

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.

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.

8

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 → RNA → protein

17

When a gene is , it forms many molecules.

18

molecules get into proteins.

19

Define an allele

20

Define polymorphism (in the context of DNA)

21

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

22

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

23

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

24

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

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

18

17

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

RNA molecules get translated into proteins.

20

19

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

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

22

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.

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

24

23

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

25

Genes can be , or .

26

Define genotype.

27

Define phenotype

28

The phenotype is controlled by derived from , and the .

29

What bloodgroup is made from two co-dominant alleles?

30

Blood groups:

	I^A	I^B	i
I^A	<input type="text"/>	<input type="text"/>	<input type="text"/>
I^B	<input type="text"/>	<input type="text"/>	<input type="text"/>
i	<input type="text"/>	<input type="text"/>	<input type="text"/>

31

Allele frequency is linked to to its in a given .

32

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.

26

25

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

27

AB

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

30

29

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

Blood groups:

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

32

31

Define genetic fitness

33

If an allele provides _____, it is likely to _____ and become _____ in a given population.

34

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 _____.

35

Carriers of _____ alleles are _____ and get protection from malaria.

36

Carriers of _____ alleles die if they are _____ since their haemoglobin does not function well.

37

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 _____.

38

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

39

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

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 its 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

<p><i>Environmental factors that cause mutations include...</i></p>	<p><i>Intrinsic factors causing mutations include...</i></p>
49	50
<p><i>Macro mutations occur during [redacted] or in [redacted]</i></p>	<p><i>Mutations during meiosis include...</i></p>
51	52
<p><i>Single chromosome macro-mutations include...</i></p>	<p><i>Examples of diseases caused by macro-mutations include [redacted], [redacted] and [redacted].</i></p>
53	54
<p><i>What are the three types of substitution micro-mutations and what are they caused by?</i></p>	<p><i>How does a nonsense mutation occur?</i></p>
55	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

_____ can cause great disturbances to a protein through _____ unless the number of bases _____, so there is no _____

59

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

60

_____ 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).

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 _____ to sequence _____ base pairs. DNA from _____ individuals of _____ was taken.

63

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!

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

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

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

65

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

68

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

They act as landmarks for us as scientists!

70

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

69

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

Genome wide association studies

71

<p><i>GWAS aim to identify the common SNP's associated with [REDACTED] by testing at least [REDACTED] of SNP's in large population samples.</i></p> <p>73</p>	<p><i>Where are the samples for GWAS taken from</i></p> <p>74</p>
<p><i>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.</i></p> <p>75</p>	<p><i>If a patient has SNP's associated with a disease, what does it mean?</i></p> <p>76</p>
<p><i>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).</i></p> <p>77</p>	<p><i>What is pharmacogenomics?</i></p> <p>78</p>
<p><i>In 2005, [REDACTED] SNP's were known to be associated with diseases, in 2008, it was [REDACTED] and now it's over [REDACTED].</i></p> <p>79</p>	<p><i>What was the aim of the 1000 genomes project?</i></p> <p>80</p>

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

74

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

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

76

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

How do patients genomes affect their response to a treatment?

78

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

To establish the most detailed catalogue of human genetic variations.

80

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

<p>On average, each person carries [REDACTED] loss of function variants in annotated genes, and [REDACTED] previously implicated in inherited disorders.</p> <p>81</p>	<p>How many new disease causing mutations were identified in the 1000 genomes project?</p> <p>82</p>
<p>In [REDACTED] the 100,000 genomes project was started by [REDACTED]. It was split between helping [REDACTED] and [REDACTED].</p> <p>83</p>	<p>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])</p> <p>84</p>
<p>[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.</p> <p>85</p>	<p>Immlumina tests healthy adults interested in learning about their risk for [REDACTED], assessing their [REDACTED] status and understanding their response to certain [REDACTED].</p> <p>86</p>
<p>How many different types of cell are there in humans?</p> <p>87</p>	<p>What is the first cell created by the fusion of the egg and sperm?</p> <p>88</p>

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

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.

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.

The zygote.

220 cell types.

What are the initial cells formed from the zygote called?

