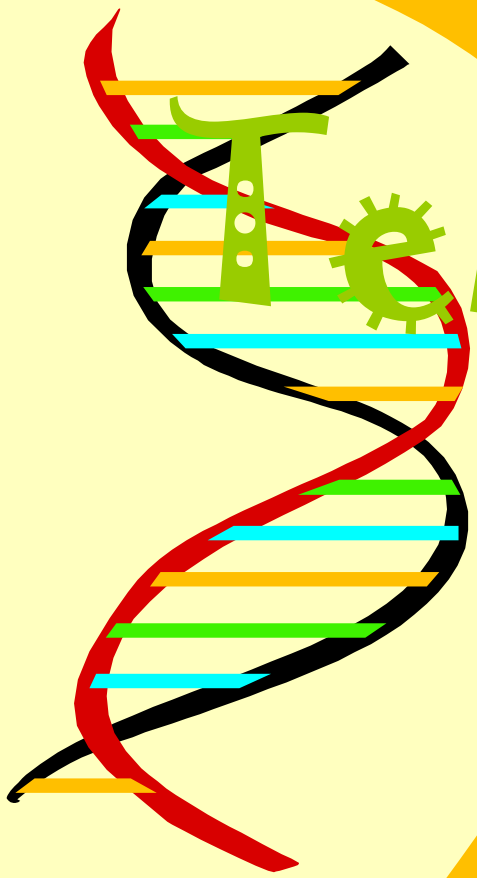
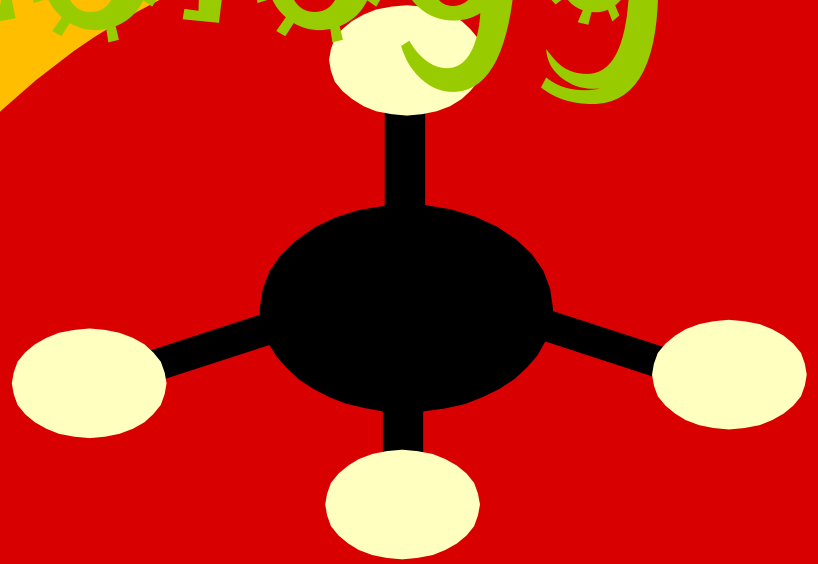
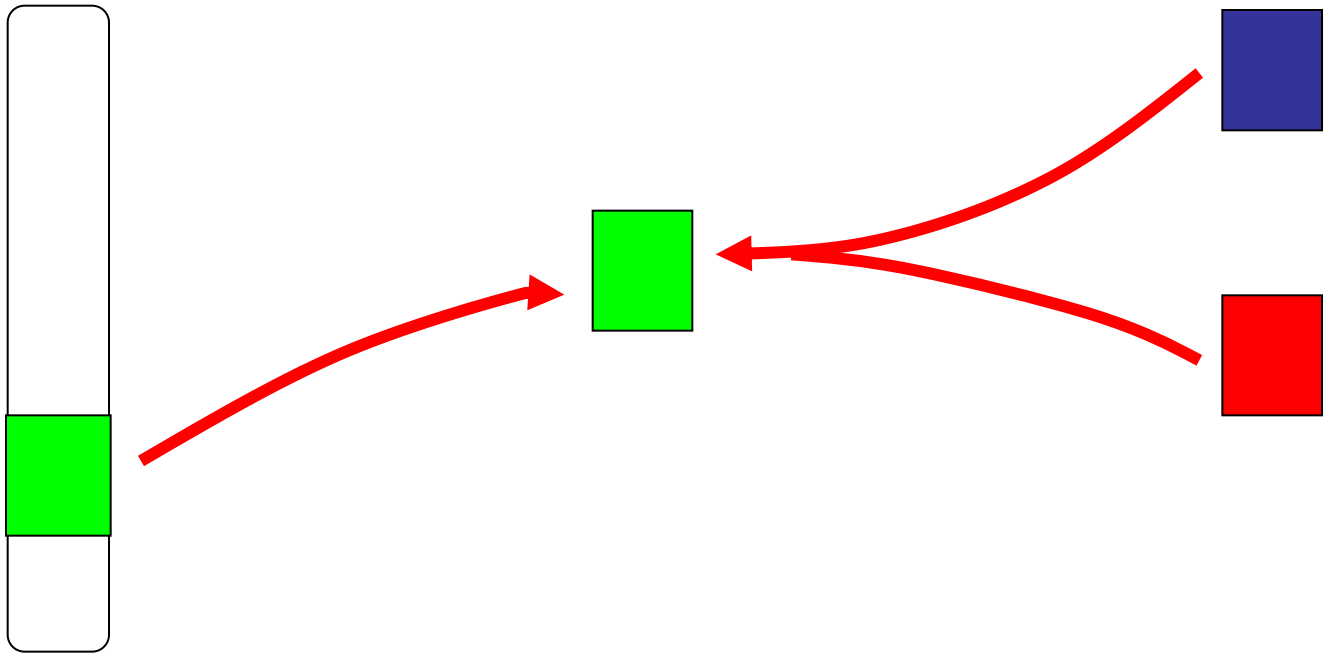
Genetic Terminology





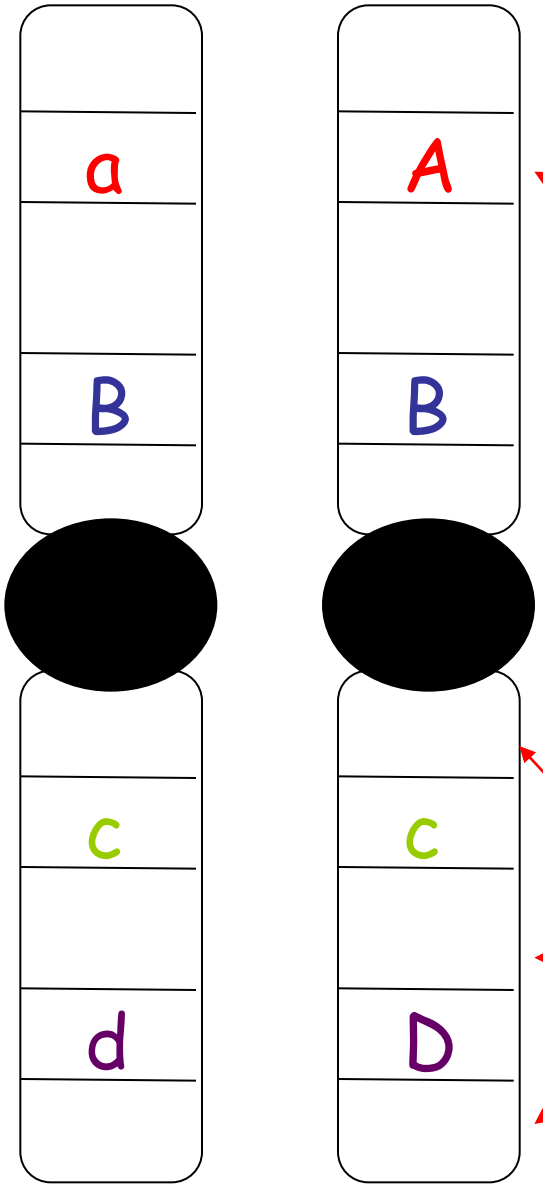
chromosome



locus of gene

gene

alleles



Each chromosome consists of;

- A linear sequence of genes
- A centromere
- Regions of repetitive DNA NOT organised into genes

worksheet
genetic definitions

TERM	DEFINITION
PHENOTYPE	Physical and chemical characteristics of an individual, caused by environment **and** genes e.g.
GENOTYPE	Combination of genes carried by an individual
GENE	Segments of nucleic acids specifying the amino acid sequence of polypeptides and proteins

TERM	DEFINITION
LOCUS	Position of a gene on a chromosome
ALLELES	Different nucleic acid combinations for a specific gene e.g.
HOMOLOGOUS CHROMOSOMES	Chromosomes carrying the same loci and which pair during prophase of meiosis I
DIPLOID CELL	A cell with 2 sets of chromosomes
HAPLOID CELL	A cell with 1 set of chromosomes

TERM	DEFINITION
GENE PAIR	The 2 copies of a particular gene found in a diploid cell
HOMOZYGOUS GENE PAIR	A gene pair with identical alleles
HETEROZYGOUS GENE PAIR	A gene air with different alleles

TERM	DEFINITION
DOMINANT ALLELE	An allele whose effect is expressed in the phenotype even in the presence of a recessive allele (capital letter)
RECESSIVE ALLELE	An allele whose effect is expressed in the phenotype ONLY in the presence of another identical allele
CROSS	Mating of 2 organisms
SELFING	Fertilising female gamete with male gamete from the same organisms (usually plants)
F1 GENERATION	The first generation of a cross
F2 GENERATION	The generation produced by crossing F1 organisms

VARIATION

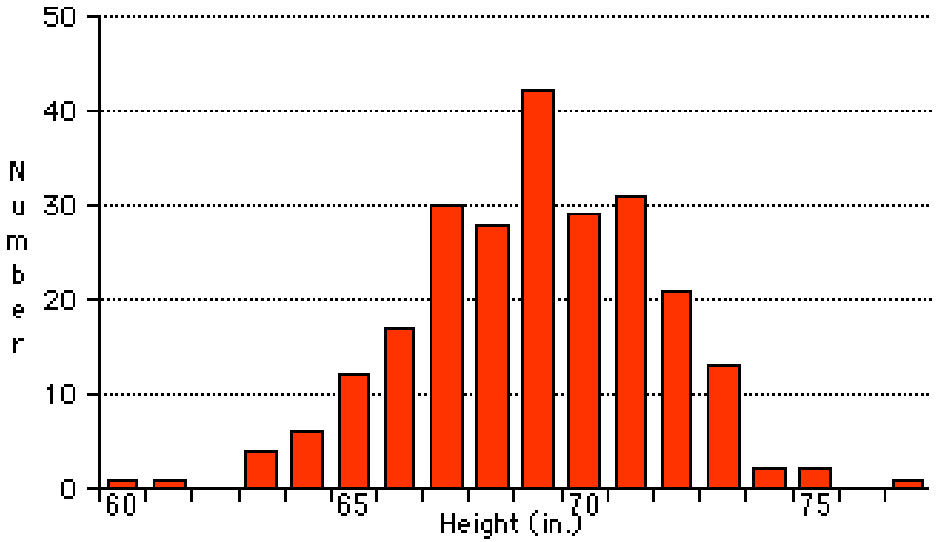
all variation is caused by a combination of the genotype AND environment

