A. VOCABULARY

TERM	MEANING
GENE	A **sequence of DNA bases** that **codes** for a **specific protein** e.g. gene for eye colour
ALLELE	A **different form** of the **same gene** e.g. alleles for brown eyes, blue eyes, green eyes
HOMOZYGOUS	Having **two identical alleles** of a gene e.g. TT or tt
HETEROZYGOUS	Having **two different alleles** of a gene e.g. Tt
DOMINANT ALLELE	The allele that is **always expressed** if **present** The allele that is **expressed** when even **one copy** is present
RECESSIVE ALLELE	The allele that is **only expressed** when the **dominant allele is absent** The allele that is **only expressed** when **two copies** are **present**
GENOTYPE	The **alleles carried** for a gene e.g. TT, Tt or tt
PHENOTYPE	The **outward expression** of a gene. The **characteristics** of an organism. e.g. tongue roller, non-roller
PHENOTYPIC RATIO	The **ratio** of **phenotypes** possible in the **offspring** from a genetic cross.
LOCUS	The **position** of a gene on a chromosome i.e. where it is on a chromosome
AUTOSOME	A **non-sex** chromosome i.e. not the last pair!
SEX-LINKED	When a gene is **located** on a **sex chromosome**
X-LINKED	When a gene is **located** on the **X chromosome**
F1 GENERATION	The **offspring from the first mating** (sons and daughters)
F2 GENERATION	The **offspring from the second mating** (grandsons and granddaughters)
CO-DOMINANCE	When two or more **alleles** have the **same level of dominance.** In **heterozygotes**, **both alleles** will be **expressed.** **Three phenotypes** are often seen instead of the usual two
PURE BREEDING	When an individual is **homozygous** for a **gene** e.g. TT or tt
MULTIPLE ALLELES	When **more than two alleles** for a **gene** exist. Remember though, that a person can only carry two of these.

B. THE WORK OF GREGOR MENDEL

- He studied **seed shape** in peas, which is controlled by **one** gene.
- The dominant allele, **S**, produces **smooth** seeds and the recessive allele, **s**, produces **wrinkled** seeds.
- He did the following cross:

S = Smooth seeds
s = wrinkled seeds

Parental phenotypes: Pure breeding Smooth seeds x wrinkled seeds

Parental genotypes: (**SS**) (**ss**)

	s	**s**
S	Ss	Ss
S	Ss	Ss

F1 (First Generation) Offspring genotypes

F1 (First Generation) Offspring phenotypes: All wrinkled seeds

- He then crossed two of the F1 (first generation) plants, which had smooth seeds:

F1 offspring Smooth seeds x F1 offspring Smooth seeds

Parental genotypes: (**Ss**) (**Ss**)

	S	**s**
S	SS	Ss
s	Ss	ss

F2 (Second Generation) Offspring genotypes

F2 (Second Generation) Offspring phenotypic ratio: **3** Smooth seeds : **1** wrinkled seeds

Crossing two heterozygous individuals gives a 3 : 1 ratio for the offspring phenotypes

C. MENDEL'S LAW OF SEGREGATION

Individual's Genotype	Gametes That Can Be Produced
Ss	S and s

The two alleles of a gene separate (segregate) into different haploid gametes during meiosis

D. MONOHYBRID CROSSES

- These are crosses that involve **one** gene.

Cystic fibrosis is a lung disease. It is caused by a recessive allele, c.

John and Sue are both **healthy** but **carry the allele for cystic fibrosis**.

Use a genetic diagram to show the probability of this couple producing a child with cystic fibrosis.

C = Health
c = cystic fibrosis

Define the alleles

	John	x	Sue
Parental phenotypes:	(Healthy)		(Healthy)
Parental genotypes:	(Cc)		(Cc)

Gametes

	C	c
C	Cc	Cc
c	Cc	cc

Offspring genotypes:

Probability of producing a child with cystic fibrosis = 1/4 = ¼ = **25%**
(**NOTE:** probability of producing a daughter with cystic fibrosis = ½ x 25 = 12.5%)

Huntington's disease is caused by a dominant allele, H. It affects the nervous system.