89

After there are more than 8 blastomeres, what is there?

90

What is the trophoblast?

91

Where does the embryo form from?

92

When the [redacted] is dividing, the cells become smaller since they are partitioning the [redacted] cytoplasm via mitosis.

93

What lets the embryo attach to the wall of the uterus?

94

[redacted] is driven by the [redacted]. The [redacted] expands and changes shape and location, but is still [redacted].

95

Once attached to the uterus wall, the inner cell mass sets the [redacted]. The [redacted] is the [redacted] axis.) The body is symmetrical along this.

96

A blastocyst a ball of cells.

Blastomeres

90

89

The inner cell mass, not the trophoblast.

The embryo after it was a blastocyst (5 days). Separate from the inner cell mass

92

91

The trophoblast

When the inner cell mass is dividing, the cells become smaller since they are partitioning the zygote cytoplasm via mitosis.

94

93

Once attached to the uterus wall, the inner cell mass sets the axis of the body. The primitive streak is the anterior posterior (head to tail) axis.) The body is symmetrical along this.

uterine implantation is driven by the trophoblast. The Inner cell mass expands and changes shape and location, but is still only one type of cell.

96

95

<p>After setting the axis, [redacted] takes place. This is where cells migrate, along the bottom, endoderm form, [redacted] in the middle [redacted] at the top. [redacted] will be the skin and nerves, [redacted] forms [redacted] and the [redacted] forms the [redacted]</p> <p>97</p>	<p>What is a highly coordinated cell movement?</p> <p>98</p>
<p>What structures become the vertebrae?</p> <p>99</p>	<p>What do somites eventually form into?</p> <p>100</p>
<p>Growing organs is called...</p> <p>101</p>	<p>By saying organogenesis is progressive, we mean</p> <p>102</p>
<p>What is used as a reference for growing specialised cells in an embryo?</p> <p>103</p>	<p>What is a differentiated cell?</p> <p>104</p>

Gastrulation

After setting the axis, gastrulation takes place. This is where cells migrate, along the bottom, endoderm form, mesoderm in the middle and ectoderm at the top. ectoderm will be the skin and nerves, mesoderm forms muscles, blood, skeleton, heart etc and the endoderm forms the digestive system, lungs etc

98

97

Muscles, vertebral column and dermis of the skin. They are landmarks for organ formation during development.

Somites; they emit signals telling what organs to form where.

100

99

That the organs grow in stages, e.g. there is a little growth for the arm first, then it gets longer, then it gets digits etc.

Organogenesis

102

101

One where the shape, structure and function is well defined.

The head to tail framework.

104

103

<p>The gurdon experiment was done on...</p>	<p>The gurdon experiment involves...</p>																												
105	106																												
<p>Cells developmental potential (potency) changes how as it gets more specialised?</p>	<p>What is involved in a grafting experiment?</p>																												
107	108																												
<p>The fate of a cell [redacted] before differentiation. They can sometimes [redacted] a new situation, up to [redacted].</p>	<table><tr><th>Source</th><th>Potential</th><th>Type of cell</th><th>Can develop into</th></tr><tr><td>Zygote</td><td>[redacted]</td><td>-</td><td>Whole organism.</td></tr><tr><td>[redacted]</td><td>[redacted] and self-renewing</td><td>Embryonic stem cell</td><td>Any cell type</td></tr><tr><td>Adult</td><td>Multipotent, [redacted]</td><td>[redacted] multipotent</td><td>Some cell types</td></tr><tr><td>Organ</td><td>Limited potential and renewal</td><td>[redacted]</td><td>Choice of between [redacted] types</td></tr><tr><td>-</td><td>Limited division</td><td>committed progenitor</td><td>1 type, locked fate.</td></tr><tr><td>-</td><td>No division</td><td>Differentiated</td><td>No division.</td></tr></table>	Source	Potential	Type of cell	Can develop into	Zygote	[redacted]	-	Whole organism.	[redacted]	[redacted] and self-renewing	Embryonic stem cell	Any cell type	Adult	Multipotent, [redacted]	[redacted] multipotent	Some cell types	Organ	Limited potential and renewal	[redacted]	Choice of between [redacted] types	-	Limited division	committed progenitor	1 type, locked fate.	-	No division	Differentiated	No division.
Source	Potential	Type of cell	Can develop into																										
Zygote	[redacted]	-	Whole organism.																										
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109	110																												
<p>Once a cell is differentiated...</p>	<p>Cells have the same genes, but it's how they express their genes that makes them different.</p>																												
111	112																												