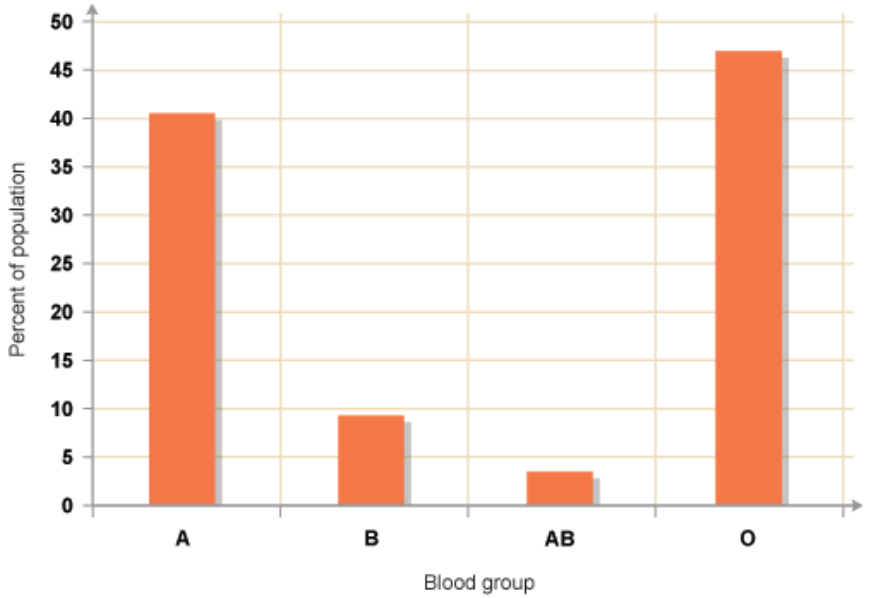
CONTINUOUS

A range of characteristics between 2 extremes

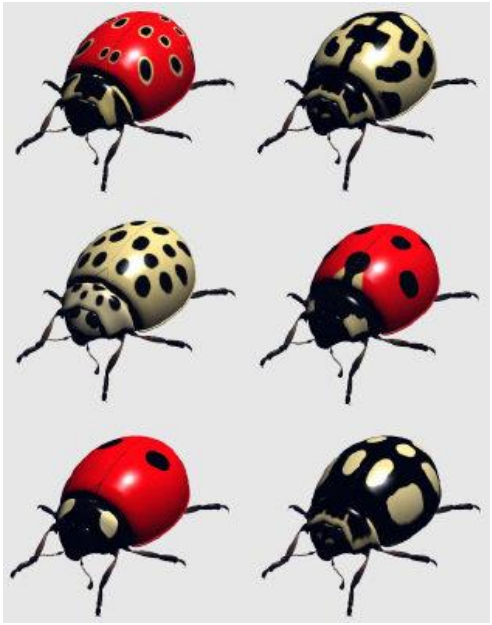
DISCONTINUOUS

Characteristics fall into one group or another





continuous or discontinuous?



DISCONTINUOUS VARIATION

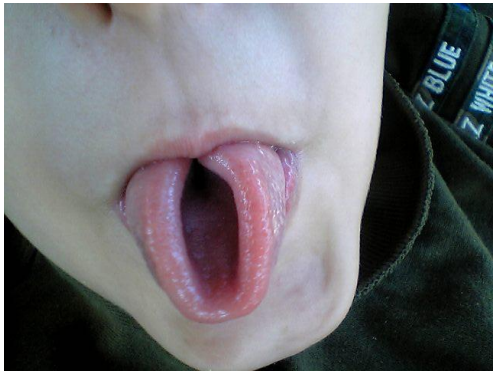
is the result of single genes

CONTINUOUS VARIATION

is caused by many genes interacting

MONOHYBRID INHERITANCE

A single gene is responsible for a single characteristic.



DOMINANT & RECESSIVENESS

Each gene has 2 alleles e.g. height in pea plants:

