yellow peas x green peas



Result - Gen 1

all of offspring produced yellow peas

③ offspring were cross-bred

Result - Gen 2

~75% of offspring were yellow peas

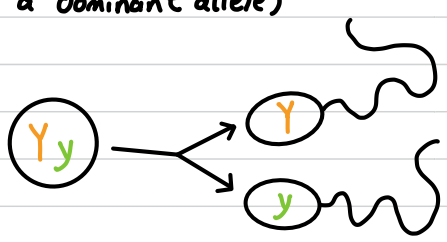
~25% of offspring were green peas

HOW?

Mendel's conclusions - Laws of Inheritance

① **Law of Dominance** - In a heterozygote, a dominant allele will mask the expression of a recessive allele
- ex: yellow pea (Y) is dominant to green pea (y) → Yy only yellow expressed (green is masked)
- the only way to express a recessive allele is as a homozygote (absence of a dominant allele)
- ex: homozygous recessive (yy) - green peas

② **Law of Segregation** - a diploid individual has 1 pair of alleles for each gene
- when gametes are formed, this pair is randomly separated into 2 gametes
- each gamete are **haploid**: contain 1 copy of each chromosome
- which allele is passed to offspring is random

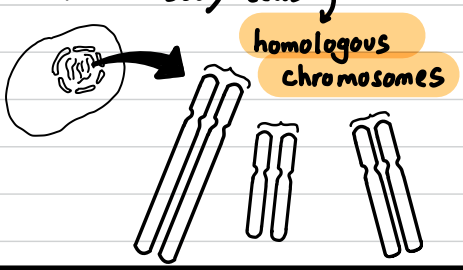


∴ 1 copy of each gene (1 allele)

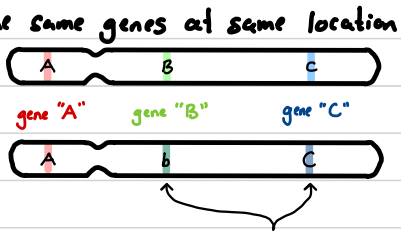
③ **Law of Independant assortment** - separate genes for separate traits are passed independently of one another from parent to offspring
- every possible combination of alleles for every gene is equally likely to occur
- ex: yellow peas + purple flowers and green peas + purple flowers can equally occur

Key concepts

Many eukaryotes are **diploid**: two copies of each chromosome in each of their body cells



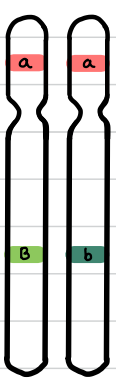
homologous chromosomes are the same size, contain the same genes at same location



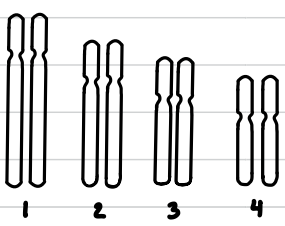
BUT they may have different versions of the gene (allele)

Genotype: combination of alleles

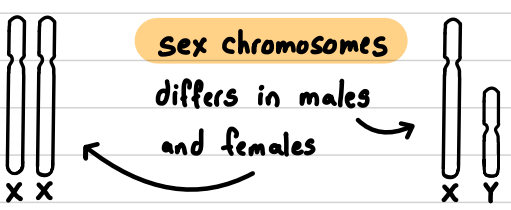
- **Homozygous**: identical alleles for a gene
- **Heterozygous**: different alleles for a gene



autosomes
- numbered chromosomes
- same for male and female
- humans have 22 pairs



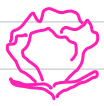
sex chromosomes
- differs in males and females



Punnett Squares

Punnett square: (named after geneticist Reginald C. Punnett) is a diagram used to predict genotypes of a cross