Steve is heterozygous for Huntington's disease. His wife, Mary, does not carry allele H.

Use a genetic diagram to show the phenotypic ratio of offspring produced by this couple.

H = Huntington's disease
h = healthy

Steve	x	Mary
(Heterozygous)		(Does not carry allele H)
(**Hh**)		(hh)

	H	**h**
h	Hh	hh
h	Hh	hh

Phenotypic ratio of offspring = 2 Huntington's : 2 healthy = **<u>1 Huntington's : 1 healthy</u>**

<u>E. PEDIGREE CHARTS</u>

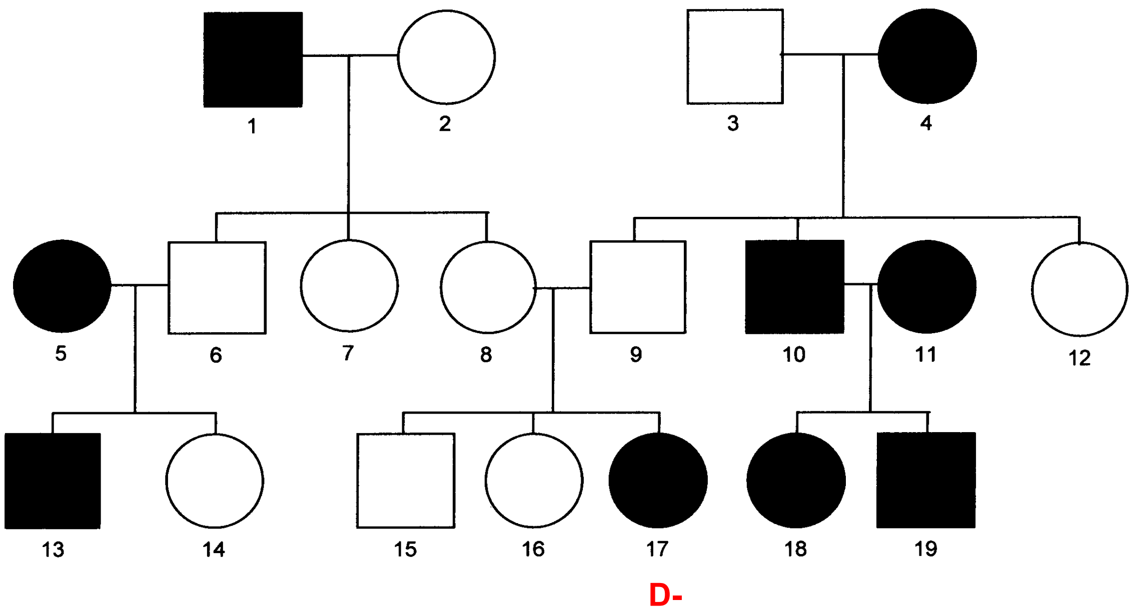
- Always **write** the **genotypes** of individuals **on** these, **next** to their **symbols**. It makes questions much easier.
- They show **generations** of a family, not just two parents.
- Examiners can ask you to prove that a genetic disorder is **caused** by a **recessive allele** or a **dominant allele** from a pedigree chart.

The easiest way to do this is to show that it **CANNOT** be caused by the **OTHER ALLELE**

Prove That A Genetic Disorder Is Caused By	What To Do
Recessive allele	Pretend it is caused by the **dominant** allele Look for **two healthy parents** producing an **affected child** Show that this **does not work**
Dominant allele	Pretend it is caused by the **recessive** allele Look for **two affected parents** producing a **healthy child** Show that this **does not work**