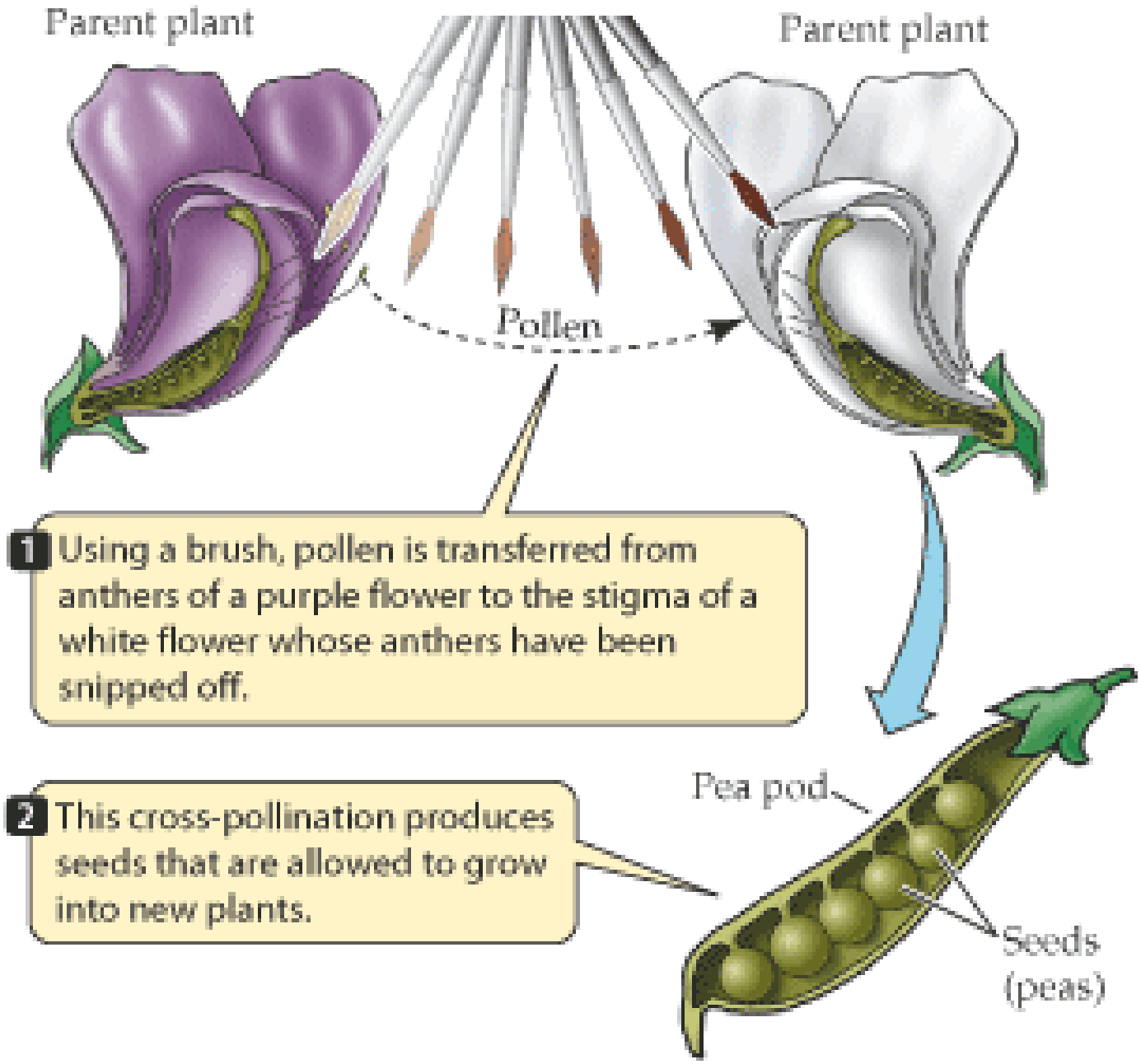
Allele T = dominant

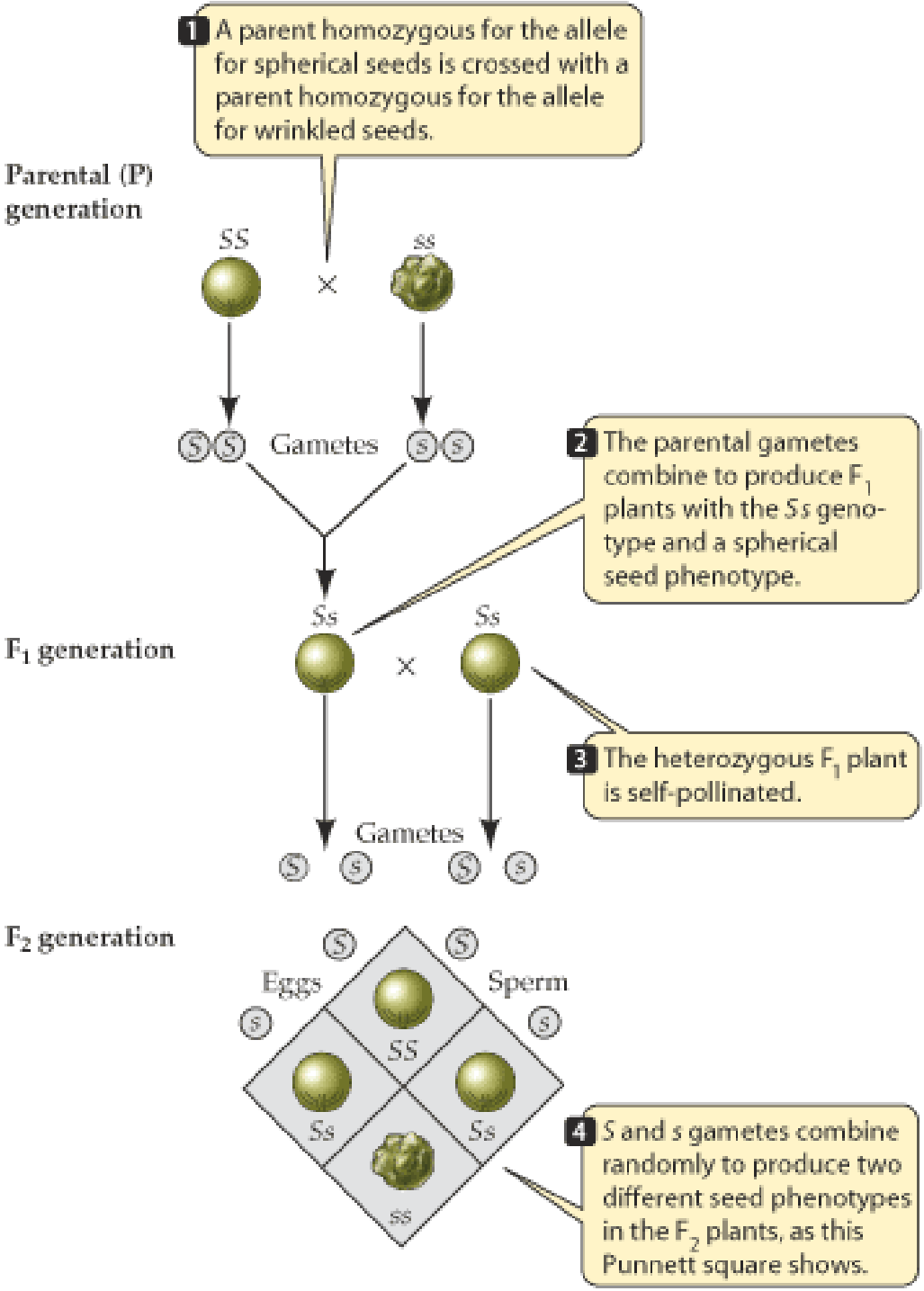
Allele t = recessive

Gametes are produced by meiosis so have only ONE copy of each allele

- TT only produces gametes with allele T
- tt only produces gametes with allele t
- Tt produces gametes with T and gametes t

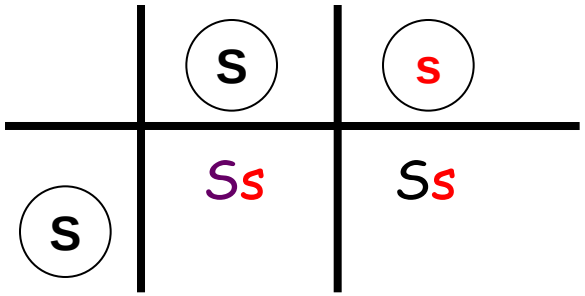
Pea flower cross-pollination




1 A parent homozygous for the allele for spherical seeds is crossed with a parent homozygous for the allele for wrinkled seeds.
Parental (P) generation
SS
ss
Gametes
2 The parental gametes combine to produce F1 plants with the Ss genotype and a spherical seed phenotype.
F1 generation
Ss
Ss
3 The heterozygous F1 plant is self-pollinated.
Gametes
F2 generation
Eggs
Sperm
SS
Ss
Ss
ss
4 S and s gametes combine randomly to produce two different seed phenotypes in the F2 plants, as this Punnett square shows.

Parent phenotype:	spherical seed	spherical seed
Parent genotype:	SS	Ss
Gametes	S	S or s

Cross
Punnett square



Genotype ratio: 100% Ss

Phenotype ratio: 100% spherical

examples

1. The allele for purple flowers is P and for white flowers is p. Show a genetic cross between two heterozygous purple flowered plants and give the ratio of the offspring phenotype.
2. Explain why a homozygous purple pea plant cannot produce white flowered offspring when crossed with a white flowered plant.

Mendel's
FIRST LAW OF INHERITANCE

The characteristics of an organism are determined by alleles which occur in pairs.

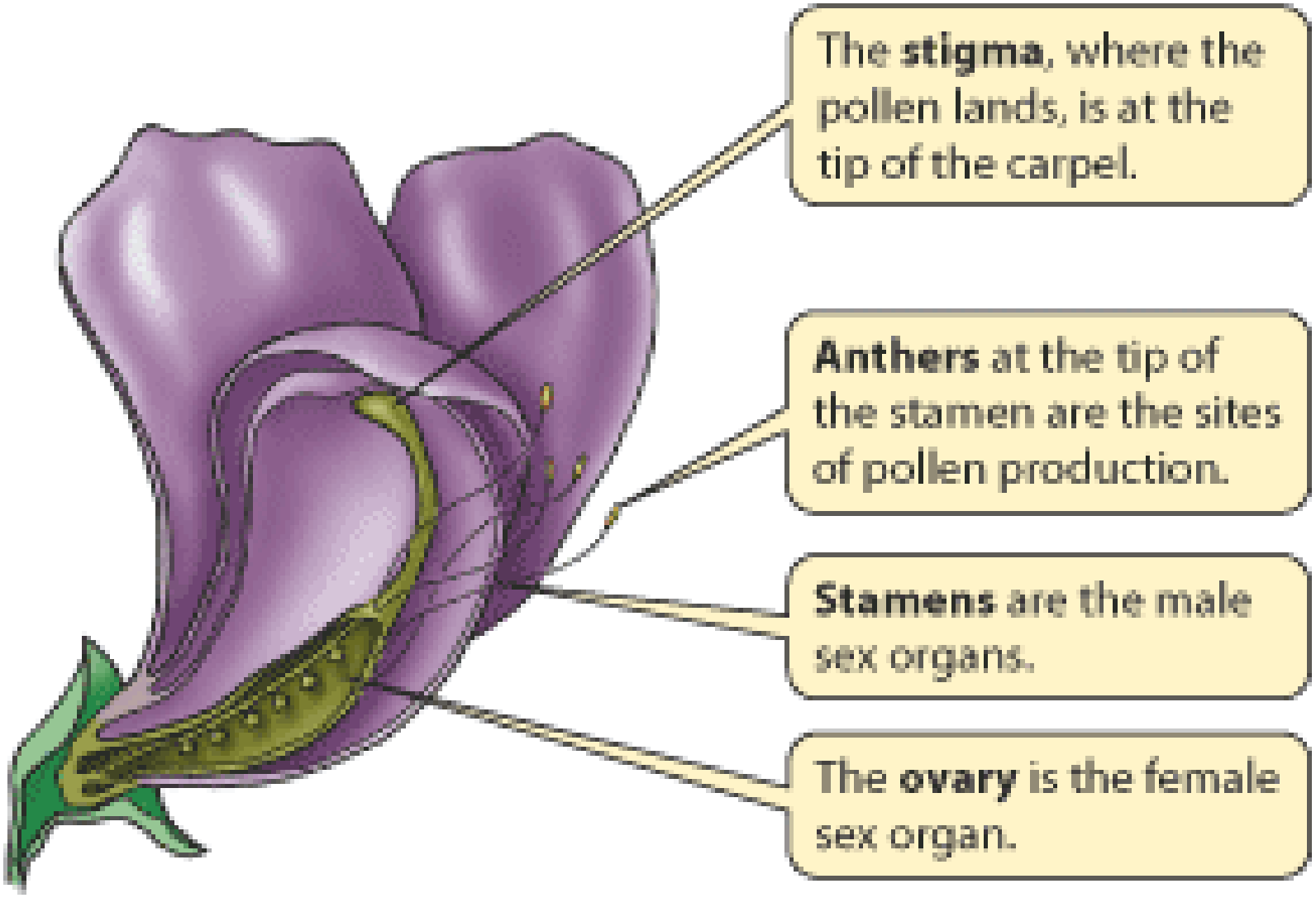
Only one of a pair of alleles can be present in a single gamete.

A TEST CROSS

This is used to determine if an organism is homozygous or heterozygous for a dominant allele

A Guinea pig test cross

- A guinea pig may have the genotype BB or Bb each giving the same black phenotype.
- Crossing the guinea pigs with a recessive white guinea pig will have different phenotype ratios in the offspring.
- BB will only produce black offspring with the genotype Bb
- Bb will also produce black offspring with genotype Bb but also white ones with genotype bb as they will inherit b from both mother and father.


The **stigma**, where the pollen lands, is at the tip of the carpel.
Anthers at the tip of the stamen are the sites of pollen production.
Stamens are the male sex organs.
The **ovary** is the female sex organ.

co-dominance

- Both alleles are dominant
- Both alleles are independently expressed

Examples include:

- Roan cattle
- MN blood groups

blood groups

- MN blood group proteins are produced by a single gene with co-dominant alleles M and N.
- The possible genotype and phenotypes are :

  MM protein M on RBC membrane

  MN proteins M + N on RBC membrane

  NN protein N on RBC membrane

The alleles are sometimes written using a letter for the gene and a superscript for the allele,

e.g. A is the gene for antigen on the red blood cells, so the alleles are A^M or A^N

The possible genotypes are then $A^M A^M$ or $A^N A^N$ or $A^M A^N$

Show the following crosses:

- Homozygous M and homozygous N
- 2 heterozygotes

lethal allelic combinations

worksheet

multiple alleles

Many genes have 3 or more alleles.

e.g. ABO blood group

An immunoglobin gene (I) which produces antigens on the plasma membrane of red blood cells has 3 alleles:

I^A produces antigen A

I^B produces antigen B

I^O produces antigen O

A diploid cell can only have 2 copies of the alleles.

