

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

41

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

42

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

43

Define a mutation

44

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

45

SNP mutations are [ ], chromosome rearrangements are [ ]

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.																										
[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.																										
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

<p>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 [REDACTED] 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].</p>	<p>[REDACTED] stem cells can become any cell.</p>
<p>[REDACTED] have the minimum level of specialisation, [REDACTED] have the maximum level of specialisation.</p>	<p>[REDACTED] cells are not stem cells, but [REDACTED] cells are.</p>
<p>[REDACTED] cell stage is the limit for totipotency in humans.</p>	<p>Stem cells in the ICM are [REDACTED].</p>
<p>Adult stem cells are [REDACTED].</p>	<p>Adult stem cells are found in...</p>

121

122

123

124

125

126

127

128

*Totipotent stem cells can become any cell.*

122

*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.*

121

*Committed progenitor cells are not stem cells, but progenitor  
cells are.*

124

*totipotent stem cells have the minimum level of specialisation,  
differentiated cells have the maximum level of specialisation.*

123

*Stem cells in the ICM are pluripotent.*

*8 cell stage is the limit for totipotency in humans.*

126

125

*Brain, Skin, Bone Marrow, Skeletal muscle, Intestines (any  
cell that needs regrowth).*

*Adult stem cells are multipotent.*

128

127



<p>Embryonic stem cells are [redacted] and [redacted].</p>	<p>In order to control ESL's in vitro, we can [redacted] culture medium, or [redacted].</p>
129	130
<p>Adult stem cells are [redacted] to grow in the lab than [redacted] but do show [redacted].</p>	<p>Describe the plasticity of ASC's</p>
131	132
<p>What are the most apparently plastic cells?</p>	<p>Why are UC-MSc's better than BM-MSc's?</p>
133	134
<p>How many proteins are usually considered for immuno-compatibility?</p>	<p>What is GVHD?</p>
135	136

*In order to control ESL's in vitro, we can change the chemical composition of the culture medium, or insert specific genes into cells.*

130

*Embryonic stem cells are immortal and pluripotent.*

129

*Most adult stem cells can trans-differentiate in the lab, but this is a low efficiency process.*

132

*Adult stem cells are harder to grow in the lab than ESC's but do show some plasticity.*

131

- *Less immunogenic*
- *longer telomeres*
- *less DNA damage*
- *non-invasive to harvest*
- *same plasticity as BM-MSCs.*

134

*Mesenchymal stem cells (mesoderm), which can transform into liver cells (endoderm) and brain cells (ectoderm)*

133

*Graft Vs Host Disease, where the immune cells in the transplant attack the host.*

5

136

135

<p><i>Why are neonatal (UC cells) less immunogenic?</i></p> <p>137</p>	<p><i>Neonatal cells have longer [REDACTED] (which [REDACTED]), since they get shorter [REDACTED] since they do not get replicated, and neonatal cells have not divided many times.</i></p> <p>138</p>
<p><i>In ESC's what enzyme is expressed that stops a telomeres from getting shorter?</i></p> <p>139</p>	<p><i>When is telomerase turned off?</i></p> <p>140</p>
<p><i>What enzyme do most cancer cells produce and why?</i></p> <p>141</p>	<p><i>What is a bank of ESC lines?</i></p> <p>142</p>
<p><i>What are the three sources of human stem cells?</i></p> <p>143</p>	<p><i>How could we make a stem cell with only some skin cells?</i></p> <p>144</p>

*Neonatal cells have longer telomeres (which indicate the age of the cell), since they get shorter at each cell division since they do not get replicated, and neonatal cells have not divided many times.*

*Embryos and fetuses have to evade the mother's immune system, so there are less surface markers on cells. Also, newly born babies have no/little immune system so there is less chance of GVHD.*

138

137

*Before the baby is born*

*Telomerase*

140

139

*A bank of embryonic stem cells, where each 'line' of cells is derived from a single embryo.*

*Telomerase so that the cells are immortal and divide indefinitely.*

142

141

*Make it into an induced pluripotent stem cell in the lab.*

*Embryonic SC's, Neonatal SC's, adult SC's (bone marrow, fat tissue (liposuction), skin).*

