Subject: Biology

Grade: 12

Section: LS

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Unit: Reproduction and Genetics

Chapter 3: Genetic variation and polymorphism

Document 2: Genetic identity of individuals. P:60

Because of the large number of polymorphisms observed in humans, it is virtually certain that each of us is genetically unique (with the exception of identical twins). It follows that genetic variation could be used to identify individuals much as a conventional fingerprint does.

How do you determine the genetic identity of an individual?

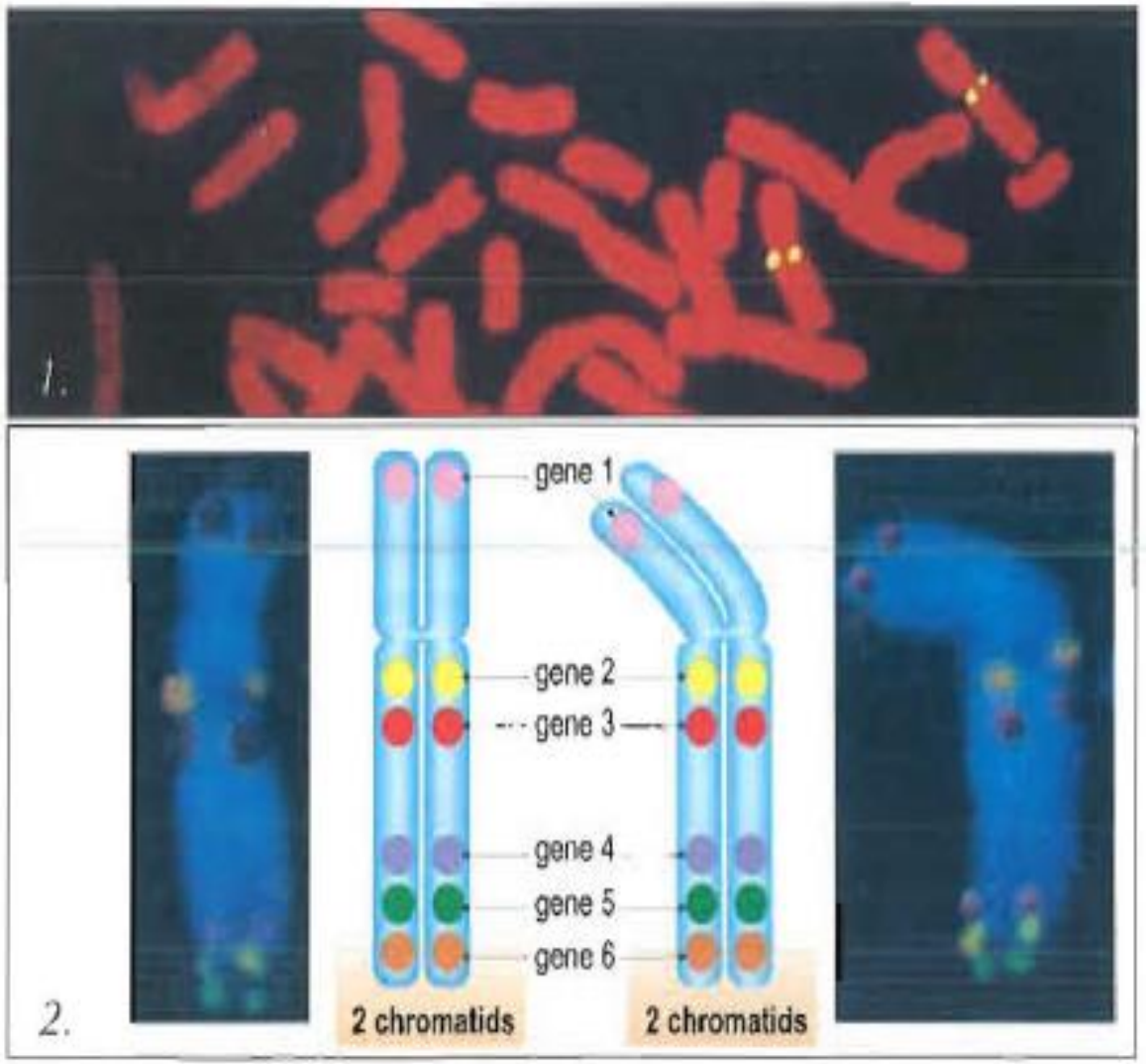
1 - Localization of a gene on a chromosome

A relatively new method allows to locate a DNA segment having a specific nucleotide sequence in the genome. This technique, known as Fluorescence In Situ Hybridization (FISH), uses fluorescent DNA probes which are known sequences of single stranded DNA. A probe is capable of binding (hybridizing) to a specific DNA sequence.

In this method, chromosomes adhering to a microscope slide are **denatured** (DNA strands are partially separated) and then put in the presence of a probe.

