
HL Paper 1

Which is a source of chromosomes for pre-natal diagnosis of abnormalities by karyotyping?

- A. Sperm
 - B. Ovaries
 - C. Erythrocytes
 - D. Chorionic villi
-

The presence of freckles is a characteristic controlled by a dominant gene. Two parents who are heterozygous for the characteristic have three children, all of whom have freckles. Which statement is true if they have a fourth child?

- A. There is a 100 % chance that their next child will have freckles.
 - B. There is a 75 % chance that their next child will have freckles.
 - C. There is a 50 % chance that their next child will have freckles.
 - D. The next child will have no freckles as the ratio is 3 with freckles to 1 without freckles.
-

What is a plasmid?

- A. Chloroplast DNA
 - B. Mitochondrial DNA
 - C. Small circle of DNA that can transfer genes to or from a prokaryote
 - D. The bacterial chromosome
-

The feather colour of a certain breed of chicken is controlled by codominant alleles. A cross between a homozygous black-feathered chicken and a homozygous white-feathered chicken produces all speckled chickens. What phenotypic ratios would be expected from a cross between two speckled chickens?

- A. All speckled
 - B. 1 black feathers : 1 white feathers
 - C. Speckled, black feathers and white feathers in equal numbers
 - D. 1 black feathers : 2 speckled feathers : 1 white feathers
-

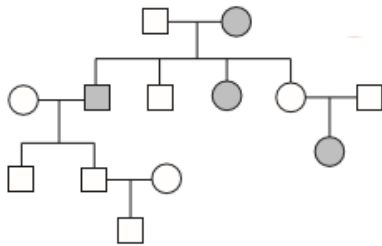
What is the major health issue resulting from the Chernobyl nuclear accident in 1986?

- A. Coronary thrombosis
- B. Cholera
- C. Sex-linked diseases
- D. Thyroid cancer

How are enzymes used during gene transfer involving plasmids?

	To cut plasmids	To extract gene from DNA	To rejoin DNA
A.	✓	✓	✓
B.	—	✓	—
C.	✓	✓	—
D.	✓	—	✓

Alkaptonuria is an inherited condition in humans that affects phenylalanine and tyrosine metabolism, resulting in the production of black-coloured urine. What deduction can be made about the allele for this condition from the pedigree chart?



Key:

- Unaffected male
- Affected male
- Unaffected female
- Affected female

- A. It is autosomal dominant.
- B. It is autosomal recessive.
- C. It is X-linked recessive.
- D. It is Y-linked recessive.

What is a gene mutation?

- A. Failure of chromosome pairs to separate properly during cell division
- B. Changes to genes caused by natural selection
- C. Changes to the nucleotide sequence of the genetic material
- D. Changes in karyotypes

What does therapeutic cloning involve?

- A. Developing genetically identical cultures of human cells for use in drug testing
 - B. Helping infertile couples to conceive by injecting the father's sperm into the mother's egg
 - C. Producing embryonic stem cells for medical use
 - D. Replacing a mutant allele in an embryo to prevent genetic disease
-

A new allele that provides herbicide resistance is identified in soybean plants. The allele is dominant. Which of the following would be carried out in a herbicide-resistant plant to find out if it is homozygous or heterozygous for the gene?

- A. Gel electrophoresis
 - B. Karyotyping
 - C. Test cross
 - D. DNA profiling
-

A certain breed of mouse can have fur that is either black or brown. A mouse breeder performs a test cross using a brown mouse to determine the unknown genotype of a black mouse. There are four offspring produced by the cross. What conclusions can be drawn?

- A. If the black mouse is homozygous, one of the four offspring must be brown.
 - B. If the black mouse is heterozygous, three of the four offspring must be black.
 - C. The black mouse must be homozygous if all four offspring are black.
 - D. The black mouse must be heterozygous if any of the offspring are brown.
-

In a human with type A blood, what determines the blood group?

- A. Sex chromosomes
 - B. One or two alleles
 - C. Multiple alleles
 - D. Codominant alleles
-

What is the chromosome number in a human gamete with non-disjunction?

- A. 46
 - B. 45
 - C. 24
 - D. 23
-

What is a characteristic of the human Y chromosome?