Taking egg cells, removing the nuclei and inserting nuclei from either a small embryo or a developed intestine cell. The former usually develop into tadpoles, but the latter mostly stop developing before the tadpole stage.

Frogs

106

105

Cells from an early gastrula (early embryo) that would form an eye are taken and transplanted into an host embryo (oldest), as well as ones from an neurala (older embryo than gastrula). The ones from the younger embryo develop into anything depending where they are implanted, the ones from the older embryo develop into eyes.

It decreases.

108

107

Source	Potential	Type of cell	Can develop into
Zygote	Totipotent	-	Whole organism.
Blasocyst	Pluripotent and self-renewing	Embryonic stem cell	Any cell type
Adult	Multipotent, self-renewing	multipotent stem cells	Some cell types
Organ	Limited potential and renewal	Progenitor	Choice of between 2-6 types
-	Limited division	committed progenitor	1 type, locked fate.
-	No division	Differentiated	No division.

The fate of a cell can be locked before differentiation. They can sometimes not adapt to a new situation, up to 4 generations before.

110

109

Cells have the same genes, but it's how they express their genes that makes them different.

It has a clear cut identity and expresses specific proteins for morphology and function.

112

111

At any given time, each cell expresses around [REDACTED] of its genes

113

About [REDACTED] of the [REDACTED] active genes are developmental genes.

114

Developmental genes control:

115

One small difference in gene expression can [REDACTED].

116

Proteins inside the egg are [REDACTED].

117

After two [REDACTED] of the zygote (egg to two cells, to four), the [REDACTED] are in the cytoplasm. After division two, the cells have different maternal proteins after division, so they have different gene expressions and more differences occur after each cell division onwards.

118

A differentiated cell can give rise to a new organism ([REDACTED]), which means genes are [REDACTED] as a cell specialises.

119

[REDACTED] are proteins that bind to the DNA and [REDACTED]. They change how the DNA is shaped so that different parts can be accessed.

120

About ten percent of the 20 percent active genes are developmental genes.

114

At any given time, each cell expresses around 20 percent of its genes

113

One small difference in gene expression can create a cascade of changes downstream.

116

Proteins that regulate genes expression (turn genes on and off), proteins involved in cell communication or signalling (tell other cells what genes to turn on and off).

115

After two cleavage divisions of the zygote (egg to two cells, to four), the same maternal proteins are in the cytoplasm. After division two, the cells have different maternal proteins after division, so they have different gene expressions and more differences occur after each cell division onwards.

118

Proteins inside the egg are not uniformly distributed.

117

Epigenetics are proteins that bind to the DNA and retrieve totipotency. They change how the DNA is shaped so that different parts can be accessed.

120

A differentiated cell can give rise to a new organism (totipotent), which means genes are not lost as a cell specialises.

119

*The embryo starts with a zygote ([REDACTED] .
It becomes a [REDACTED] with a [REDACTED] and [REDACTED]
([REDACTED])*

*Before full differentiation, cells [REDACTED]
and are [REDACTED]*

At the gene level, cells become different by

The initial differences come from the

*[REDACTED] being unevenly distributed
in the [REDACTED] . As [REDACTED] form from
[REDACTED] , they end up not having the same
[REDACTED] .*

*The embryo starts with a zygote (totipotent.
It becomes a blastocyst with a trophoblast and ICM
(pluripotent)
Before full differentiation, cells become locked in their fate and
are determined
At the gene level, cells become different by expressing different
developmental genes
The initial differences come from the maternal developmental
proteins being unevenly distributed in the egg cytoplasm. As
blastomeres form from cleavage divisions, they end up not
having the same developmental proteins.*