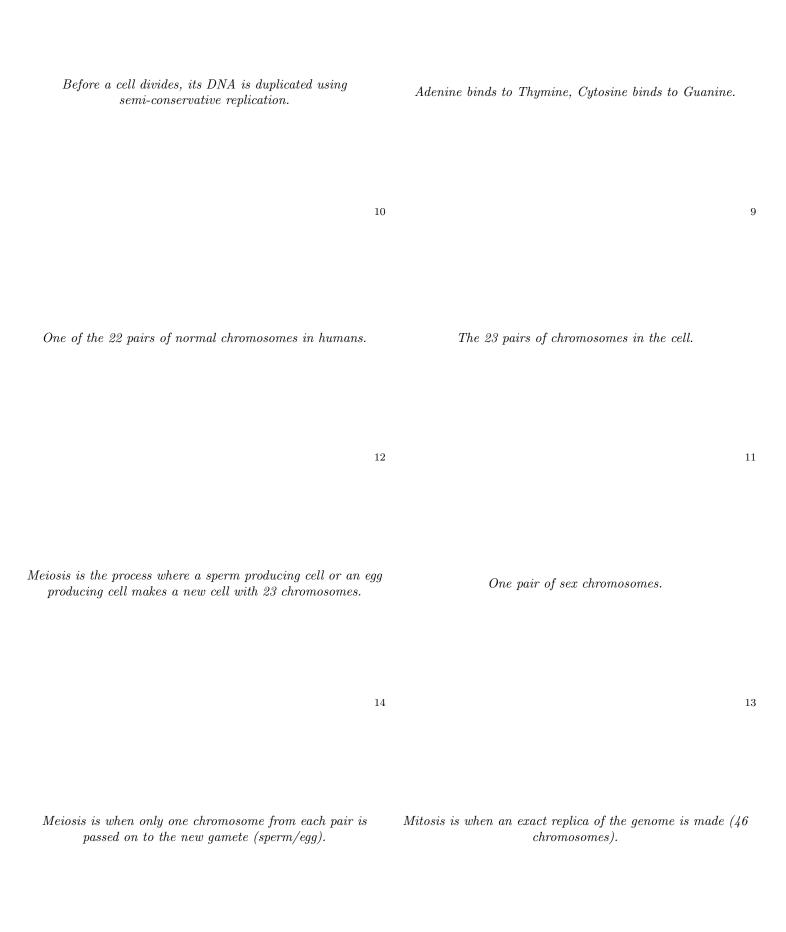
$Each\ DNA\ molecule\ is\ packed\ into\ a$.	$contain\ instructions\ for\ making$
The two strands of DNA twist to form a $oxedsymbol{1}$.	When replicating, the between the DNA strands break, and come to bind with the exposed ones on the separated strands to form new strands.
Proteins act alone or in functions. to perform many cellular	The four DNA bases are 6
A backbone provides structure for the DNA .	bonds hold the two strands of DNA together.

Genes contain instructions for making proteins.	Each DNA molecule is packed into a chromosome.
	2 1
When replicating, the hydrogen bonds between the DNA strands break, and new bases come to bind with the exposed ones on the separated strands to form new strands.	The two strands of DNA twist to form a double helix.
	4 3
Adenine, Thymine, Guanine, Cytosine	Proteins act alone or in complexes to perform many cellular functions.
	6 5
$Hydrogen\ bonds\ hold\ the\ two\ strands\ of\ DNA\ together.$	$A \ sugar-phosphate \ backbone \ provides \ structure \ for \ the \ DNA.$

binds to binds to	Before a cell divides, its DNA is duplicated using ${}^{\circ}$.
$What \ is \ the \ Karyotype?$	$What \ is \ an \ autosome?$
In addition to the autosomes, what other chromosomes are there?	is the process where a sperm producing cell or an egg producing cell makes a new cell with 23 chromosomes.
is when an exact replica of the genome is made (46 chromosomes).	is when only one chromosome from each pair is passed on to the new (sperm/egg).



$DNA \longrightarrow RNA \longrightarrow protein$	When a gene is, it forms many
molecules get into proteins.	Define an allele
Define polymorphism (in the context of DNA)	is when a person has two copies of one allele on a gene locus.
is when a person has two different alleles on a gene locus.	A gene is if the protein that it produces can be compensated for by the correct protein produced by



A gene is recessive if the mutated protein that it produces can

be compensated for by the correct protein produced by an

 $alternative \ allele.$

24

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Heterozygous is when a person has two different alleles on a

gene locus.