- A. It is made of DNA and histones covered by phospholipids.
 - B. It contains some genes that are not present on the X chromosome.
 - C. It is the largest chromosome in the human karyotype.
 - D. It has a condensed length of approximately 100 μm .
-

What is the difference between the alleles of a gene?

- A. Their position on the chromosome
 - B. Their amino acid sequence
 - C. Their pentose sugars
 - D. Their base sequence
-

What is meiosis?

- A. Division of a diploid nucleus to form diploid nuclei
 - B. Reduction division of a haploid nucleus to form diploid nuclei
 - C. Reduction division of a diploid nucleus to form haploid nuclei
 - D. Division of a haploid nucleus to form haploid nuclei
-

Which is a characteristic of the haploid number of eukaryotic chromosomes?

- A. It doubles in mitosis.
 - B. It is fixed for each species.
 - C. It is an even number for all species.
 - D. It is positively correlated with an animal's mass.
-

Which statement about the polymerase chain reaction (PCR) is correct?

- A. PCR allows DNA to be cloned in a test tube and millions of copies can be made in hours.
 - B. PCR allows the synthesis of RNA from DNA molecules.
 - C. PCR is a natural process carried out by some viruses to amplify DNA molecules.
 - D. PCR is a man-made technique used to identify the nucleotide sequence of DNA.
-

What causes genetic variety in the formation of gametes during meiosis?

- A. Crossing over in prophase I and random orientation of homologous chromosomes in metaphase I
- B. Crossing over in metaphase I and random orientation of homologous chromosomes in metaphase II
- C. Linkage of genes in prophase I and crossing over in metaphase I

D. Linkage of genes in metaphase I and random orientation of homologous chromosomes in metaphase II

What was an aim of genetic modification of organisms?

- A. To provide stem cells from embryos for medical use
 - B. To make crop plants resistant to herbicides
 - C. To provide sperm cells for *in vitro* fertilization (IVF)
 - D. To produce genetically identical sheep
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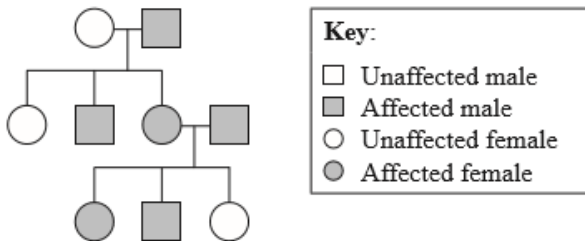
In humans the ABO blood groups are an example of inheritance involving multiple alleles. In a family the parents have blood group A and blood group B respectively. Their first child has blood group O. What is the probability that their next child will have blood group B?

- A. 100 %
 - B. 75 %
 - C. 50 %
 - D. 25 %
-

What makes gene transfer between species possible?

- A. All species use the same genetic code.
 - B. All species have the same genetic material.
 - C. All species produce the same polypeptides.
 - D. All species transcribe genes using plasmids.
-

The diagram shows a pedigree.



According to the pedigree shown, which pattern of inheritance is indicated?

- A. Sex-linked recessive trait
 - B. Autosomal recessive trait
 - C. Autosomal dominant trait
 - D. Codominant alleles
-

One type of gene mutation involves a base substitution.

Original DNA sequence: GAC TGA GGA CTT CTC TTC AGA

mutated sequence 1: GAC TGA GGA CAT CTC TTC AGA

mutated sequence 2: GAC TGA GGA CTC CTC TTC AGA

mRNA codons for valine GUU GUC GUA GUG

mRNA codons for glutamic acid GAA GAG

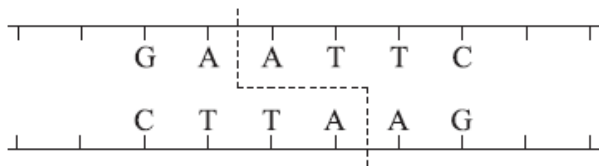
What are the consequences of the base substitutions in the two new sequences of DNA?

- A. Both are mutations that would result in different polypeptides.
- B. Sequence 2 would result in a changed polypeptide but sequence 1 would not.
- C. All three DNA sequences would translate into the same polypeptide.
- D. Only the original DNA and sequence 2 would translate into the same polypeptide.

What is characteristic of homologous chromosomes?