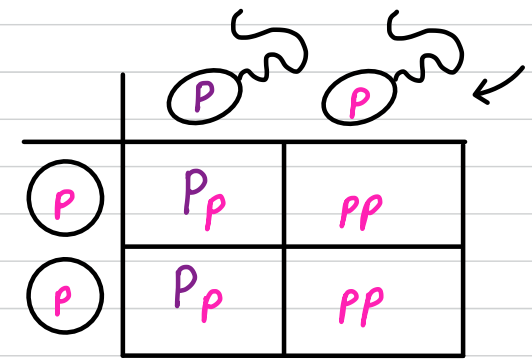
purple is - P dominant



white is - p recessive

Heterozygous purple pea plant (Pp)
x
white pea plant (pp)

monohybrid cross (crossing 1 gene)



represents 25% chance of this particular genotype

50% chance of Purple flowers
50% chance of white flowers

1:1 ratio

How to predict the probability of genotypes

Example

In squirrels, grey is dominant and brown is recessive. A brown female is crossed with a homozygous grey male
Deduce the probability that their offspring will be brown.

① write the given information

male phenotype: grey
genotype: GG
female phenotype: brown
genotype: gg

② draw Punnett square with parent genotypes

	G	G
g		
g		

③ Drop down the alleles to each box

	G	G
g		
g		

④ Each box represents 25%

	G	G
g	Gg	Gg
g	Gg	Gg

- 100% chance of grey
- 0% chance of brown

phenotypic ratio Grey : brown = 1:0
genotypic ratio GG : Gg : gg = 0:1:0

Example

Brown eyes is dominant to blue eyes. A brown eyed man, whose mother has blue eyes marries a blue eyed woman.
Deduce the probability that their offspring will have blue eyes.

male phenotype: brown
genotype BB or Bb
female phenotype: blue
genotype bb

mother: bb
∴ must pass b allele to son
∴ Bb

	B	b
b	Bb	bb
b	Bb	bb

- 50% chance of brown
- 50% chance of blue

phenotypic ratio Brown : blue = 1:1
genotypic ratio BB : Bb : bb = 0:1:1

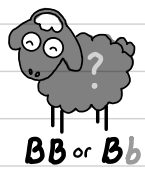
Can always deduce genotype of homozygous recessive individuals from the phenotype → bb
but not so with dominant phenotypes → either BB or Bb : which one???

Solution: Test crosses!

Example

Black wool is dominant to white in sheep. You have a black sheep but are unsure of their genotype.
Explain how you can deduce its genotype using crosses

① cross the unknown dominant genotype with homozygous recessive



BB or Bb × bb

will always be masked
∴ phenotype of offspring reflect other parent

if unknown is BB

	B	B
b	Bb	Bb
b	Bb	Bb

100% of offspring black

if unknown is Bb

	B	b
b	Bb	bb
b	Bb	bb

50% Black
50% white

② Repeat cross many times / examine large numbers of offspring

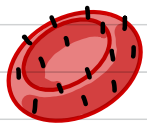
*note: probabilities are random independent events
50% black could still mean out of 4 offspring all are black

ex: coin flip is 50/50 but it's possible to flip 'heads' 5 times in a row

Co-Dominance & Sex-Linkage

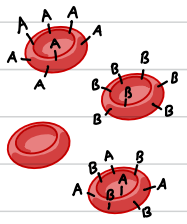
Co-dominance: when neither allele masks the expression of the other - BOTH are equally expressed in phenotype

↳ **ABO blood groups**

the ABO gene codes for blood cell antigen →  surface proteins

3 different alleles:
- I^A A allele
- I^B B allele
- i O allele

A ← allele
gene (lowercase for recessive)

Phenotype	RBC antigen	RBC antibody	Genotype
Blood type A		anti-B	I^AI^A or I^Ai
Blood type B		anti-A	I^BI^B or I^Bi
Blood type O		anti-B and anti-A	ii
Blood type AB		none	I^AI^B

A allele (I^A) is co-dominant with B allele (I^B)
O allele (i) is recessive to both A + B

Example

A man is type AB and his wife is heterozygous B. What is the phenotypic ratio of their potential offspring?