If a mutated gene produces proteins that fulfil a new function, then it may be sometimes, since the original function will be fulfilled by .	Genes can be, or
Define genotype.	Define phenotype
The phenotype is controlled by derived from , and the	What bloodgroup is made from two co-dominant alleles?
Blood groups: $\begin{array}{c ccccccccccccccccccccccccccccccccccc$	Allele frequency is linked to its in a given.

Genes can be recessive, dominant or co-dominant.

If a mutated gene produces proteins that fulfil a new function, then it may be co-dominant, since the original function will be fulfilled by the other allele.

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The physical appearance of an individual, including its observable or measurable traits.

The genetic make-up of an individual, which includes the genes or alleles present in it.

28

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AB

The phenotype is controlled by proteins derived from genes, and the environment.

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Allele frequency is linked to the fitness it provides to its carriers in a given environment.

$$Blood\ groups: \begin{array}{c|cccc} & I^A & I^B & i \\ \hline I^A & A & AB & A \\ I^B & AB & B & B \\ i & A & B & O \\ \end{array}$$

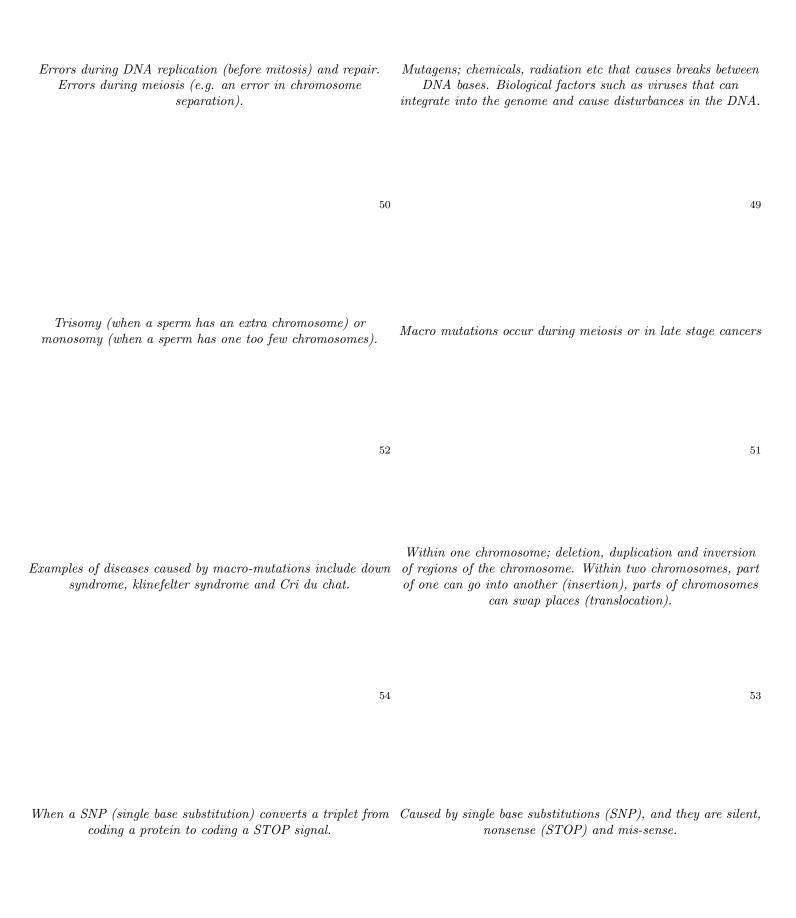
Define genetic fitness	If an allele provides , it is likely to and become in a given population.
Mutations have allowed us to our diet. This includes a mutation that lets us produce during adulthood (to drink milk) and another one that reduces the function of a allowing us to eat broccoli and sprouts! This is an example of .	Carriers of alleles are and get protection from malaria.
Carriers of alleles die if they are since their haemoglobin does not function well.	People for a mutation affecting are asymptomatic and immune to HIV. Probably because this gave protection against and in the past. This mutation is less effective against pathogens from .
Environment interaction can influence the genotype. and are sensitive to temperature, and change colour at different temperatures. This is caused by temperature sensitive.	The environment affects the phenotype; a can make a human twin grow to be smaller, and flowers have based on the soil.



