Each DNA molecule is packed into a [redacted].

1

[redacted] *contain instructions for making* [redacted].

2

The two strands of DNA twist to form a [redacted].

3

When replicating, the [redacted] *between the DNA strands break, and* [redacted] *come to bind with the exposed ones on the separated strands to form new strands.*

4

Proteins act alone or in [redacted] *to perform many cellular functions.*

5

The four DNA bases are...

6

A [redacted] *backbone provides structure for the DNA.*

7

[redacted] *bonds hold the two strands of DNA together.*

8

Genes contain instructions for making proteins.

2

Each DNA molecule is packed into a chromosome.

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.

4

The two strands of DNA twist to form a double helix.

3

Adenine, Thymine, Guanine, Cytosine

6

Proteins act alone or in complexes to perform many cellular functions.

5

Hydrogen bonds hold the two strands of DNA together.

8

A sugar-phosphate backbone provides structure for the DNA.

7

binds to , *binds to* .

9

Before a cell divides, its DNA is duplicated using .

10

What is the Karyotype?

11

What is an autosome?

12

In addition to the autosomes, what other chromosomes are there?

13

is the process where a sperm producing cell or an egg producing cell makes a new cell with 23 chromosomes.

14

is when an exact replica of the genome is made (46 chromosomes).

15

is when only one chromosome from each pair is passed on to the new (sperm/egg).

16

Before a cell divides, its DNA is duplicated using semi-conservative replication.

10

Adenine binds to Thymine, Cytosine binds to Guanine.

9

One of the 22 pairs of normal chromosomes in humans.

12

The 23 pairs of chromosomes in the cell.

11

Meiosis is the process where a sperm producing cell or an egg producing cell makes a new cell with 23 chromosomes.

14

One pair of sex chromosomes.

13

Meiosis is when only one chromosome from each pair is passed on to the new gamete (sperm/egg).

16

Mitosis is when an exact replica of the genome is made (46 chromosomes).

15

DNA [redacted] *RNA* [redacted] *protein*

17

When a gene is [redacted]*, it forms many* [redacted] *molecules.*

18

[redacted] *molecules get* [redacted] *into proteins.*

19

Define an allele

20

Define polymorphism (in the context of DNA)

21

[redacted] *is when a person has two copies of one allele on a gene locus.*

22

[redacted] *is when a person has two different alleles on a gene locus.*

23

A gene is [redacted] *if the* [redacted] *protein that it produces can be compensated for by the correct protein produced by* [redacted]*.*

24

When a gene is transcribed, it forms many RNA molecules.

18

$DNA \xrightarrow{transcription} RNA \xrightarrow{translation} protein$

17

Any of several forms of a gene, usually arising through mutation. Alleles are responsible for hereditary variation.

20

RNA molecules get translated into proteins.

19

Homozygous is when a person has two copies of one allele on a gene locus.

22

The existence of several alleles for one gene locus. Individuals have one or two alleles per locus.

21

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

Heterozygous is when a person has two different alleles on a gene locus.

23

If a mutated gene produces proteins that fulfil a new function, then it may be [redacted]*, since the original function will be fulfilled by* [redacted].

25

Genes can be [redacted]*,* [redacted] *or* [redacted].

26

Define genotype.

27

Define phenotype

28

The phenotype is controlled by [redacted] *derived from* [redacted]*, and the* [redacted].

29

What bloodgroup is made from two co-dominant alleles?

30

Blood groups:

	I^A	I^B	i
I^A	[redacted]	[redacted]	[redacted]
I^B	[redacted]	[redacted]	[redacted]
i	[redacted]	[redacted]	[redacted]

31

Allele frequency is linked to [redacted] *to its* [redacted] *in a given* [redacted].

32

Genes can be recessive, dominant or co-dominant.

26

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.

25

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

28

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

27

AB

30

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

29

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

32

Blood groups:

	I^A	I^B	i
I^A	A	AB	A
I^B	AB	B	B
i	A	B	O

31

Define genetic fitness

33

If an allele provides [redacted] *, it is likely to* [redacted] *and become* [redacted] *in a given population.*

34

Mutations have allowed us to [redacted] *our diet. This includes a mutation that lets us produce* [redacted] *during adulthood (to drink milk) and another one that reduces the function of a* [redacted] *allowing us to eat broccoli and sprouts! This is an example of* [redacted] *.*

35

Carriers of [redacted] *alleles are* [redacted] *and get protection from malaria.*

36

Carriers of [redacted] *alleles die if they are* [redacted] *since their haemoglobin does not function well.*

37

People [redacted] *for a mutation affecting* [redacted] *are asymptomatic and immune to HIV. Probably because this gave protection against* [redacted] *and* [redacted] *in the past. This mutation is less effective against pathogens from* [redacted] *.*

38

Environment interaction can influence the genotype. [redacted] *and* [redacted] *are sensitive to temperature, and change colour at different temperatures. This is caused by temperature sensitive* [redacted] *.*

39

The environment affects the phenotype; a [redacted] *can make a human twin grow to be smaller, and flowers have* [redacted] *based on the soil* [redacted] *.*

40

If an allele provides an advantage, it is likely to persist and become more prominent in a given population.