- A. They have an identical DNA sequence.
- B. They are of the same length in karyograms.
- C. They form pairs in prokaryotes.
- D. They carry the same alleles.

What type of enzyme could be used to cut a DNA molecule as indicated by the dotted line on the diagram below?



- A. DNA ligase
- B. DNA polymerase
- C. Helicase
- D. Restriction enzyme

What could be achieved by DNA profiling using gel electrophoresis?

- A. The chromosome number of an organism could be counted.
- B. It could be proven that human tissue found at the site of a crime did not come from a person suspected of having committed the crime.
- C. A karyotype could be produced.
- D. Extinct species of living organisms could be brought back to life.

Which process can be used to amplify small fragments of DNA?

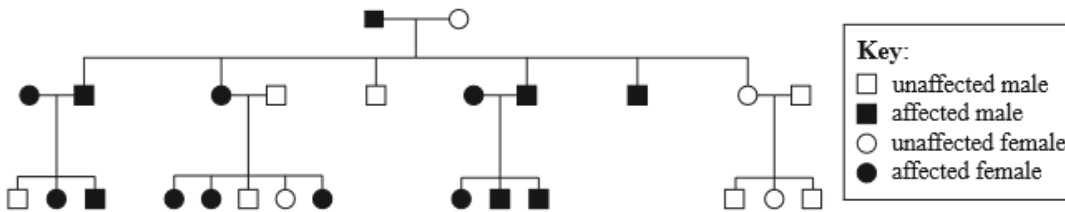
- A. Gel electrophoresis
- B. Polymerase chain reaction
- C. DNA profiling
- D. Electron microscopy

In a cross between red haired cattle and white haired cattle the offspring produced are always a colour called roan (light red). If the roan cattle are interbred they produce white, roan and red offspring.

How many alleles are controlling this character?

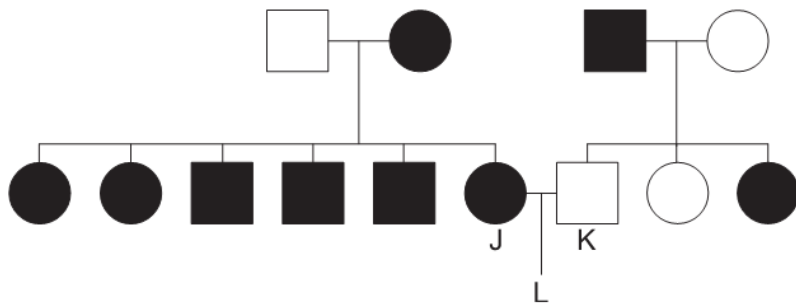
- A. 1
- B. 2
- C. 3
- D. 4

What evidence is given in the pedigree chart below to prove that the condition is caused by a dominant allele?



- A. Two unaffected parents have unaffected children.
- B. Two affected parents have affected children.
- C. An affected parent and an unaffected parent have affected children.
- D. Two affected parents have an unaffected child.

A dominant autosomal allele for lactase persistence allows humans to digest milk as adults. People who lack this allele are lactose intolerant in adulthood.



- Key:**
- lactase persistent male
 - lactose intolerant male
 - lactase persistent female
 - lactose intolerant female

If J and K have a child L, what is the probability that L will be lactase persistent?

- A. 25 %
- B. 50 %
- C. 75 %
- D. 100 %

When genes are transferred between species, the amino acid sequence of the polypeptide translated from them is unchanged. Why is this so?

- A. All organisms use ribosomes for protein synthesis.
- B. DNA replication is semi-conservative.
- C. The enzymes used are substrate specific.
- D. The genetic code is universal.

What is a definition of a clone?

- A. A group of cells derived from a single parent cell
- B. Differentiated cells that retain the capacity to divide
- C. A fetus developed specifically for medical use
- D. A group of cells that have lost the ability to differentiate

Which of the following statements about homologous chromosomes is correct?

- A. Each gene is at the same locus on both chromosomes.
- B. They are two identical copies of a parent chromosome which are attached to one another at the centromere.
- C. They always produce identical phenotypes.
- D. They are chromosomes that have identical genes and alleles.

What event occurs only in meiosis?