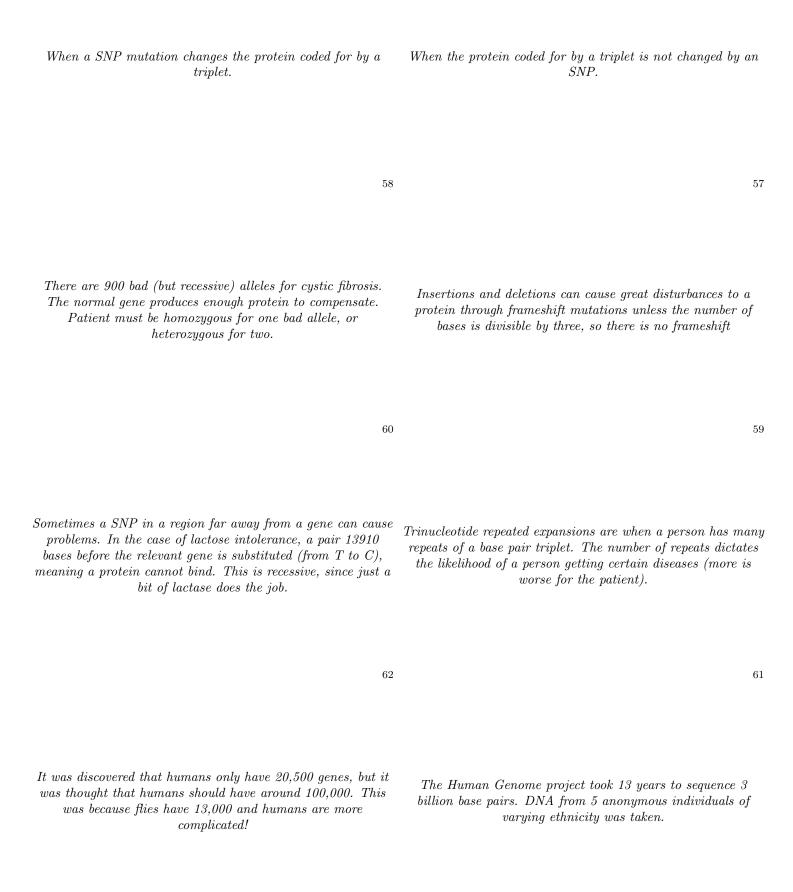
Most are due to several genes and the environment (e.g. , , , ,).	A greater similarity between for a particular compared to provides evidence that factors play a role.
twins share all their genes and their home environment. twins share their genes and a home environment.	Defineamutation
The size of mutations ranges from (- SNP) to	SNP mutations are, chromosome rearrangements are
Define a hereditary mutation.	Define an acquired (somatic) mutation.



$Environmental\ factors\ that\ cause\ mutations\ include$	Intrinsic factors causing mutations include
Macro mutations occur during or in	Mutations during meiosis include 52
$Single\ chromosome\ macro-mutations\ include$	Examples of diseases caused by macro-mutations include and .
33	04
What are the three types of substitution micro-mutations and what are they caused by?	How does a nonsense mutation occur?
55	56