144

143

<p><i>What are the currently approved stem cell based therapies?</i></p> <p>145</p>	<p><i>How does a bone marrow transplant to cure leukaemia work?</i></p> <p>146</p>
<p><i>Give an example of tissue engineering</i></p> <p>147</p>	<p><i>What is ex-vivo and in-situ cartilage engineering</i></p> <p>148</p>
<p><i>List advantages of MSC's</i></p> <p>149</p>	<p><i>MSC's might be good for [REDACTED] e.g. with HMC's since they help other stem cells to graft</i></p> <p>150</p>
<p><i>Clinical trials take [REDACTED], and [REDACTED] therapies are in phase 3 for stem cell treatments. Foreign clinics advertise MSC treatments, but none have published data from clinical trials.</i></p> <p>151</p>	<p><i>Most trials for stem cell therapies are carried out with MSC's ([REDACTED]), HSC's count for [REDACTED]. ESC's are around [REDACTED] and are being tested with [REDACTED] since they are [REDACTED].</i></p> <p>152</p>

1. Get a matching donor 2. Replicate stem cells ex vitro 3. Destroy bone marrow in patient using irradiation and chemotherapy 4. transplant stem cells into patient.

*Skin grafts, Hematopoietic SC transplant from adult bone marrow or neonatal cells.*

146

145

*Growing new cartilage outside the body and in the body respectively (using MSC's to stimulate growth).*

*Remove cells from lungs, hips and nose, remove a donor trachea (from cadaver) and remove all cells, grow cells around trachea and transplant in patient.*

148

147

*MSC's might be good for cotransplants e.g. with HMC's since they help other stem cells to graft*

- Easy to isolate
- Plastic (not literally!) in the lab
- Can be frozen and thawed
- Possess potent immuno-suppression and anti-inflammation effects
- Capable of homing (going to site of injury)
- Stimulate regeneration

150

149

*Most trials for stem cell therapies are carried out with MSC's (70 percent), HSC's count for 20 percent. ESC's are around 2 percent and are being tested with eyes since they are immuno-privileged.*

*Clinical trials take a long time, and less than 10 therapies are in phase 3 for stem cell treatments. Foreign clinics advertise MSC treatments, but none have published data from clinical trials.*

152

151

<p><i>SC's can be used for [REDACTED],</i> <i>[REDACTED]</i> <i>and [REDACTED].</i></p> <p>153</p>	<p><i>For repairing and replacing cells, what type of cell should we use?</i></p> <p>154</p>
<p><i>What is an induced pluripotent stem cell?</i></p> <p>155</p>	<p><i>How to do Parkinson's in a dish?</i></p> <p>156</p>
<p><i>The traditional approach to medicine is [REDACTED].</i></p> <p>157</p>	<p><i>The traditional approach to medicine does not take into account [REDACTED], which is successful for some, but not all patients.</i></p> <p>158</p>
<p><i>What is stratified medicine?</i></p> <p>159</p>	<p><i>Personalised medicine (aka [REDACTED]) takes into account</i></p> <p>160</p>

*The patients own cells (autologous transplants). This requires adult stem cells that are reasonably plastic though, and its hard to isolate ASC's in the lab. Otherwise, use donor SC's with low immunogenicity.*

154

*SC's can be used for replacing cells (e.g. transplants), repairing cells (e.g. genetically modify SC's outside the body and re-implant) and protecting via MSC immunosuppression.*

153

- *Collect skin cells*
- *Re-program them into stem cells*
- *Grow brain cells from them (induce brain cell differentiation)*
- *Stress out the brain cells with toxins*
- *Observe Parkinson's-like features*

156

*When you reprogram a normal (e.g. skin) cell by inserting genes (via viruses or otherwise). Only 3-4 gene insertions required.*

155

*The traditional approach to medicine does not take into account individual differences between patients, which is successful for some, but not all patients.*

*The traditional approach to medicine is one size fits all.*

158

157

*Personalised medicine (aka precision medicine) takes into account*

*Targeting different types of specific diseases made up of lots of different genes e.g. maturity onset diabetes*