34

The reproductive success of a genotype, measured as the number of offspring produced by and individual that survive to a reproductive age relative to the average age for the population.

33

Carriers of sickle cell anaemia alleles are asymptomatic and get protection from malaria.

36

Mutations have allowed us to diversify our diet. This includes a mutation that lets us produce lactase during adulthood (to drink milk) and another one that reduces the function of a bitter substance taste receptor allowing us to eat broccoli and sprouts! This is an example of natural selection.

35

People homozygous for a mutation affecting CCR5 are asymptomatic and immune to HIV. Probably because this gave protection against the plague and smallpox in the past. This mutation is less effective against pathogens from developing countries.

38

Carriers of sickle cell anaemia alleles die if they are homozygous since their haemoglobin does not function well.

37

The environment affects the phenotype; a worse diet can make a human twin grow to be smaller, and flowers have different colours based on the soil pH.

40

Environment interaction can influence the genotype. Himalayan rabbits and arctic foxes are sensitive to temperature, and change colour at different temperatures. This is caused by temperature sensitive tyrosine.

39

Most [redacted] *are due to several genes and the environment (e.g.* [redacted]*,* [redacted]*,* [redacted]*).*

41

A greater similarity between [redacted] *for a particular* [redacted] *compared to* [redacted] *provides evidence that* [redacted] *factors play a role.*

42

[redacted] *twins share all their genes and their home environment.* [redacted] *twins share* [redacted] *their genes and a home environment.*

43

Define a mutation

44

The size of mutations ranges from [redacted] *(*[redacted] *- SNP) to* [redacted] *(*[redacted]*)*

45

SNP mutations are [redacted]*, chromosome rearrangements are* [redacted]

46

Define a hereditary mutation.

47

Define an acquired (somatic) mutation.

48

A greater similarity between identical twins for a particular trait compared to fraternal twins provides evidence that genetic factors play a role.

42

Most phenotypes are due to several genes and the environment (e.g. skin colour, height, weight).

41

*A **permanent** alteration in the DNA sequence passed on into daughter cells (and sometimes gametes).*

44

Identical twins share all their genes and their home environment. Fraternal twins share half their genes and a home environment.

43

SNP mutations are micro-mutations, chromosome rearrangements are macro-mutations

46

The size of mutations ranges from a single base pair (single nucleotide polymorphism - SNP) to large segments of a chromosome (chromosome rearrangement)

45

When a mutation occurs at some point in a person's life, and is present only in the cell that it occurred and it's daughter cells (through mitosis).

48

A mutation inherited from a parent gamete and present throughout a person's life and in every cell in their body. This can be passed on to progeny through meiosis.

47

Environmental factors that cause mutations include...

49

Intrinsic factors causing mutations include...

50

Macro mutations occur during [redacted] *or in* [redacted]

51

Mutations during meiosis include...

52

Single chromosome macro-mutations include...

53

Examples of diseases caused by macro-mutations include [redacted], [redacted] *and* [redacted].

54

What are the three types of substitution micro-mutations and what are they caused by?

55

How does a nonsense mutation occur?

56

Errors during DNA replication (before mitosis) and repair. Errors during meiosis (e.g. an error in chromosome separation).

50

Mutagens; chemicals, radiation etc that causes breaks between DNA bases. Biological factors such as viruses that can integrate into the genome and cause disturbances in the DNA.

49

Trisomy (when a sperm has an extra chromosome) or monosomy (when a sperm has one too few chromosomes).

52

Macro mutations occur during meiosis or in late stage cancers

51

Examples of diseases caused by macro-mutations include down syndrome, klinefelter syndrome and Cri du chat.

54

Within one chromosome; deletion, duplication and inversion of regions of the chromosome. Within two chromosomes, part of one can go into another (insertion), parts of chromosomes can swap places (translocation).

53

When a SNP (single base substitution) converts a triplet from coding a protein to coding a STOP signal.

56

Caused by single base substitutions (SNP), and they are silent, nonsense (STOP) and mis-sense.

55

What is a silent mutation?

57

What is a mis-sense mutation?

58

[redacted] *can cause great disturbances to a protein through* [redacted] *unless the number of bases* [redacted]*, so there is no* [redacted]

59

There are [redacted] *bad (but* [redacted]*) alleles for cystic fibrosis. The normal gene* [redacted]*. Patient must be* [redacted] *for one bad allele, or* [redacted] *for two.*

60

[redacted] *are when a person has many repeats of a base pair triplet.* [redacted] *dictates the likelihood of a person getting certain diseases (more is worse for the patient).*

61

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.