Genotype	Antigen on RBC	Blood Group phenotype
$I^A I^A$		
$I^B I^B$		
I^O		

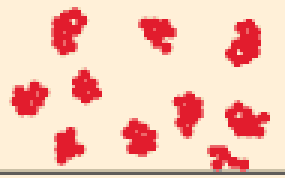
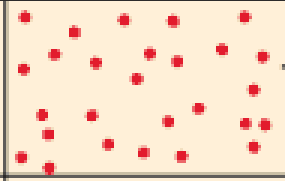
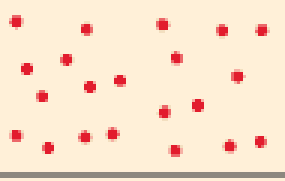
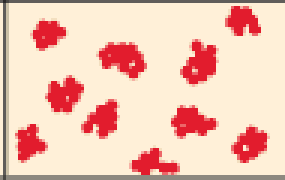
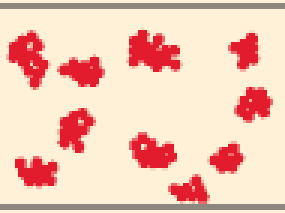
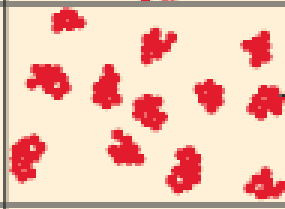
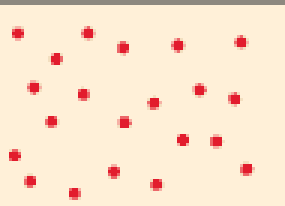
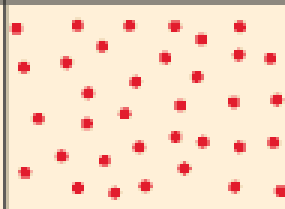
Questions

What is the evidence that:

1. I^O is recessive
2. I^A and I^B are co-dominant?

Answers

1. Phenotype O is only present when the individual is homozygous for the allele I^o
2. When these alleles are both present in the genotype the both appear separatley in the phenotype (if incomplete dominance there would be a new phenotype)

Blood type of cells	Genotype	Antibodies made by body	Reaction to added antibodies	
			Anti-A	Anti-B
A	I^AI^A or I^Ai^O	Anti-B		
B	I^BI^B or I^Bi^O	Anti-A		
AB	I^AI^B	Neither anti-A nor anti-B		
O	i^Oi^O	Both anti-A and anti-B		

Red blood cells that do not react with antibody remain evenly dispersed.

Red blood cells that react with antibody clump together (speckled appearance).

monohybrid crosses in humans

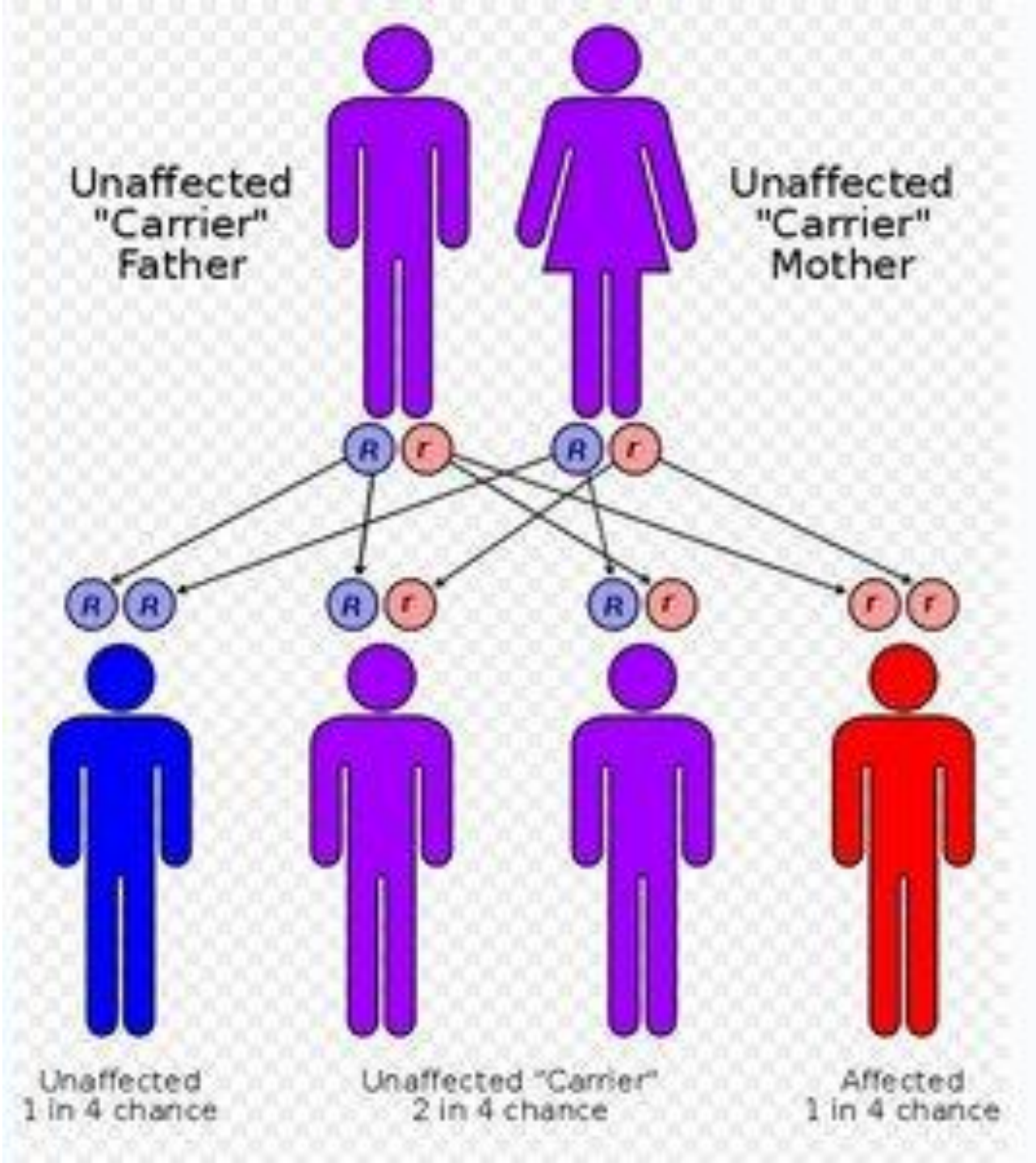
- Read the information on the following genetic disorders and decide whether it is caused by
- A dominant allele
- A recessive allele
- Incomplete dominance
- Co-dominance