160

159



<p><i>Examples of historical personalised medicine include...</i></p> <p>161</p>	<p><i>When the human genome project started, █ drugs had pharmacogenetic information. After it ended, █ drugs had this information and ten years later, there █ drugs. Now the █, █, █ and █ are examined.</i></p> <p>162</p>
<p><i>Genetic changes of interest include █, █, █ and █. These all change how much of the proteins coded for by an affected gene is produced.</i></p> <p>163</p>	<p><i>What are the advantages of personalised medicine (6 things)?</i></p> <p>164</p>
<p><i>What genes increase your risk of breast and ovarian cancer and how much by?</i></p> <p>165</p>	<p><i>There are over █ predictive tests looking at █ genes. They can █ of treating patients.</i></p> <p>166</p>
<p><i>Even if a predictive test for a gene doesn't have an associated drug to lower risk, you can █.</i></p> <p><i>Sergey Brin does this for Alzheimer's!</i></p> <p>167</p>	<p><i>It's easy to take biopsy of cancer tumours (because they're by definition, not needed), so they can have their genome sequenced to see what genes the cancers have.</i></p> <p>168</p>

When the human genome project started, 4 drugs had pharmacogenetic information. After it ended, 46 drugs had this information and ten years later, there 104 drugs. Now the genome, proteome, metabolome and epigenome are examined.

Inheritance of alkaptonuria, blood transfusions using blood capability testing, genetic basis of selective toxicity of an antimalarial drug.

162

161

- Shift reaction to prevention
- Predict susceptibility of developing a disease
- Improve dosing of drugs (increase efficiency, reduce side effects)
- Reduce cost, time and attrition rate in drug development
- Decrease adverse affects of drugs, increase diagnostic and detection power for disease

Genetic changes of interest include SNP's, base insertions, copy-number variations and variable number tandem repeats. These all change how much of the proteins coded for by an affected gene is produced.

164

163

There are over 15000 predictive tests looking at 2800 genes. They can save the cost of treating patients.

BRAC1, BRAC2; 85 percent higher lifetime chance of breast cancer and 60 percent chance of ovarian cancer.

166

165

It's easy to take biopsy of cancer tumours (because they're by definition, not needed), so they can have their genome sequenced to see what genes the cancers have.

Even if a predictive test for a gene doesn't have an associated drug to lower risk, you can change environmental factors (e.g. eat better, stop smoking etc). Sergey Brin does this for Alzheimer's!

168

167

<p><i>There are drugs (Ivacaftor) that target the [redacted] of diseases rather than just treating symptoms.</i></p> <p>169</p>	<p><i>What does metastatic cancer mean?</i></p> <p>170</p>
<p><i>Enzymes metabolise drugs, and [redacted] metabolise over [redacted] percent of drugs. There are [redacted] in genes that code for these enzymes. Some people metabolise fast (and are at risk of [redacted]), or even ultra-fast metabolisers (meaning the drugs [redacted]).</i></p> <p>171</p>	<p><i>After a stent has been put into an artery, the body recognises it as foreign and blood will clot around it. A drug is given to stop clotting, but one enzyme (CYP 2C19) converts the drug from inactive to active. Variations in this enzyme mean not as much is converted, meaning the blood can clot possibly causing a heart attack or stroke.</i></p> <p>172</p>
<p><i>What are some problems with personalised medicine?</i></p> <p>173</p>	<p><i>What are the ethical problems with personalised medicine (5 things)?</i></p> <p>174</p>
<p><i>[redacted] mutations can be involve with genes. Drugs need to target driver mutations in order to be effective.</i></p> <p>175</p>	<p><i>Define biomarker</i></p> <p>176</p>

*When the cancer has moved from the original site to other areas of the body.*

170

*There are drugs (Ivacaftor) that target the gene underlying cause of diseases rather than just treating symptoms.*

169

*After a stent has been put into an artery, the body recognises it as foreign and blood will clot around it. A drug is given to stop clotting, but one enzyme (CYP 2C19) converts the drug from inactive to active. Variations in this enzyme mean not as much is converted, meaning the blood can clot possibly causing a heart attack or stroke.*

172

*Enzymes metabolise drugs, and one family of enzymes metabolise over 90 percent of drugs. There are thousands of mutations in genes that code for these enzymes. Some people metabolise fast (and are at risk of overdose toxicity), or even ultra-fast metabolisers (meaning the drugs are broken down before they have an effect).*