<u>Prove that the genetic disorder below is caused by a recessive allele.</u>

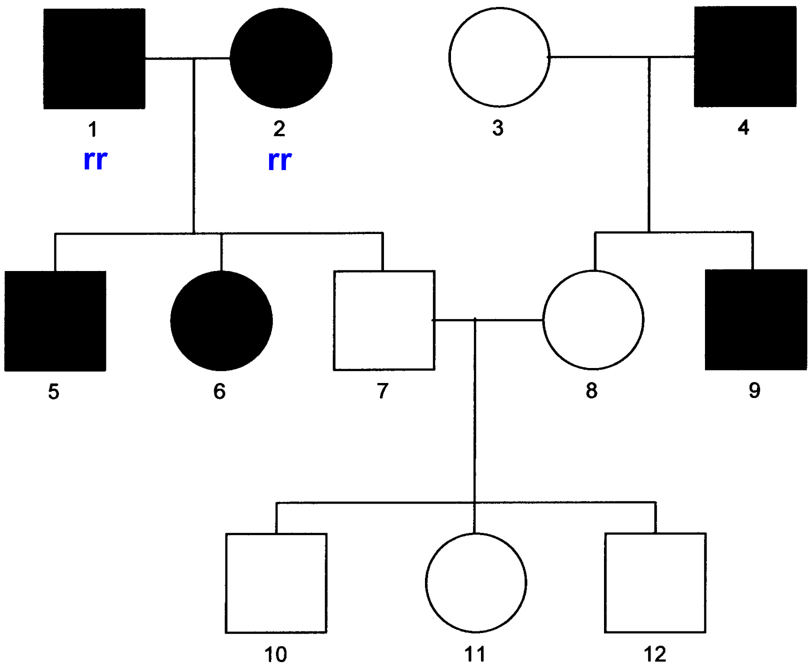
- Pretend it is caused by the **dominant** allele
- Look for **two healthy parents** producing an **affected child**
- Show that this **does not work**



- Focus on individuals **8**, **9** and **17**.
- If **caused by a dominant allele**, **D**, **17** <u>must</u> carry this allele.
- **17** got this dominant allele from (at least) one of its **parents**, **8** or **9**.
- (So) **at least one** of its **parents**, **8** or **9**, <u>must</u> be **affected**.
- They are both healthy, so it **does not work** – this must be caused by a **recessive allele**.

<u>Prove that the genetic disorder below is caused by a dominant allele.</u>

- Pretend it is caused by the **recessive** allele
- Look for **two affected parents** producing a **healthy child**
- Show that this **does not work**



- Focus on individuals **1**, **2** and **7**.
- **If caused by a recessive allele**, **r**, **1 & 2** <u>must</u> be homozygous recessive (rr).
- (So) they can **only** produce **affected** children (rr).
- (But) they produce a **healthy** child / 7.
- (So) it **does not work** – this must be caused by a **dominant allele**.

F. CO-DOMINANCE

- When two or more **alleles** have the **same level of dominance.**
- In **heterozygotes**, **both alleles** will be **expressed.**
- **Three** (or more) **phenotypes** are often seen instead of the usual two.

The ABO Blood Group System

- Alleles $\mathbf{I^A}$ (A) and $\mathbf{I^B}$ (B) are **co-dominant**.
- Allele **i** (O) is **recessive** to $\mathbf{I^A}$ and $\mathbf{I^B}$.

Blood Group	Possible Genotype(s)
A	$I^A I^A$ and $I^A i$
B	$I^B I^B$ and $I^B i$
AB	$I^A I^B$
O	$i\ i$

The ABO Blood Group System is an example of **MULTIPLE ALLELES**.

There are **three** different alleles but a person can only carry **two** of these.

Jason is blood group AB and his wife, Sarah, is blood group O.

Use a genetic diagram to show the phenotypic ratio of offspring that they can produce.

Jason	x	Sarah
(AB)		(O)
($I^A I^B$)		(**i i**)

	I^A	I^B
i	$I^A i$	$I^B i$
i	$I^A i$	$I^B i$

Phenotypic ratio of offspring = 2 A : 2 B = **1 Group A : 1 Group B**

David is blood group A and his wife, Jessie, is blood group B.

They produce a child that has a different blood group to **both of them**.

Use a genetic diagram to show how this is possible.