  - Cystic fibrosis
  - Huntington's chorea
  - Sickle cell anaemia

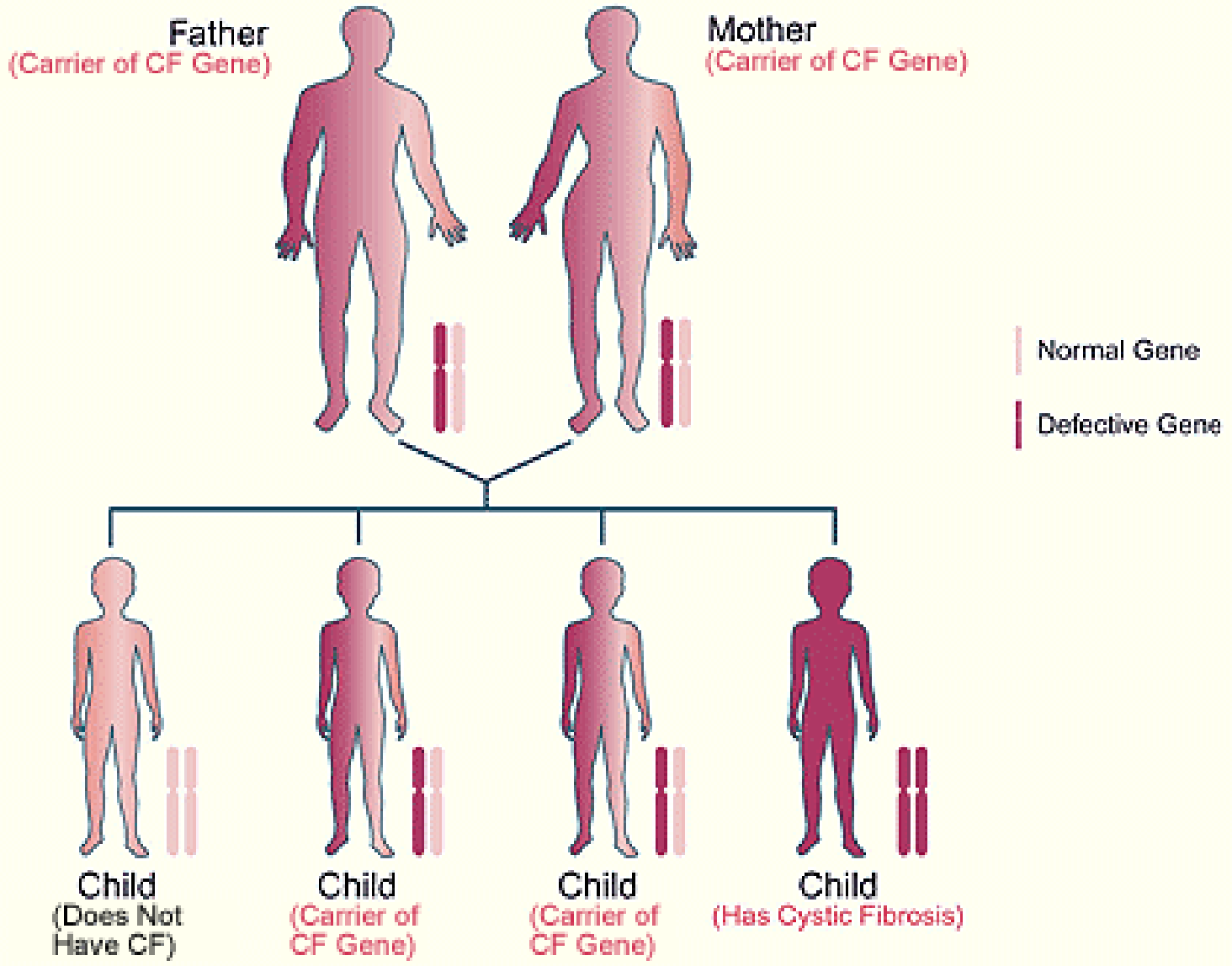
Cystic Fibrosis

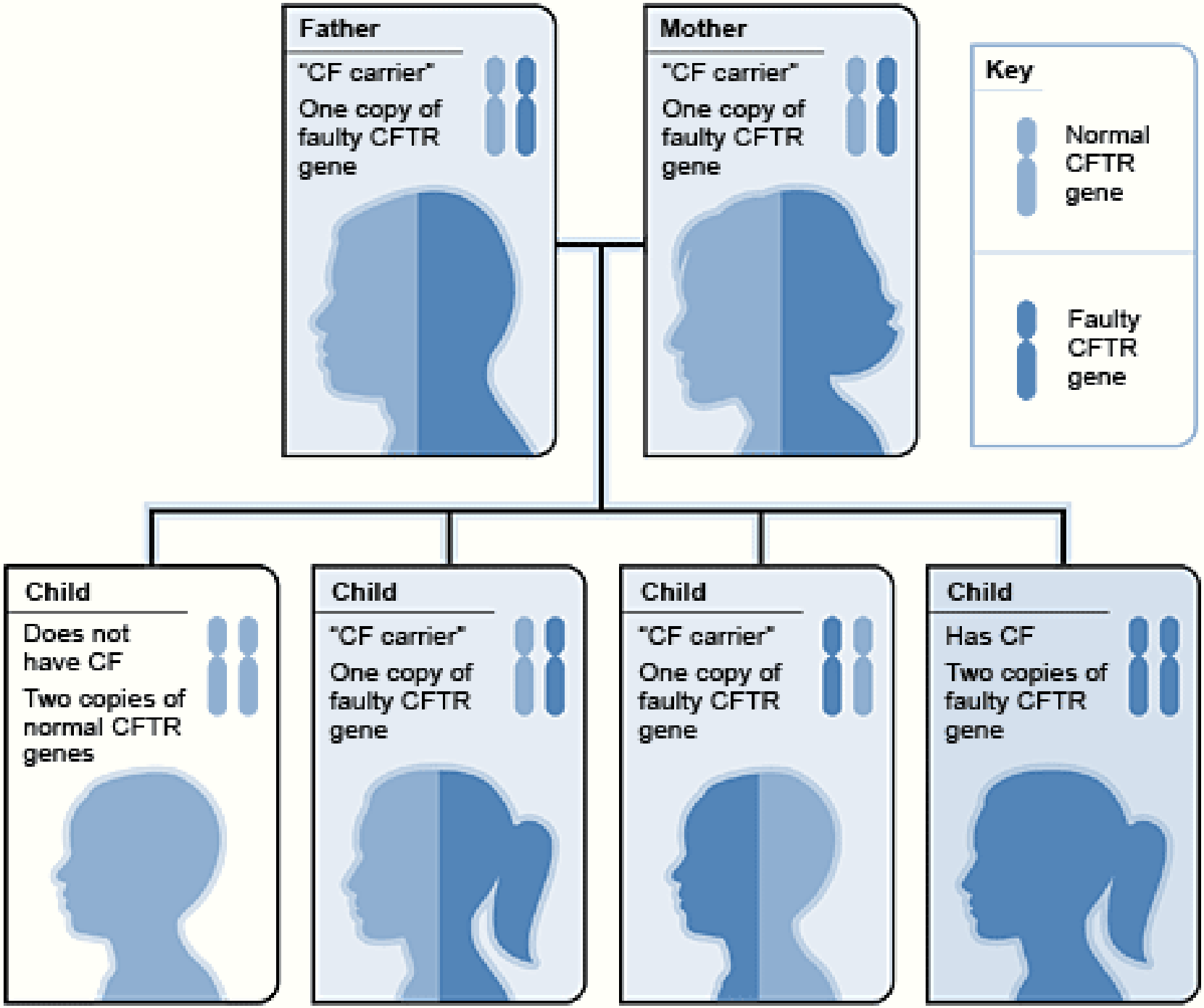
- CF results in the production of thick mucus in lungs, digestive system + reproductive tracts due to the production of a faulty carrier protein in the cell membrane
- c is recessive to C
- CC = Normal
- Cc = carrier (DO NOT suffer from CF)
- cc = CF
- carrier X carrier = 25%; 1 in 4; 1:3 chance of CF child


ERECT


Unaffected
"Carrier"
Father
Unaffected
"Carrier"
Mother
R
r
R
r
R R
R r
R r
r r
Unaffected
1 in 4 chance
Unaffected "Carrier"
2 in 4 chance
Affected
1 in 4 chance

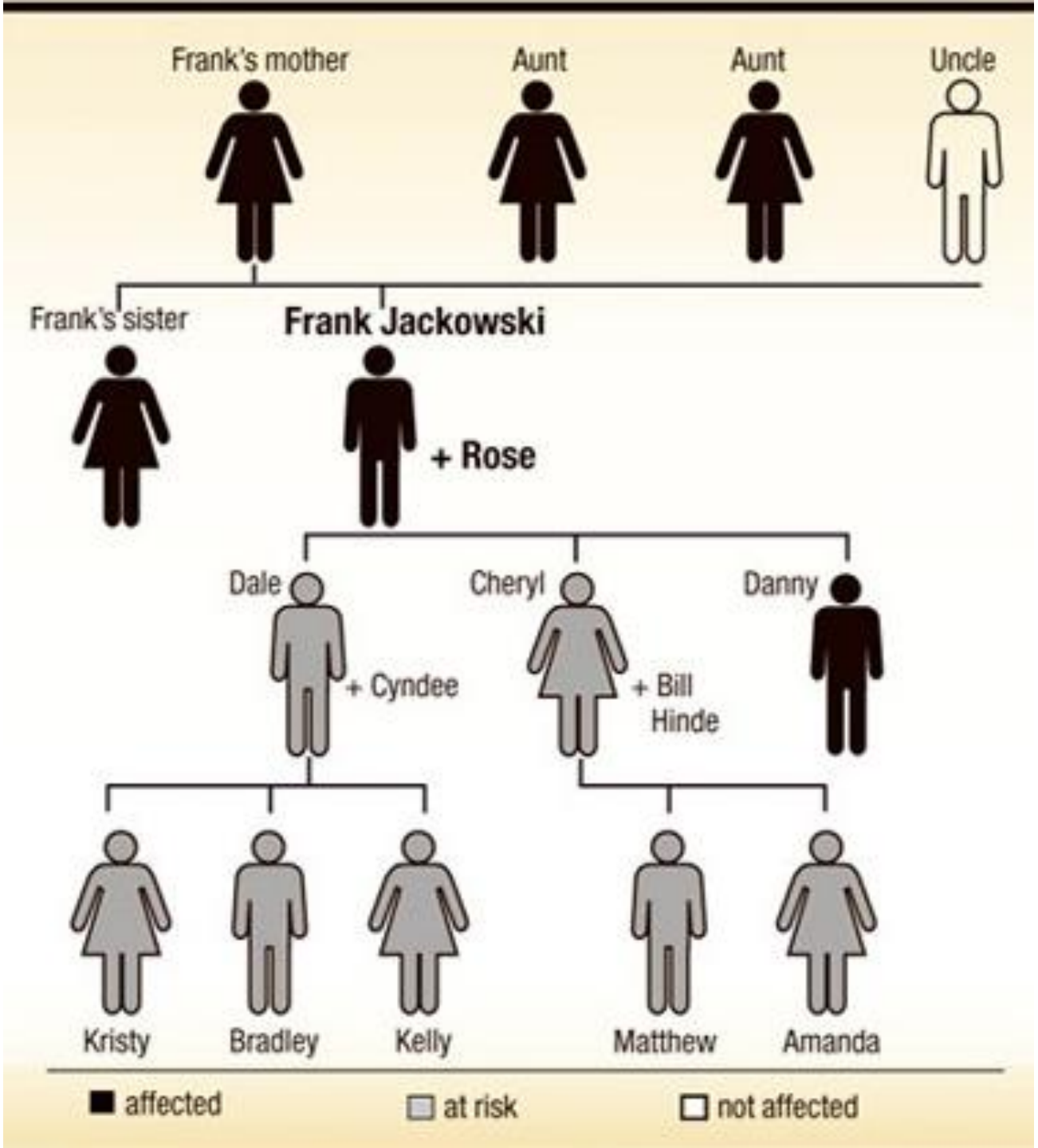
Inheritance of Cystic Fibrosis (CF)

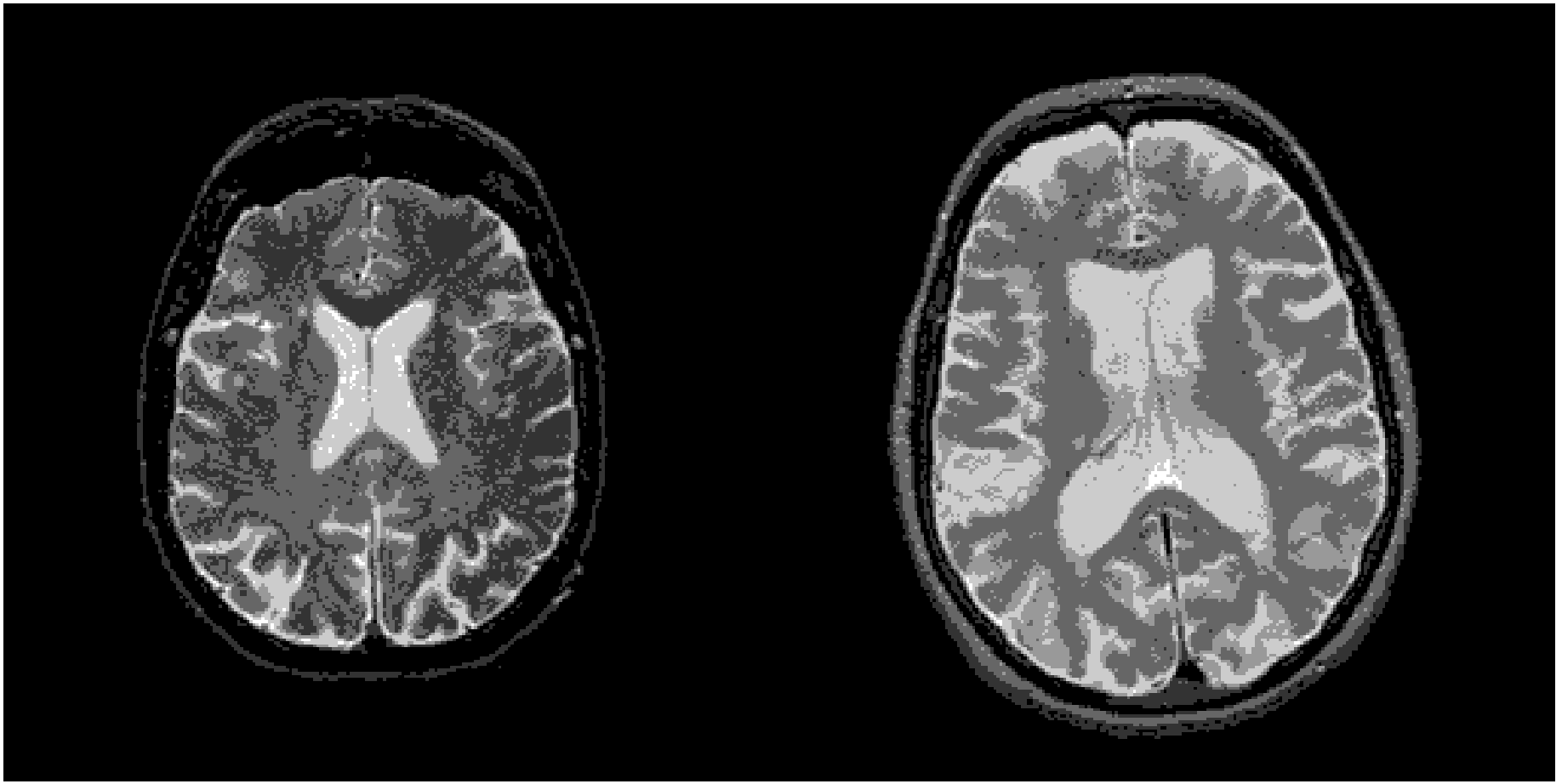



Father
"CF carrier"
One copy of faulty CFTR gene
Mother
"CF carrier"
One copy of faulty CFTR gene
Key
Normal CFTR gene
Faulty CFTR gene
Child
Does not have CF
Two copies of normal CFTR genes
Child
"CF carrier"
One copy of faulty CFTR gene
Child
"CF carrier"
One copy of faulty CFTR gene
Child
Has CF
Two copies of faulty CFTR gene