171

*Who sees the data? How will it be stored? How will it be used? Could it be used against us? What legal protection do we have?*

174

*Ethics, multiple gene variations per disease, quantity of data*

173

*A naturally occurring molecule, gene or characteristic by which a particular pathological or physiological process, disease etc can be identified.*

176

*Driver and passenger mutations can be involve with genes. Drugs need to target driver mutations in order to be effective.*

175

*Why are biomarkers helpful?*

177

\_\_\_\_\_ can be used to build up a signature, telling us how multiple \_\_\_\_\_ etc contribute towards a disease

178

\_\_\_\_\_ and \_\_\_\_\_ are used to identify genes involved in diseases.

179

Given a patient, we can use \_\_\_\_\_, \_\_\_\_\_ and \_\_\_\_\_ (detecting antigens on the surface of cells) to determine their biomarkers. These can be gotten from \_\_\_\_\_, \_\_\_\_\_, \_\_\_\_\_, \_\_\_\_\_ etc. Anywhere where we \_\_\_\_\_ in the body.

180

*How do DNA chips work?*

181

*What is the name for a test that goes with a drug?*

182

Oncotype Dx identifies \_\_\_\_\_ genes associated with \_\_\_\_\_ and \_\_\_\_\_ housekeeping genes (used as a control). These are used to give a score of 1-100 giving the likely reoccurrence of \_\_\_\_\_ within the next \_\_\_\_\_ years. It also predicts the response to \_\_\_\_\_. This costs \_\_\_\_\_.

183

\_\_\_\_\_ determines how aggressive a \_\_\_\_\_ tumor is (i.e. whether there is a high or low risk of \_\_\_\_\_). It measures the mRNA of \_\_\_\_\_ genes. A biopsy is taken and determined to make sure that \_\_\_\_\_ or more cells are cancerous, then the tissue is used for a \_\_\_\_\_.

184

*multiple biomarkers can be used to build up a signature, telling us how multiple genes/proteins etc contribute towards a disease*

178

*Because they help with prediction, diagnosis, progression, regression, or the outcome of treatment of a disease.*

177

*Given a patient, we can use DNA sequencing, microarrays and immunohistochemistry (detecting antigens on the surface of cells) to determine their biomarkers. These can be gotten from normal or diseased tissue, blood, saliva, sweat etc. Anywhere where we can find protein or DNA in the body.*

180

*GWAS and microarrays are used to identify genes involved in diseases.*

179

*A companion diagnostic (CDx).*

*First, sample DNA is taken, then it is amplified using PCR. It is then placed on a DNA chip with many probes, where it will bind to probes that it is complementary to. The chip is washed to remove the non-bound DNA, then scanned, where the bound probes will be visible.*

182

181

*MammaPrint determines how aggressive a breast cancer tumor is (i.e. whether there is a high or low risk of metastasis). It measures the mRNA of 1900 genes. A biopsy is taken and determined to make sure that thirty percent or more cells are cancerous, then the tissue is used for a microarray.*

184

*Oncotype Dx identifies 16 genes associated with breast cancer and 5 housekeeping genes (used as a control). These are used to give a score of 1-100 giving the likely reoccurrence of the tumor within the next ten years. It also predicts the response to chemotherapy. This costs \$4175.*

183

*What is ecosystem services?*

185

*Give examples of ecosystem services.*

186

*Biological resources include...*

187

*What are the social benefits to biodiversity?*

188

*Define biodiversity*

189

*What are the three main levels of biodiversity.*

190

*Define genetic diversity*

191

*What does a low genetic diversity mean?*

192

*FILL IN*

*Involves putting a value on a service that protects biodiversity.  
E.g. instead of building a dam, work out how much preserving  
a forest can help water retention (protecting water resources).*

186

185

*Research, recreation and tourism, culture.*

*Food, medicinal resources and pharmaceutical drugs (e.g. stuff  
in rainforests), wood, ornamental plants, breeding stocks, gene  
diversity.*