Man phenotype: AB
genotype: I^AI^B
woman phenotype: B
genotype: I^Bi

	I^A	I^B
I^B	I^AI^B	I^BI^B
i	I^Ai	I^Bi

25% type A
50% type B
25% type AB

phenotypic ratio A : B : O : AB = 1 : 2 : 0 : 1
genotypic ratio $I^Ai : I^BI^B : I^Bi : I^AI^B$ = 1 : 1 : 1 : 1

Example

Alice has blood type A blood and her husband has blood type B blood.
Their first child, Amanda, has type O blood. There second child has type AB blood.
Deduce the genotype of both Alice and her husband

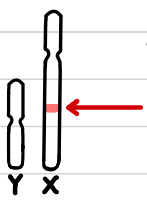
Alice phenotype: type A
genotype: I^AI^A or I^Ai
husband phenotype: type B
genotype: I^BI^B or I^Bi
Amanda phenotype: type O
genotype: ii

→ must recieve a recessive i allele from each parent
∴ both parents contain the i allele

	I^A	i
I^B	I^AI^B	I^Bi
i	I^Ai	ii ↑ Amanda

Alice: I^Ai
husband: I^Bi

Sex-linkage: gene found on sex chromosome (X or y) → pattern of inheritance is different for males and females



X-linked means gene found on X, not Y

females have homologous X chromosomes ∴ inheritance similar to autosomal traits
males only have 1 copy of X chromosome ∴ no masking - if allele present always expressed

↳ **Hemophilia**

gene for a blood clotting factor is defective and non-functional
∴ affected people's ability to clot blood is impaired
→ bleeding a lot after injury and easy bruising

2 alleles:
- X^H normal clotting factor - Dominant
- X^h defective clotting factor - recessive

Genotype: X^Hy X^hy X^HX^H X^hX^h X^HX^h → **carrier:** has the affected recessive allele but does not express it

Phenotype: normal male | hemophilia male | normal female | hemophilia female | normal female

Example

A hemophilic woman and normal man want to have children. What is the probability of:
a) having a child with hemophilia b) a girl with hemophilia c) a boy with hemophilia ?

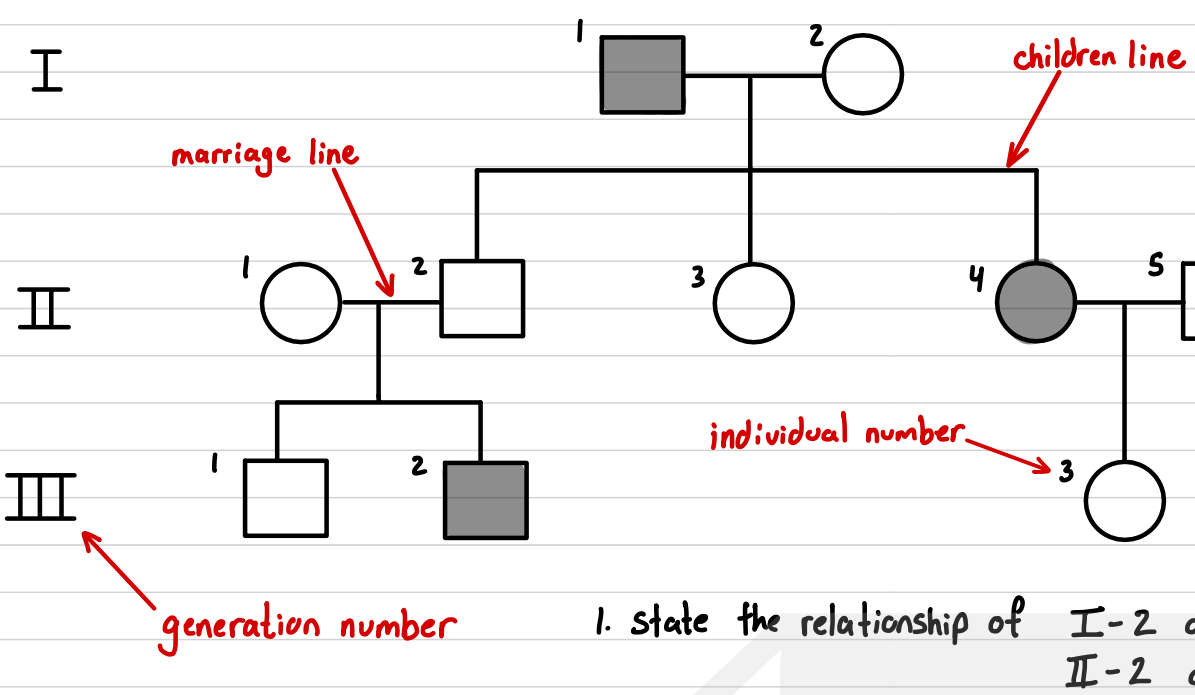
man phenotype: normal
genotype: X^Hy
woman phenotype: hemophilia
genotype: X^hX^h