Huntington's chorea

- HC causes cells of the brain to degenerate affecting a person's coordination. They become moody, depressed and memory is affected. Movements are clumsy + jerky, eventually all movement is lost and death follows.
- HC first affects sufferers in their 30s and 40s, by which time many have reproduced, unknowingly passing on the gene. H is dominant to h and so only one dominant allele is necessary to cause the disease.
- hh = Normal
- Hh and HH = HC
- Heterozygote X normal = 50%; 1 in 2; 1:1 chance of HC child

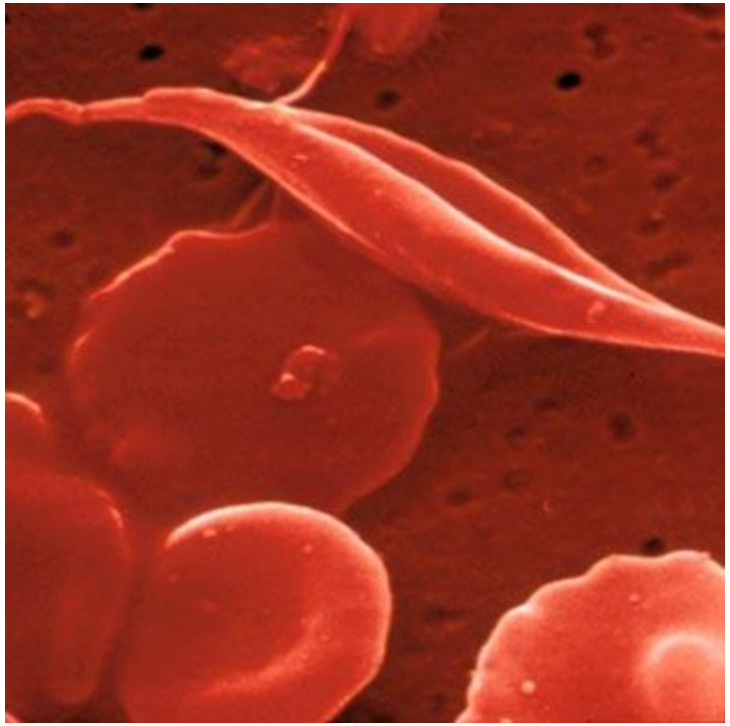

Frank's mother
Aunt
Aunt
Uncle
Frank's sister
Frank Jackowski
+ Rose
Dale
+ Cyndee
Cheryl
+ Bill Hinde
Danny
Kristy
Bradley
Kelly
Matthew
Amanda
affected
at risk
not affected

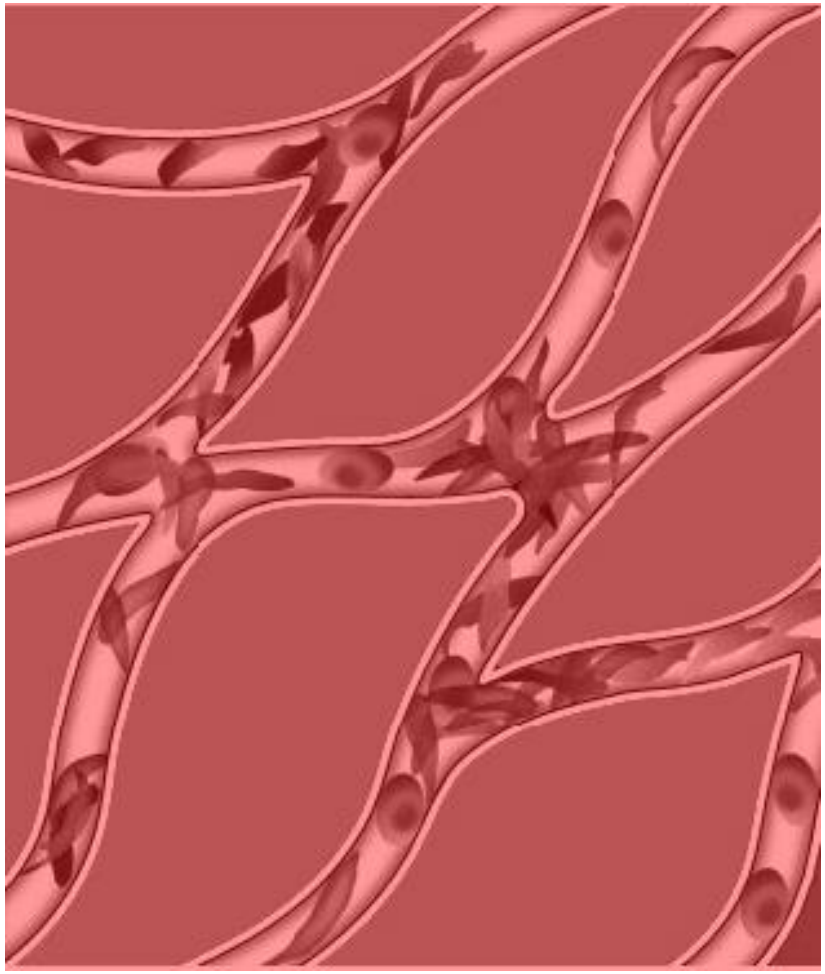


MRI of normal (non Huntington's) brain on the left compared to a Huntingon's brain on the right. Note the loss of brain mass (larger light areas in the Huntinton's brain)

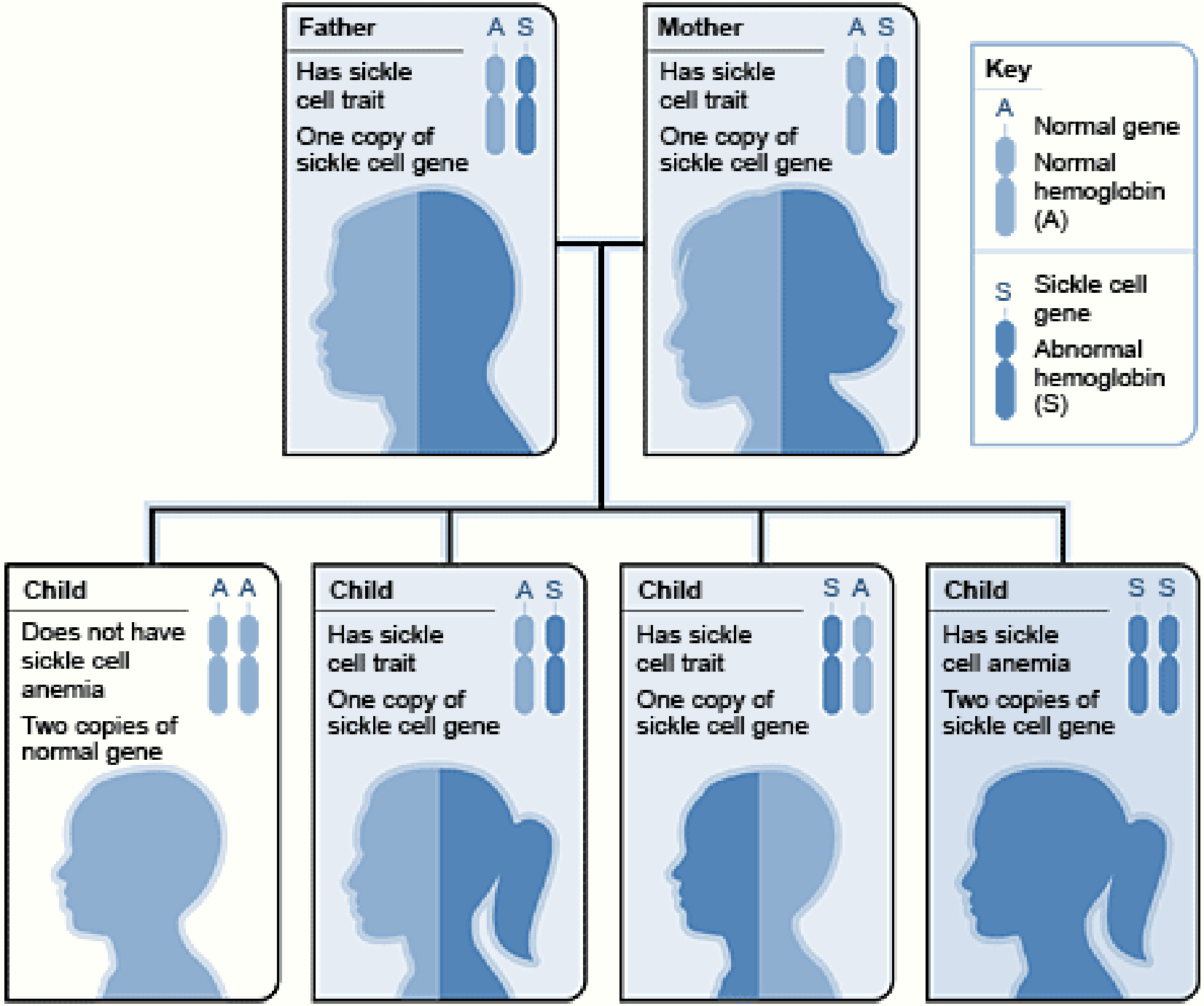
Sickle Cell Anaemia

- SCA is a blood disease which causes RBCs to become crescent shaped when O_2 levels are low. These cells then stick in capillaries blocking circulation and can cause early death.
- A heterozygote (carrier) has some normal and some sickle cells, so are not so badly affected.
- SS = Normal
- Ss = carrier (DO NOT suffer from SCA)
- ss = SCA
- carrier X carrier =
- 25% normal: 50% carrier: 25% SCA






Fatigue
Jaundice
Breathlessness
Back pain
Hand Swelling
Arm and
Leg pains
Joint pain
Foot swelling