- A. Fusion of gametes to promote genetic variation
- B. Random separation of chromatids
- C. Random separation of homologous chromosomes

D. Replication of chromosomes

What description is matched with the correct phase in meiosis I?

A.	Prophase I	recombination occurs only between sister chromatids
B.	Metaphase I	homologous chromosomes join together at each end of the cell
C.	Anaphase I	homologous chromosomes are pulled apart
D.	Telophase I	two diploid nuclei are produced

What does the universal nature of the genetic code allow?

- A. Change of genetic code in the same species
 - B. Transfer of genes between species
 - C. Formation of clones
 - D. Infection by bacteria
-

If a father with A-type blood and a mother with B-type blood have a child, what is the probability that the child will have O-type blood?

- A. 50 % chance if both parents have the recessive allele.
 - B. 25 % chance if both parents have the recessive allele.
 - C. 0 % chance because neither parent has the allele.
 - D. 50 % chance if either parent has the recessive allele.
-

What is the effect of dominant alleles?

- I. They mask the effect of recessive alleles.
 - II. They become more frequent than recessive alleles in a population.
 - III. They have a joint effect with recessive alleles when characteristics are co-dominant.
- A. I only
 - B. I and II only
 - C. I and III only
 - D. I, II and III
-

The sequence of the first six amino acids of the normal β hemoglobin (Hb^A) chain are listed.

valine – histidine – leucine – threonine – proline – glutamic acid

Which sequence of amino acids could there be in the first six amino acids of the sickle-cell β hemoglobin (Hb^S) chain?

- A. glutamic acid – histidine – leucine – threonine – proline – valine
 - B. valine – valine – histidine – leucine – threonine – proline
 - C. glutamic acid – histidine – leucine – threonine – proline – glutamic acid
 - D. valine – histidine – leucine – threonine – proline – valine
-

Which genetic condition can be diagnosed by karyotyping?

- A. Trisomy 21
 - B. Sickle-cell anemia
 - C. Hemophilia
 - D. Colour blindness
-

What term describes the failure of sister chromatids to separate during anaphase II?

- A. Sex linkage
 - B. Karyotyping
 - C. Non-disjunction
 - D. Semi-conservative replication
-

Which sequence of DNA would be suitable in DNA profiling?

- A. ---ATTCGTGAATCAGCC--
 - B. ---ATTCGTGAATTTGCC--
 - C. ---ATTCGTGATTGCAGC--
 - D. ---ATTCGTGATTTCGTGA--
-

In humans a V-shaped hair line is dominant to a straight hair line. A woman with a V-shaped hair line and a man with a straight hair line have children.

The woman has a mother with a straight hair line. What is the proportion of children who are likely to have a V-shaped hair line?

- A. Half of the children
 - B. A quarter of the children
 - C. All of the children
 - D. None of the children
-

Which of the following is an inherited disease that is due to a base substitution mutation in a gene?

- A. Trisomy 21
 - B. Sickle cell anemia
 - C. AIDS
 - D. Type II diabetes
-

What commonly causes Down syndrome in humans?

- A. Non-disjunction
 - B. Base substitution
 - C. Amniocentesis
 - D. Gene mutation
-

In a person who is heterozygous for sickle-cell anemia, where is the mutation found?

- A. In every gamete produced
 - B. Only in gametes carrying an X chromosome
 - C. In all brain cells
 - D. In blood plasma
-

Red-green colour blindness is a sex-linked condition. Which of the following always shows normal vision?

- A. A homozygous male
 - B. A homozygous female
 - C. A heterozygous male
 - D. A heterozygous female
-

Which technique causes fragments of DNA to move in an electric field?

- A. Polymerase chain reaction (PCR)
 - B. Genetic modification
 - C. Therapeutic cloning
 - D. Gel electrophoresis
-

Which of the following genotypes is possible in the offspring of a homozygous male with blood group A and a female with blood group B?

- A. $I^A I^A$
- B. $I^A i$
- C. ii

D. I^Bi

What do **all** human males inherit from their mother?