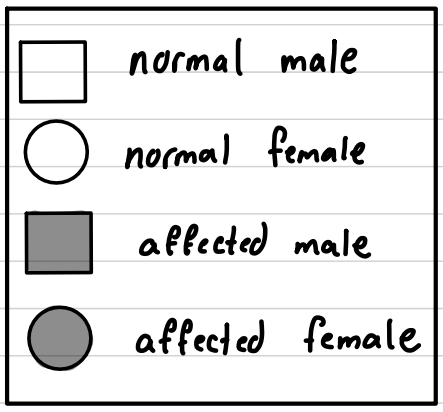
	X^H	y
X^h	X^hX^H	X^hy
X^h	X^hX^H	X^hy

50% child with hemophilia
0% girl with hemophilia * girls do not have it as they can be carriers
50% boy with hemophilia * boys guaranteed to have it as they inherit X from mother

Pedigrees

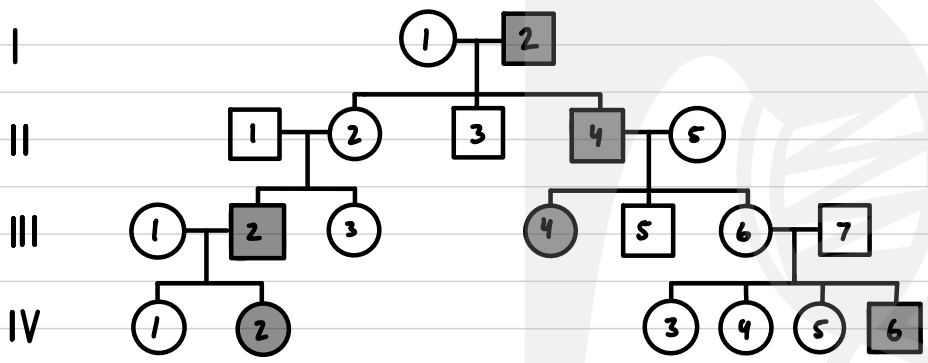
Pedigree: a chart visually showing a pattern of inheritance across generations





1. state the relationship of
   - I-2 and III-2 — grandmother and grandson
   - II-2 and II-5 — brother-in-law
   - III-1 and III-3 — 1st cousins
2. how many children did I-1 and I-2 have? 3
3. disorder is autosomal recessive. Deduce genotypes of I and II generation
   - I-1 ee I-2 Ee - has to be, otherwise nobody in II would be affected
   - II-1 Ee - has a ee child II-2 Ee II-3 Ee II-4 ee II-5 EE or Ee
   - III-1 Ee or EE III-2 ee III-3 Ee

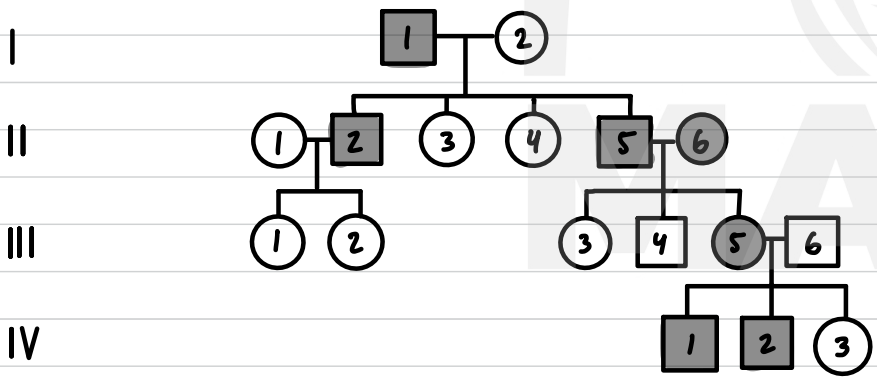
Deducing the type of inheritance of a condition can be done through analysis