Father
A S
Has sickle cell trait
One copy of sickle cell gene
Mother
A S
Has sickle cell trait
One copy of sickle cell gene
Key
A
Normal gene
Normal hemoglobin (A)
S
Sickle cell gene
Abnormal hemoglobin (S)
Child
A A
Does not have sickle cell anemia
Two copies of normal gene
Child
A S
Has sickle cell trait
One copy of sickle cell gene
Child
S A
Has sickle cell trait
One copy of sickle cell gene
Child
S S
Has sickle cell anemia
Two copies of sickle cell gene

NOTE

- Straight recessive/dominant alleles give 2 phenotypes : dominant and recessive
- Incomplete + co-dominant give 3 phenotypes ALLWAYS LOOK AT THE HETEROZYGOTES:
  - Co-dom 2 proteins will be produced (e.g. roan = red AND white)
  - Incomplete ONLY one NEW protein produced (e.g. R+W = pink)

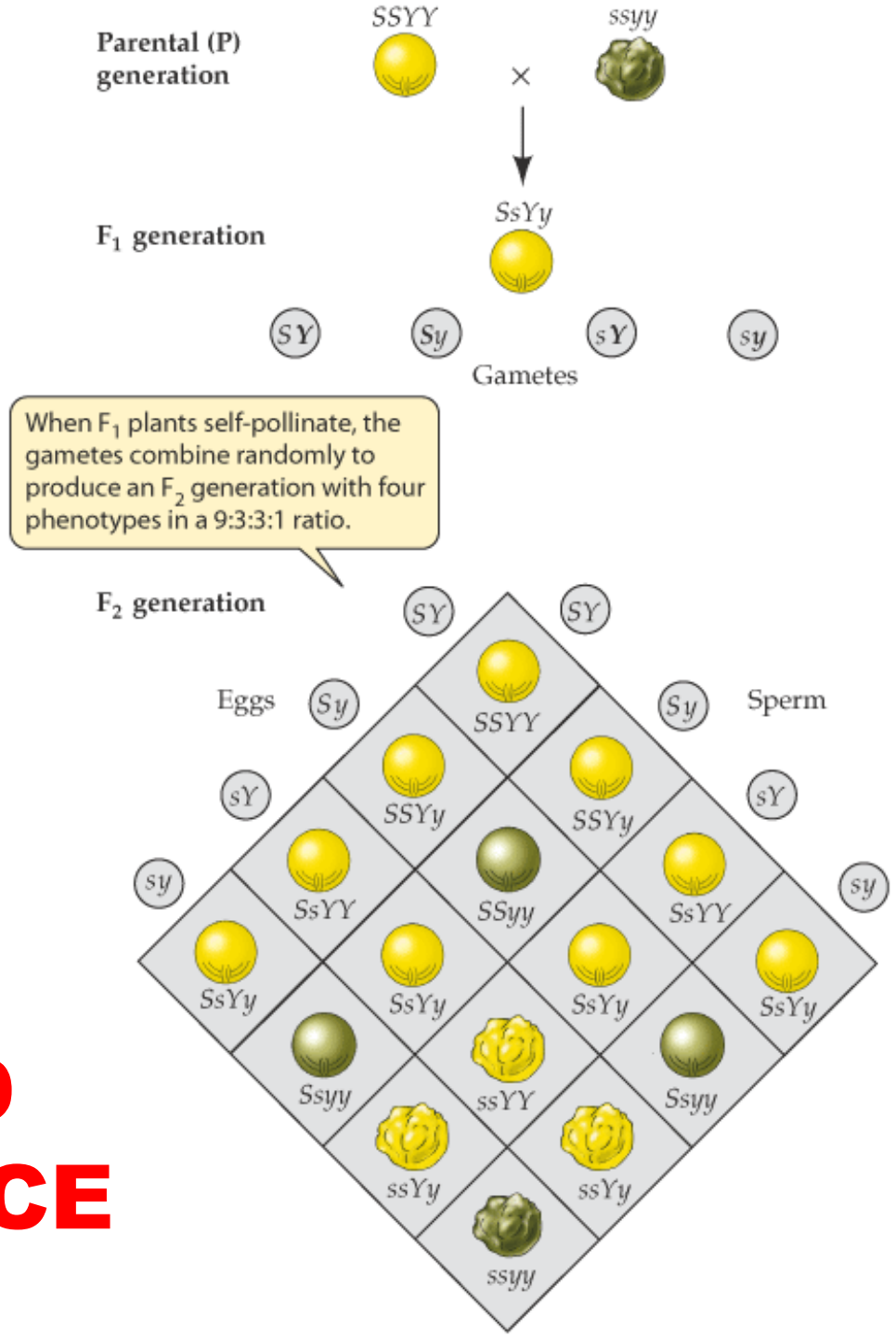
ANSWERS

- CYSTIC FIBROSIS
  - Recessive allele
  - Simple monohybrid cross
- HUNTINGTON'S CHOREA
  - Dominant allele
  - Simple monohybrid cross
- SICKLE CELL ANAEMIA
  - Co-dominance

Mendel's
SECOND LAW OF INHERITANCE

The alleles of unlinked genes separate independently during meiosis

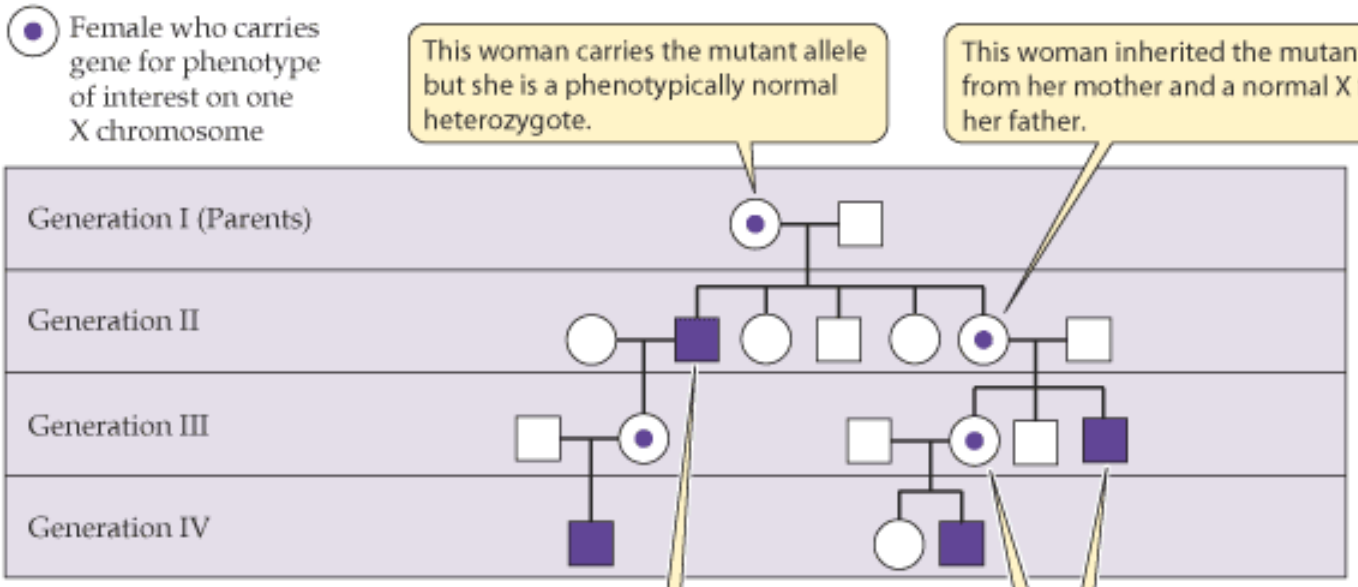
DIHYBRID INHERITANCE

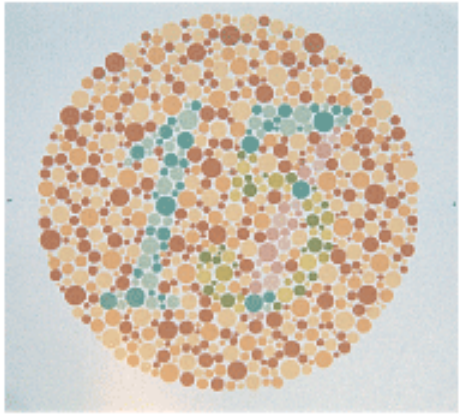


10.1 Mendel's Results from Monohybrid Crosses

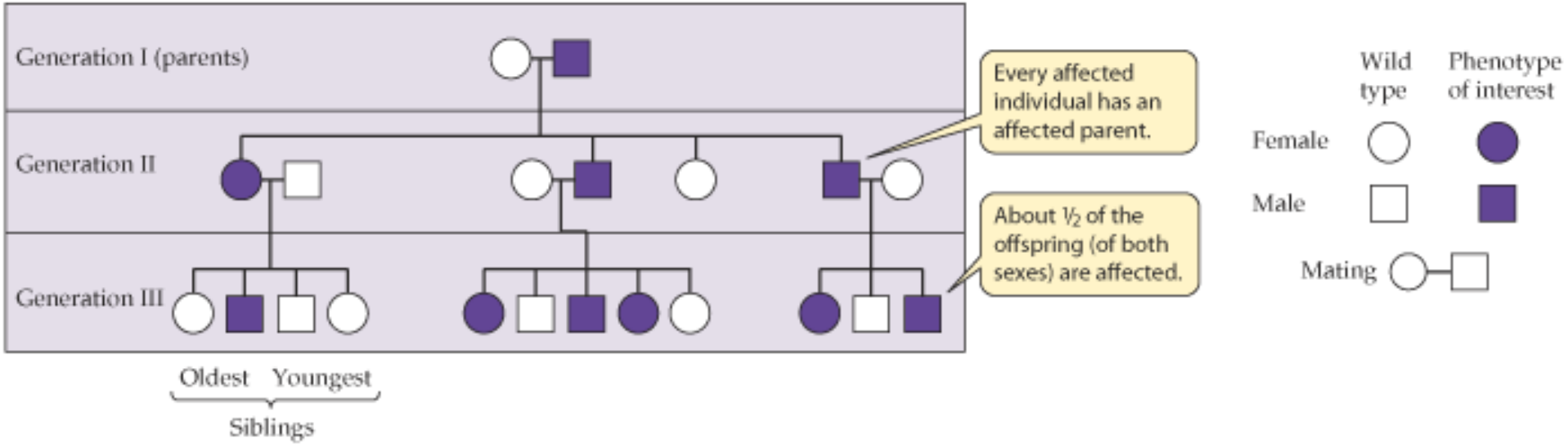
	PARENTAL GENERATION PHENOTYPES			F_2 GENERATION PHENOTYPES			
	DOMINANT	RECESSIVE		DOMINANT	RECESSIVE	TOTAL	RATIO
	Spherical seeds ×	Wrinkled seeds		5,474	1,850	7,324	2.96:1
	Yellow seeds ×	Green seeds		6,022	2,001	8,023	3.01:1
	Purple flowers ×	White flowers		705	224	929	3.15:1
	Inflated pods ×	Constricted pods		882	299	1,181	2.95:1
	Green pods ×	Yellow pods		428	152	580	2.82:1
	Axial flowers ×	Terminal flowers		651	207	858	3.14:1
	Tall stems (1 m) ×	Dwarf stems (0.3 m)		787	277	1,064	2.84:1