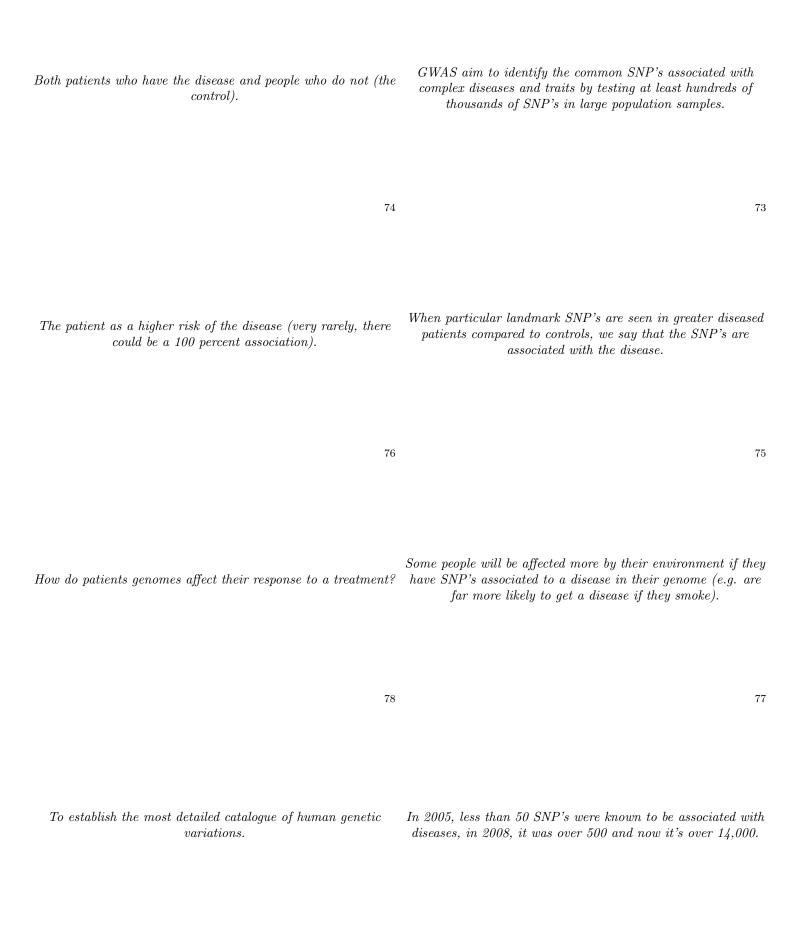
What is a silent mutation? 57	What is a mis-sense mutation? 58
can cause great disturbances to a protein through unless the number of bases , so there is no	There are bad (but) alleles for cystic fibrosis. The normal gene . Patient must be for one bad allele, or for two.
are when a person has many repeats of a base pair triplet. dictates the likelihood of a person getting certain diseases (more is worse for the patient).	Sometimes a SNP in a region far away from a gene can cause problems. In the case of lactose intolerance, a pair 13910 bases before the relevant gene is substituted (from T to C), meaning a protein cannot bind. This is recessive, since just a bit of lactase does the job.
The Human Genome project took to sequence base pairs. DNA from individuals of was taken.	It was discovered that humans only have genes, but it was thought that humans should have around . This was because flies have and humans are more complicated!



Humans share of their genes with flies, and only of the human DNA codes for genes.	Why can humans get by with so few genes?
Cells have the, but do not express the Where these are expressed determines the type of cell formed.	Humans genomes differ by about , which is about base pairs which are mostly
The frequency of SNP's is one in every base pairs. Most are and have	SNP's outside of genes are useful because
$GWAS\ stands\ for$	Most diseases result from , patients with have been found to be more at risk of developing some diseases.



GWAS aim to identify the common SNP's associated with by testing at least of SNP's in large population samples.	Where are the samples for GWAS taken from
73	74
When particular landmark SNP's are seen in greater diseased patients compared to controls, we say that the SNP's are with the disease.	If a patient has SNP's associated with a disease, what does it mean?
Some people will be affected more by if they have SNP's associated to a disease in their genome (e.g. are far more likely to get a disease if they smoke).	$What \ is \ pharmacogenomics?$
In 2005, SNP's were known to be associated with diseases, in 2008, it was and now it's over.	What was the aim of the 1000 genomes project?



On average, each person carries loss of function variants in annotated genes, and previously implicated in inherited disorders.	How many new disease causing mutations were identified in the 1000 genomes project?
In the 100,000 genomes project was started by It was split between helping and	The 100,000 genomes project sampled people including serious illness patients. cancer patient genomes (one cancer and one normal per patient), and rare disease genomes (three per patient;
and both let you get your genome sequenced. does not offer much advice or counselling, but does, and is therefore more expensive.	Immlumina tests healthy adults interested in learning about their risk for , assessing their status and understanding their response to certain .



On average, each person carries 250-300 loss of function variants in annotated genes, and 50-100 previously implicated in inherited disorders.

82

81

The 100,000 genomes project sampled 75,000 people including 40,000 serious illness patients. 50,000 cancer patient genomes (one cancer and one normal per patient), and 50,000 rare disease genomes (three per patient; one patient genome and two blood relatives))

In 2014 the 100,000 genomes project was started by the NHS. It was split between helping cancer patients and patients with rare diseases.

84

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Immlumina tests healthy adults interested in learning about their risk for a set of adult-onset conditions, assessing their carrier status and understanding their response to certain drugs.

23andMe and Illumina both let you get your genome sequenced. 23andMe does not offer much advice or counselling, but illumina does, and is therefore more expensive.