Is the shaded trait autosomal dominant or recessive?
the trait is recessive.
If it was dominant, II-1 and II-2 would be aa
it would be impossible to have a child affected (III-2)

Autosomal recessive patterns

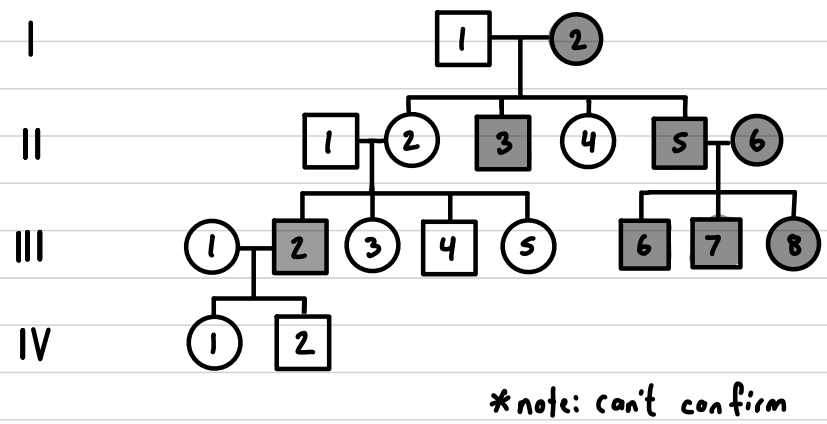
- affected people may have unaffected (heterozygous) parents
- carriers for condition - can skip generations
- same frequency of inheritance for males and females



Is the shaded trait autosomal dominant or recessive?
the trait is Dominant
If the trait is recessive, than II-5 and II-6 would be aa
it would be impossible to have a child unaffected (III-3 + 4)

Autosomal Dominant patterns

- every affected person has at least 1 affected parent
- no carriers for condition - rarely skips generations
- same frequency of inheritance for males and females



*note: can't confirm sex-linkage, only infer strongly

Is the shaded recessive trait autosomal or X-linked?
trait is X-linked
only time female is affected is when both parents carry the recessive allele (X^a) as females need two copies to be affected

X-linked recessive patterns

- all children of affected only fathers (mother not carrier) are unaffected
- all sons of affected mothers are affected (inherit X from mother)
- female carriers for condition - can skip generations
- males are affected more frequently than females (only need 1 allele)

Assessment Tasks

(1) The traits in pea plants Mendel studied are a form of inheritance called 'complete dominance'. Explain why.

(2) For the following crosses.

i) Determine the phenotype and genotype of the parents
ii) Draw a Punnett square of the cross
iii) Determine the phenotypic ratio of offspring
iv) Determine the genotypic ratio of offspring

a) Hornless (H) in cattle is dominant over horned (h). A horned bull is mated with a homozygous hornless cow

b) In tomatoes, red fruit (R) is dominant over yellow fruit. A heterozygous red is crossed with yellow.

c) In guinea pigs, short hair (S) is dominant over long (s). Two heterozygous short hairs mated

d) In chickens, black feathers (C^B) is co-dominant to white (C^W). A white rooster mated with a white/black chicken

e) Man has type O blood has children with an AB woman.

f) Red-green colour blindness is sex-linked recessive in humans (X^b). A colour blind male and carrier female

(3) Sickle-cell anemia is a co-dominant trait with two different alleles: Hb^A (normal) and Hb^S (sickle cell)

a) Using research, provide all possible phenotypes with their corresponding genotypes

b) In a malaria-prevalent area, which genotype is ideal? Explain.

(4) Using your understanding of antigens and antibodies, explain and justify which blood type is

a) the universal donor

b) the universal recipient

(5) A woman has type B blood and her husband has AB blood. Is it possible for them to have a child with O. Explain

(6) Draw a 3 generation pedigree chart clearly showing the inheritance of a sex-linked recessive disorder. Start with an affected male and unaffected female

(7) Analyze the 3 pedigrees on the previous page. For each, deduce as many of the genotypes as possible

(8) The pedigree below shows the inheritance of fur colour in mice.

a) deduce which characteristic is dominant

b) deduce the genotypes of as many individuals as possible