188

187

*Genetic diversity, species diversity, ecosystem diversity.*

*The variety of life at all levels; gene level, population level,  
species level, ecosystem level. Also, the interactions between  
these living things.*

190

189

*The key factor driving the extinction vortex is the loss of  
genetic variation since variation is necessary for evolutionary  
responses to environmental change.*

*The combination of different genes found within a population  
of a single species, and the pattern of variation found within  
different populations of the same species.*

192

191



<p><i>A small population is prone to positive feedback loops that draw it down an [redacted].</i></p> <p>193</p>	<p><i>Define genetic drift</i></p> <p>194</p>
<p><i>Extinction vortex: small population means interbreeding and genetic drift, so there is a loss of genetic diversity, meaning that there is a reduction in individual fitness and population adaptability so there is lower reproduction and a higher mortality.</i></p> <p>195</p>	<p><i>What (is the biggest thing that) makes species susceptible to extinction?</i></p> <p>196</p>
<p><i>Cheetah has a [redacted] because it had a [redacted] near the last ice age ([redacted]) and they had an isolated populations in North Africa and Asia are [redacted].</i></p> <p>197</p>	<p><i>Greater Prairie Chicken</i></p> <p>198</p>
<p><i>Define species diversity</i></p> <p>199</p>	<p><i>Define species richness</i></p> <p>200</p>

*When a population has little genetic variation, any disease that all individuals are susceptible to could kill the whole population.*

194

*A small population is prone to positive feedback loops that draw it down an extinction vortex.*

193

*Small population size; i.e. rare species are most at risk.*

*Extinction vortex: small population means interbreeding and genetic drift, so there is a loss of genetic diversity, meaning that there is a reduction in individual fitness and population adaptability so there is lower reproduction and a higher mortality.*

196

195

*Greater Prairie Chicken*

*Cheetah has a low genetic variation because it had a genetic bottleneck near the last ice age (only a few individuals survived) and they had an isolated populations in North Africa and Asia are still genetically similar.*

198

197

*The number of different species in a particular area.*

*The variety and abundance of different types of organisms which inhabit an area.*

200

199

*Define species evenness*

201

*Define ecosystem diversity*

202

*An ecosystem can [redacted] such as a whole forest or  
a [redacted] such as a pond.*

203

*Give four causes of biodiversity loss*

204

*Most threatened species are imperiled*

*[redacted].*

205

*Give the three types of (endangered) species*

206

*What is a rare species?*

207

*What is a dominant species?*

208

*Encompasses the variety of habitats that occur in a region, or the mosaic of patches found within a landscape*

*The relative abundance with which each species is represented in an area (e.g. lots more grey than red squirrels).*

202

201

*Habitat loss, intriduced species, overexploitation, pollution.*

*An ecosystem can cover a large area such as a whole forest or a small area such as a pond.*

204

203

*Rare, dominant and keystone*

*Most threatened species are imperiled for more than one reason.*

206

205

*A species that supports many other species. Saving these often helps many others.*

*A species that has a small population.*

208

207

<p><i>What is a keystone species?</i></p>	<p><i>Conserving biodiversity aims to look for [REDACTED] by mapping biodiversity by region (richness, levels of threat) and by country. These areas are ranked and preserved.</i></p>
209	210
<p><i>Hotspots have three aspects; [REDACTED], [REDACTED] and [REDACTED]. These rarely overlap.</i></p>	<p><i>There are [REDACTED] hotspots in the world containing [REDACTED] of the worlds threatened vertebrates and only cover [REDACTED] of the earth's surface.</i></p>
211	212
<p><i>FILL IN</i></p>	<p><i>FILL IN</i></p>
213	214

*Conserving biodiversity aims to look for hotspots by mapping biodiversity by region (richness, levels of threat) and by country. These areas are ranked and preserved.*

210

*A keystone species is a species whos very presence contributes to a diversity of life and whos extinction would lead to the extinction of many otheres. They help support the whole ecosystem.*

209

*There are 34 hotspots in the world containing seventy five percent of the worlds threatened vertebrates and only cover two point three percent of the earth's surface.*

212

*Hotspots have three aspects; richness, threatened species and endemic (unique to a location) species. These rarely overlap.*

211

*FILL IN*

*FILL IN*

214

213