- I. An X chromosome
- II. A Y chromosome
- III. Mitochondrial DNA

- A. I only
 - B. I and II only
 - C. I and III only
 - D. I, II and III
-

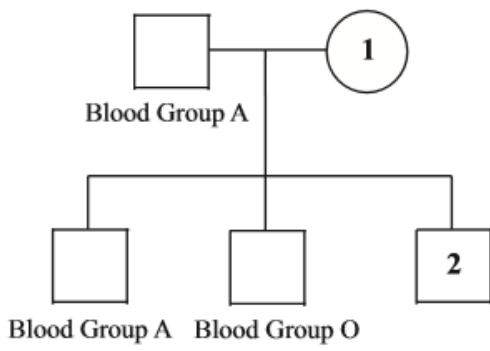
Which is a possible risk associated with a genetic modification of crops?

- A. Crop plants will become weaker with time.
 - B. It can increase mutations in the organisms that consume them.
 - C. Starch obtained from genetically modified plants will be more difficult to digest.
 - D. Resistance to herbicide genes can be transferred to weeds.
-

What maximum number of different genotypes and phenotypes are possible among the children of a mother with blood group A and a father with blood group B?

	Genotypes	Phenotypes
A.	2	2
B.	2	4
C.	4	4
D.	4	2

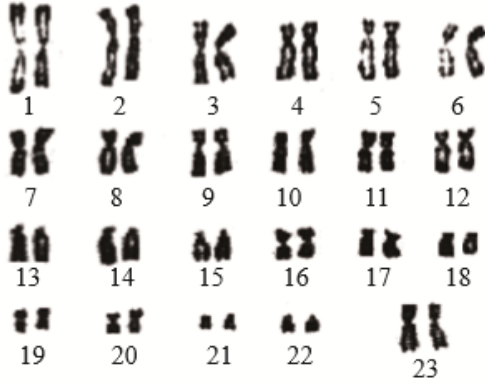
The pedigree chart below shows the blood types of three members of a family.



Which could be the blood types of individuals 1 and 2?

	Individual 1	Individual 2
A.	A	AB
B.	AB	B
C.	O	B
D.	B	A

Questions 38 and 39 refer to the following karyotype.



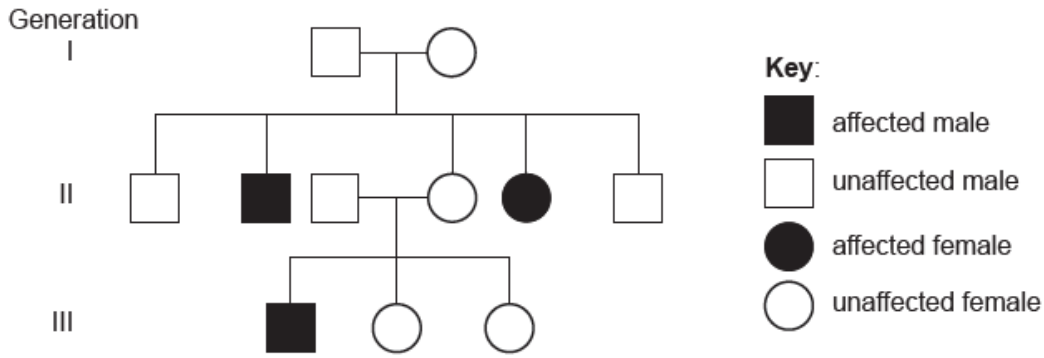
[Source: www.ds-health.com/trisomy.htm]

What procedure(s) could have been involved in the creation of the karyotype?

- I. Chorionic villus sampling
- II. DNA profiling
- III. Amniocentesis

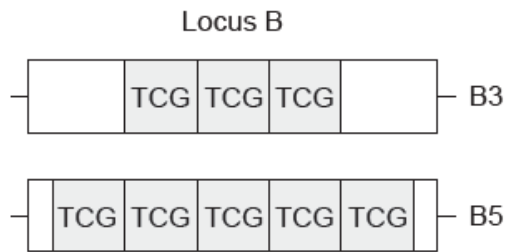
- A. I only
- B. II only
- C. I and III only
- D. I, II and III

What proves that the inheritance of the condition shown in this pedigree chart is autosomal recessive and not autosomal dominant?