David	x	Jessie
(A)		(B)
(I^A i)		(I^B i)

	I^A	i
I^B	I^A I^B	I^B i
i	I^A i	i i

It is possible to produce children who are of blood group **AB** and **O**.

The table shows the blood groups of four people in a paternity test to determine the father of a child.

Person	Blood Group
Mother	B
Child	O
Father 1	AB
Father 2	A

Use genetic diagrams to make conclusions.

Starting point: The child is **i i** (blood group O) and the mother is **I^B i** (blood group B)

Father 1 (AB)

	I^A	I^B
I^B	I^A I^B	I^B I^B
i	I^A i	I^B i

Cannot produce a child of blood group O

Father 1 **cannot** be the real father

Father 2 (A)

	I^A	i
I^B	I^A I^B	I^B i
i	I^A i	i i

Can produce a child of blood group O

Father 2 **could** be the real father

G. SEX-LINKAGE

- When a **gene** is located on a **sex chromosome**, X or Y.
- In humans, **haemophilia** and **red-green colour-blindness** are caused by **recessive alleles** located on the **X-chromosome**.
- These genes are said to be **X-linked** as they are **on** the **X-chromosome**.
- **Males** (XY) get their **X-chromosome** from their **mother**.

With this, **SEX MATTERS!**

You **must** write down:

XX or **XY** to show if an individual is female or male

The **sex** of an individual in the **phenotypic ratio**

Haemophilia is a condition in which the blood fails to clot.

It is caused by the recessive allele, h, found on the X-chromosome.

Paul and his wife, Tracy are both healthy. However, Tracy, carries the allele for haemophilia.

Use a genetic diagram to show the phenotypic ratio of offspring produced by this couple.

H = Healthy

h = haemophilia

NOTE:
This allele is **only found on the X-chromosome**, not on the Y-chromosome

Paul (haemophilia)	x	Tracy (healthy)
(X^H Y)		(X^H X^h)

NOTE:
Tracy is a still healthy as the allele for haemophilia is **recessive**

	X^H	Y
X^H	X^H X^H	X^H Y
X^h	X^H X^h	X^h Y

Phenotypic ratio of offspring = **2 healthy female : 1 healthy male : 1 haemophilia male**

Red-green colourblindness is a condition in which the colours red and green look similar.

It is caused by the recessive allele, r, found on the X-chromosome.

Max is red-green colourblind but his wife, Stacey, has normal vision.
However, Stacey's father is red-green colourblind.

Use a genetic diagram to show the phenotypic ratio of offspring produced by this couple.

R = Normal vision

r = red-green colourblind

Max (red-green colourblind) x Stacey (normal vision)

($X^r Y$) ($X^R X^r$)

NOTE:
Stacey must be heterozygous. She must have X^R for normal vision but received X^r from her father, who was colourblind

	X^r	Y
X^R	$X^R X^r$	$X^R Y$
X^r	$X^r X^r$	$X^r Y$

Phenotypic ratio of offspring = **1 normal vision female : 1 colour-blind female :**

1 normal vision male : 1 colour-blind male

H. WHY DO MORE MALES THAN FEMALES HAVE RECESSIVE X-LINKED DISEASES?

$X^h Y$

Males have one X-chromosome
Cannot be carriers/heterozygous
Cannot also carry the dominant allele to mask the effect of the recessive allele

$X^H X^h$

Females have two X-chromosomes
Can be carriers/heterozygous
Can also carry the dominant allele to mask the effect of the recessive allele

I. PEDIGREE CHARTS & SEX-LINKAGE

- Look for **much more of one sex being affected** than the other.