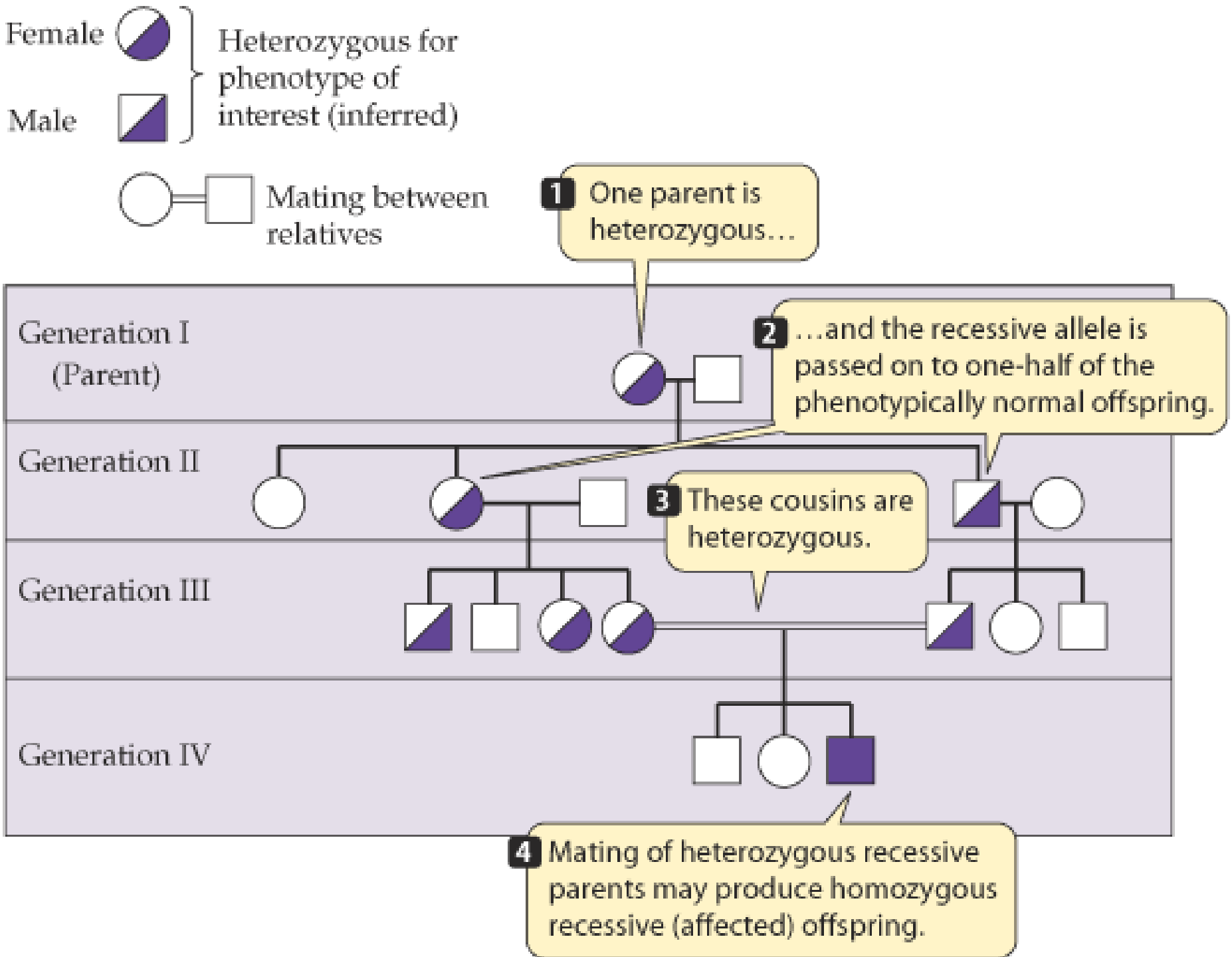
Possible genotypes	CC, Cc^{ch}, Cc^{h}, Cc	$c^{ch}c^{ch}$	$c^{ch}c^{h}$, $c^{ch}c$	$c^{h}c^{h}$, $c^{h}c$	cc
Phenotype	Dark gray	Chinchilla	Light gray	Himalayan	Albino





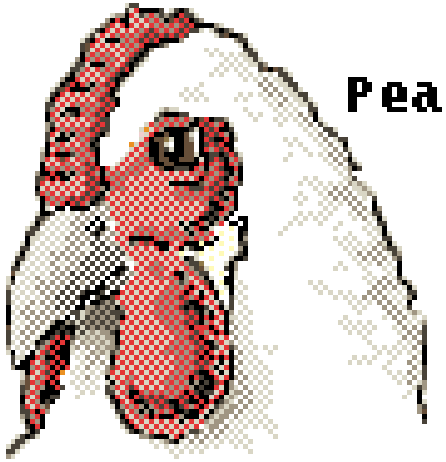
In this test for red-green color blindness, people with normal color vision will see the number 15.

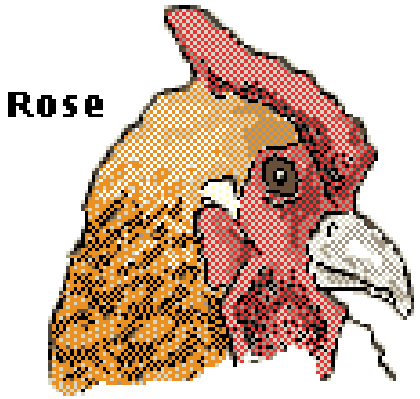

Generation I (parents)
Generation II
Generation III
Every affected individual has an affected parent.
About ½ of the offspring (of both sexes) are affected.
Wild type
Phenotype of interest
Female
Male
Mating
Oldest
Youngest
Siblings

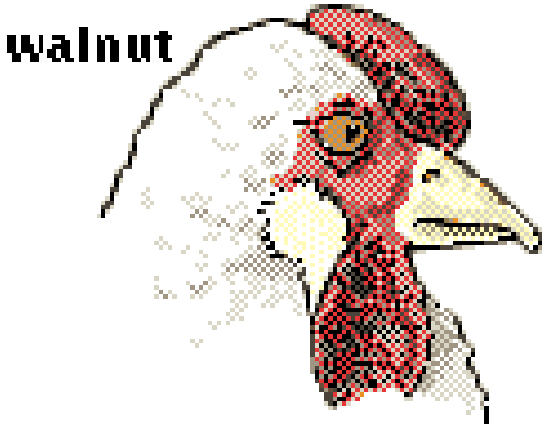

Female
Male
Heterozygous for phenotype of interest (inferred)
Mating between relatives
1 One parent is heterozygous…
2 …and the recessive allele is passed on to one-half of the phenotypically normal offspring.
3 These cousins are heterozygous.
4 Mating of heterozygous recessive parents may produce homozygous recessive (affected) offspring.
Generation I (Parent)
Generation II
Generation III
Generation IV

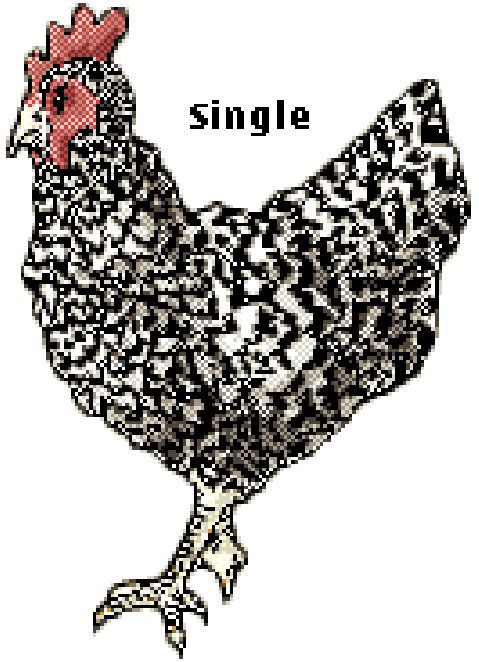
GENE INTERACTION AND EPISTASIS

- The shape of the comb of farmyard fowl is an example of a single character determined by two different genes.
- The product of these interactions are 4 possible phenotypes


Pea


Rose


walnut


Single


Rose comb


Pea comb


Walnut comb


Single comb

The genes involved, represented by the letters P and R, interact to form phenotypes:-

Pea: PPrr or Pprr

Rose: ppRR or ppRr

Walnut: PPRR or PpRR or PPRr or PpRr

Single: pprr

What do you notice about the alleles and phenotype?

- In other words, pea and rose combs are the product of dominant alleles for the character in question combined with a recessive allele for the other.
- Walnut is a product of the interaction of dominant alleles.
- Single is produced in the absence of dominant alleles.

Banding in Snails

- 2 genes interact to produce the pronounced banding in the shell of the snail *Cepaea nemoralis*.
- The dominant allele at one locus totally inhibiting the expression of an allele at a second locus.

- The banding exists in three common variants and is controlled by different genes on separate chromsomes.
- One gene determines whether or not bands are present. The allele for absence of bands (A) is dominant to the allele for presence (a).

- A second gene determines the number and position of the bands. A dominant allele codes for a 'mid-banded' condition.
- A recessive allele codes for a 'five-banded condition.
- The banded characteristic will be expressed when the genotype for banding is aa.

Interaction of genetic and environmental factors resulting in the phenotype

genotype

protein

hormones

enzymes

structural

environment

temperature

food availability

disease

other organisms

phenotype

Both genotype and environmental factors affect an organisms phenotype, resulting in variation between members of a population.

In Himalayan rabbits the genotype and environment give rise to fur colour. It has white fur over most of its body but black fur on its nose, ears, tail and feet.

The rabbit carries a gene for black fur, but the enzyme it makes is inactive at temperatures greater than 30°C therefore areas of the body that are above 30°C have white fur (no colour/albino). Only the nose, ears, tail and feet are cold enough for the enzyme to work.