62

The Human Genome project took [redacted] *to sequence* [redacted] *base pairs. DNA from* [redacted] *individuals of* [redacted] *was taken.*

63

It was discovered that humans only have [redacted] *genes, but it was thought that humans should have around* [redacted]*. This was because flies have* [redacted] *and humans are more complicated!*

64

When a SNP mutation changes the protein coded for by a triplet.

58

When the protein coded for by a triplet is not changed by an SNP.

57

There are 900 bad (but recessive) alleles for cystic fibrosis. The normal gene produces enough protein to compensate. Patient must be homozygous for one bad allele, or heterozygous for two.

60

Insertions and deletions can cause great disturbances to a protein through frameshift mutations unless the number of bases is divisible by three, so there is no frameshift

59

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.

62

Trinucleotide repeated expansions are when a person has many repeats of a base pair triplet. The number of repeats dictates the likelihood of a person getting certain diseases (more is worse for the patient).

61

It was discovered that humans only have 20,500 genes, but it was thought that humans should have around 100,000. This was because flies have 13,000 and humans are more complicated!

64

The Human Genome project took 13 years to sequence 3 billion base pairs. DNA from 5 anonymous individuals of varying ethnicity was taken.

63

Humans share [redacted] *of their genes with flies, and only* [redacted] *of the human DNA codes for genes.*

65

Why can humans get by with so few genes?

66

Cells have the [redacted]*, but do not express the* [redacted]*. Where these* [redacted] *are expressed determines the type of cell formed.*

67

Humans genomes differ by about [redacted]*, which is about* [redacted] *base pairs which are mostly* [redacted]

68

The frequency of SNP's is one in every [redacted] *base pairs. Most are* [redacted] *and have* [redacted]*.*

69

SNP's outside of genes are useful because...

70

GWAS stands for...

71

Most diseases result from [redacted]*, patients with* [redacted] *have been found to be more at risk of developing some diseases.*

72

Humans share sixty percent of their genes with flies, and only two percent of the human DNA codes for genes.

65

Alternative splicing; the same gene can produce different proteins when it is shaped differently (isoforms). This means that we can make 100k proteins with 23k genes.

66

Cells have the same genome, but do not express the same genes and isoforms. Where these proteins are expressed determines the type of cell formed.

67

Humans genomes differ by about 0.01 percent, which is about 3 million base pairs which are mostly SNP's

68

The frequency of SNP's is one in every 300 base pairs. Most are outside genes and have no effect on the phenotype.

69

They act as landmarks for us as scientists!

70

Genome wide association studies

71

Most diseases result from polygenic and environmental interactions, patients with particular groups of landmark SNP's have been found to be more at risk of developing some diseases.

72

GWAS aim to identify the common SNP's associated with [redacted] *by testing at least* [redacted] *of SNP's in large population samples.*

73

Where are the samples for GWAS taken from

74

When particular landmark SNP's are seen in greater diseased patients compared to controls, we say that the SNP's are [redacted] *with the disease.*

75

If a patient has SNP's associated with a disease, what does it mean?

76

Some people will be affected more by [redacted] *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).*

77

What is pharmacogenomics?

78

In 2005, [redacted] *SNP's were known to be associated with diseases, in 2008, it was* [redacted] *and now it's over* [redacted].

79

What was the aim of the 1000 genomes project?

80

GWAS aim to identify the common SNP's associated with complex diseases and traits by testing at least hundreds of thousands of SNP's in large population samples.

73

Both patients who have the disease and people who do not (the control).

74

When particular landmark SNP's are seen in greater diseased patients compared to controls, we say that the SNP's are associated with the disease.

75

The patient as a higher risk of the disease (very rarely, there could be a 100 percent association).

76

Some people will be affected more by their environment 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).

77

How do patients genomes affect their response to a treatment?

78

In 2005, less than 50 SNP's were known to be associated with diseases, in 2008, it was over 500 and now it's over 14,000.

79

To establish the most detailed catalogue of human genetic variations.

80

On average, each person carries [redacted] *loss of function variants in annotated genes, and* [redacted] *previously implicated in inherited disorders.*

81

How many new disease causing mutations were identified in the 1000 genomes project?

82

In [redacted] *the 100,000 genomes project was started by* [redacted]*. It was split between helping* [redacted] *and* [redacted]*.*

83

The 100,000 genomes project sampled [redacted] *people including* [redacted] *serious illness patients.* [redacted] *cancer patient genomes (one cancer and one normal per patient), and* [redacted] *rare disease genomes (three per patient;* [redacted]*)*

84

[redacted] *and* [redacted] *both let you get your genome sequenced.* [redacted] *does not offer much advice or counselling, but* [redacted] *does, and is therefore more expensive.*

85

Immlumina tests healthy adults interested in learning about their risk for [redacted]*, assessing their* [redacted] *status and understanding their response to certain* [redacted]*.*

86

671

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

83

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.

86

85