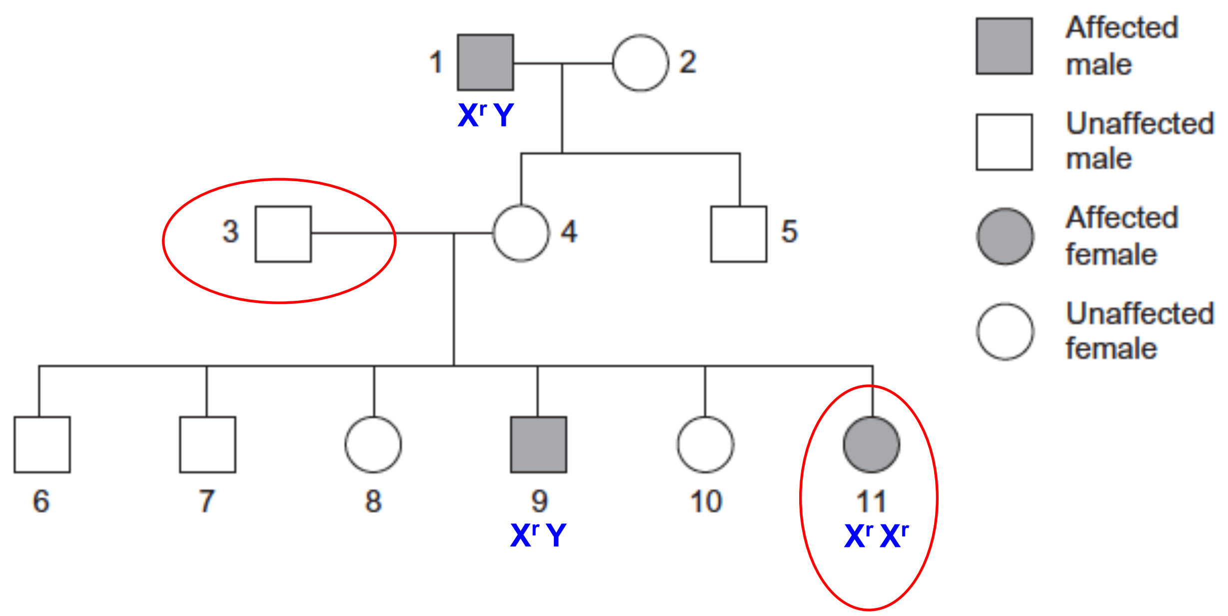
- Remember to always **write what you know on** the pedigree chart, **next to the individuals**.

- To prove that a **genetic disorder** is **not sex-linked**, pretend that it **is sex-linked** and show that it **does not work this way**.

The pedigree chart below shows a genetic disease caused by a recessive allele, r.

Prove that this disease is **not** on the **X-chromosome**.

Pretend that it **is sex-linked** and write-in **what you know** for **certain**:



- Look for a **healthy man producing an affected daughter**.
- If **sex-linked, 11** must be **$X^r X^r$**.
- (So) her **father/3** must have given her **X^r**.
- (So) her **father/3** must be **affected**.
- (But) her f**ather/3** is **healthy**.
- (So) it **does not work** – this is **not sex-linked**.

NOTE: This recessive allele **cannot be on the Y-chromosome** either

as **there are affected females**

J. TEST CROSS

- When an organism is mated with the **recessive** phenotype.

- It is used to **determine** the **genotype** of an organism, when there is **more than one possibility**.

- The **offspring phenotypes** are used to **determine** the **genotype** of the **parent**.

> In a species of fruit fly, eye colour is controlled by a gene with two alleles.
>
> The dominant allele, R, gives red eyes.
> The recessive allele, r, gives white eyes.
>
> You are given a fly with red eyes and asked to determine its genotype.
>
> Explain how you would do this.

Problem: you do not know if the fly with red eyes has the genotype **RR**, or **Rr** – both are possible.

Mate this fly with the **recessive phenotype**/a fly with **white eyes** (**rr**).

- (As) you **know** the **genotype** of a fly with **white** eyes - there is only **one** possibility (**rr**).