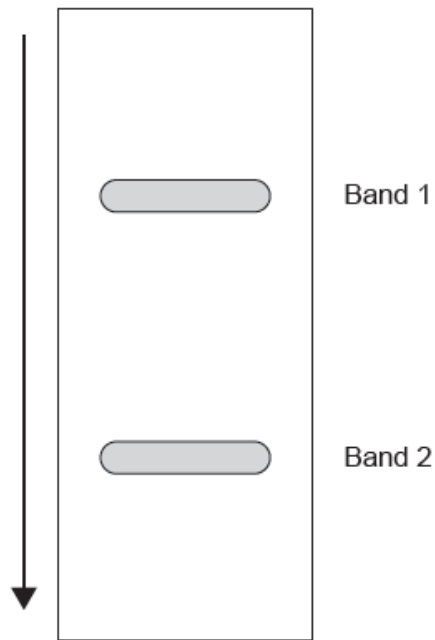
- A. There is no affected person in generation I.
- B. Both males and females are affected.
- C. Two unaffected parents have a child that is affected.
- D. Not everybody in generation III is affected.

A DNA profile was made of one individual in a paternity suit. Locus B was used to distinguish between this individual and other individuals. The individual had two alleles of the gene at locus B which are shown below:



Gel electrophoresis was used to separate and visualize the alleles B3 and B5. The gel, with two bands of DNA, is shown below.

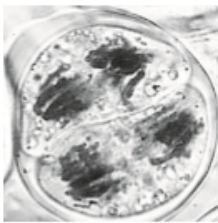
Direction of migration of DNA



What DNA is in bands 1 and 2?

	Band 1	Band 2
A.	mix of B3+B5	B3 only
B.	mix of B3+B5	B5 only
C.	B5 only	B3 only
D.	B3 only	B5 only

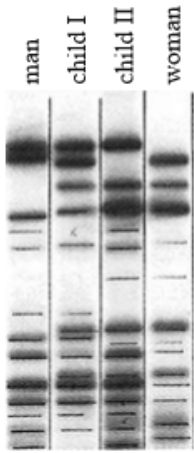
What stage of meiosis is shown in the micrograph?



[Source: www.vcbio.science.ru.nl/en/virtuallessons
Used with permission.]

- A. Prophase I
- B. Metaphase II
- C. Anaphase II
- D. Telophase I

What conclusion can be made from the following evidence from an analysis of DNA fragments?



- A. Both children are related to both parents.
- B. Child I is related to the man but child II is not.
- C. Both children are unrelated to either of the parents.
- D. Child II is related to the man but child I is not.

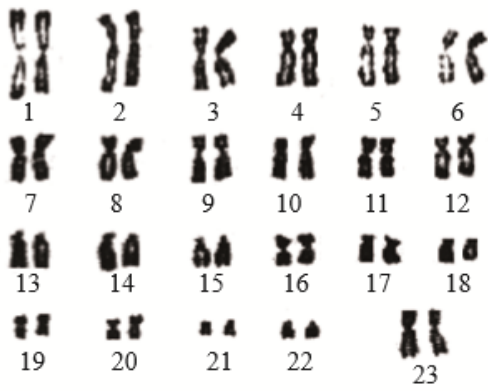
What information can be concluded from the karyotype?



[Source: http://en.wikipedia.org/wiki/File:NHGRI_human_male_karyotype.png]

- A. The person is a normal male.
- B. The person is a normal female.
- C. The person is a male with Down syndrome.
- D. The person is a female with Down syndrome.

Questions 38 and 39 refer to the following karyotype.



[Source: www.ds-health.com/trisomy.htm]

What can be concluded from the karyotype provided?

- A. There was non-disjunction during meiosis in the mother.
- B. There was non-disjunction during meiosis in the father.
- C. The fetus is male.
- D. The fetus is female.

The image shows a karyogram.



[Source: [https://commons.wikimedia.org/wiki/File:Karyotype_of_sheep_\(Ovis_aries\).png](https://commons.wikimedia.org/wiki/File:Karyotype_of_sheep_(Ovis_aries).png),
[https://commons.wikimedia.org/wiki/File:Karyotype_of_sheep_\(Ovis_aries\).png](https://commons.wikimedia.org/wiki/File:Karyotype_of_sheep_(Ovis_aries).png)), by M. Singh, X. Ma, E. Amoah and G. Kannan]

What information can be determined from this karyogram?

- A. The sex is female.
- B. The haploid number is 54.
- C. Disjunction occurred during meiosis.
- D. The species is not human.