<table>
<tr><td>If the fly with red eyes is RR:</td><td>If the fly with red eyes is Rr:</td></tr>
<tr><td>
<table>
<tr><td></td><td>R</td><td>R</td></tr>
<tr><td>r</td><td>Rr</td><td>Rr</td></tr>
<tr><td>r</td><td>Rr</td><td>Rr</td></tr>
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<tr><td></td><td>R</td><td>r</td></tr>
<tr><td>r</td><td>Rr</td><td>rr</td></tr>
<tr><td>r</td><td>Rr</td><td>rr</td></tr>
</table>
</td></tr>
<tr><td>Cannot produce flies with white eyes</td><td>Can produce flies with white eyes</td></tr>
<tr><td>If no offspring have white eyes,
parent must be RR</td><td>If any offspring have white eyes,
parent must be Rr</td></tr>
</table>

K. PREDICTED AND ACTUAL RATIOS

- The **predicted** and **actual ratios** of **offspring phenotypes** seen can **differ**.

- This can be caused by:

  - a **small sample size**
  - **fertilisation** being a **random** process
  - some **alleles** being **lethal** when combined

Example of lethal alleles: coat colour in mice

Y = Yellow coat
y = grey coat

If we breed two yellow **heterozygous** mice:

(Yellow) (Yellow)

(Yy) **(Yy)**

We would expect:

	Y	y
Y	YY	Yy
y	Yy	yy

Expected offspring phenotypic ratio = **3 Yellow: 1 Grey**

However, it is more common to an offspring phenotypic ratio of **2 Yellow : 1 Grey**

This is because mice that are **homozygous (YY) die when embryos**.

A **combination** of **two dominant alleles** is **lethal**.

L. MORE DIFFICULT CROSSES

- You are much more likely to only be tested **only** on what you have just read.
- However, examiners **have** went further before by **combining** different types of cross!
- The key things to remember here are these:

Keep the **different genes separate** when **writing genotypes**

Any allele of one gene can **end up with any allele of the other gene** in a **sex cell**

Draw **one** genetic diagram – **not two separate ones**.

The best way to show this is with examples:

1. A Gene On A Non-Sex Chromosome & Co-Dominance

James is blood group O and has cystic fibrosis.

Jane is blood group AB and does not have cystic fibrosis.
However, she does carry the allele for cystic fibrosis.

Use a genetic diagram to show the show the probability of this couple having a child that is blood group A and healthy.

Deal with **each gene separately** when writing the **phenotype** and **genotype**

James	x	Jane
(cystic fibrosis and O)		(healthy and AB)
(**cc** and **ii**)		(**Cc** and $\mathbf{I^A\ I^B}$)

Any allele of one gene can **end up with any allele of the other gene** in a **sex cell**.

This is why **James can** produce **one type of sex cell** but **Jane can** produce **four different types.**

	c i
C $\mathbf{I^A}$	Cc I^A i
c $\mathbf{I^A}$	cc I^A i
C $\mathbf{I^B}$	Cc I^B i
c $\mathbf{I^B}$	cc I^B i

Draw only **one genetic diagram** – **not** two separate ones.

Probability of child being healthy **and** blood group A = ¼ = **25%**

2. A Gene On A Non-Sex Chromosome & A Gene That Is Sex-Linked

The gene for tongue-rolling is not sex-linked and is due to a dominant allele, T.

Haemophilia is caused by a recessive allele, h, found on the X-chromosome.

Luke has **haemophilia** and is **heterozygous** for **tongue-rolling**.

His wife, Linda, **does not carry alleles for haemophilia or tongue-rolling**.

Use a genetic diagram to determine the phenotypic ratio of offspring that this couple could produce.

Deal with **each gene separately** when writing the **phenotype** and **genotype**

Luke	x	Linda
haemophilia and heterozygous roller		healthy and non-roller
(**X^h Y** and **Tt**)		(**X^H X^H** and **tt**)

Any allele of one gene can **end up with any allele of the other gene** in a **sex cell**.

'Any X or Y can go with any T or t'

	X^H t
X^h T	X^H X^h Tt
X^h t	X^H X^h tt
Y T	X^H Y Tt
Y t	X^H Y tt

Draw only **one genetic diagram** – **not** two separate ones.

Phenotypic ratio of offspring = **1 female healthy roller : 1 female healthy non-roller:**

1 male healthy roller : 1 male healthy non-roller