This probe fixes "in situ" (at a specific location) to the chromosome.

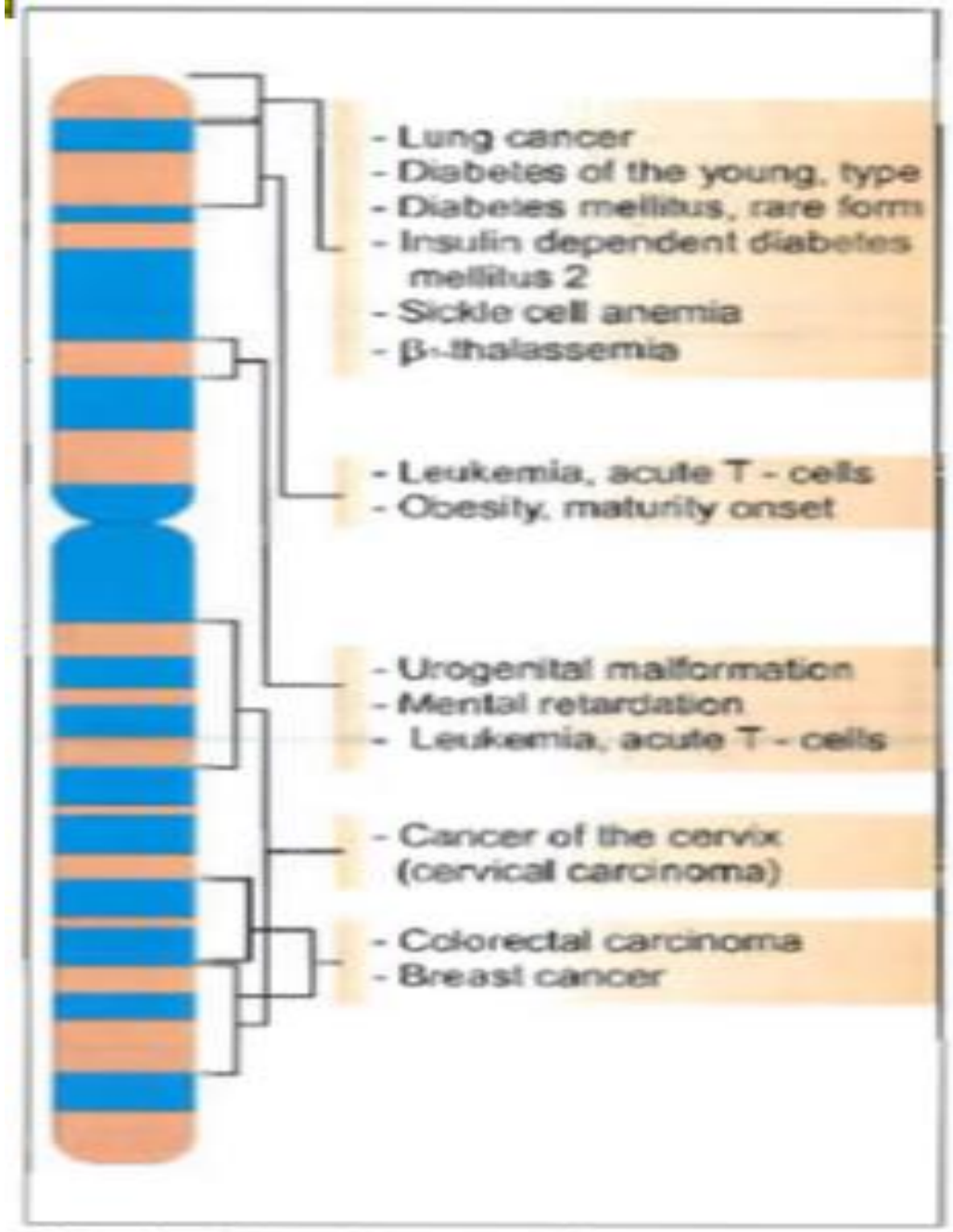
As a result, the chromosome shows a fluorescent dot and hence the gene or the DNA sequence is located (Doc. a).



Doc.a *Genes located by the FISH technique.*
1. A mono-locus probe specific for a given gene.
2. Two human metaphase chromosomes labeled by six different "mono-locus" probes.

2 - Genetic maps

FISH, and other mapping techniques, have allowed scientists to construct **human genetic maps**. The indicated genes are involved in normal cellular functions, and only in the event of their dysfunction or mutation would the diseased state become apparent.



Doc.b Genetic map of chromosome 11.

3- DNA fingerprint

The complete sequencing of the human genome, published in February 2001, revealed that each cell has 30 000 to 40 000 genes. At a given locus, each gene has two alleles that may be different or identical. Suppose a specific gene has only two alleles, rather than several, namely T and t, then a particular individual may possess two identical alleles TT or tt; alternatively he/she may possess two different alleles as in Tt.

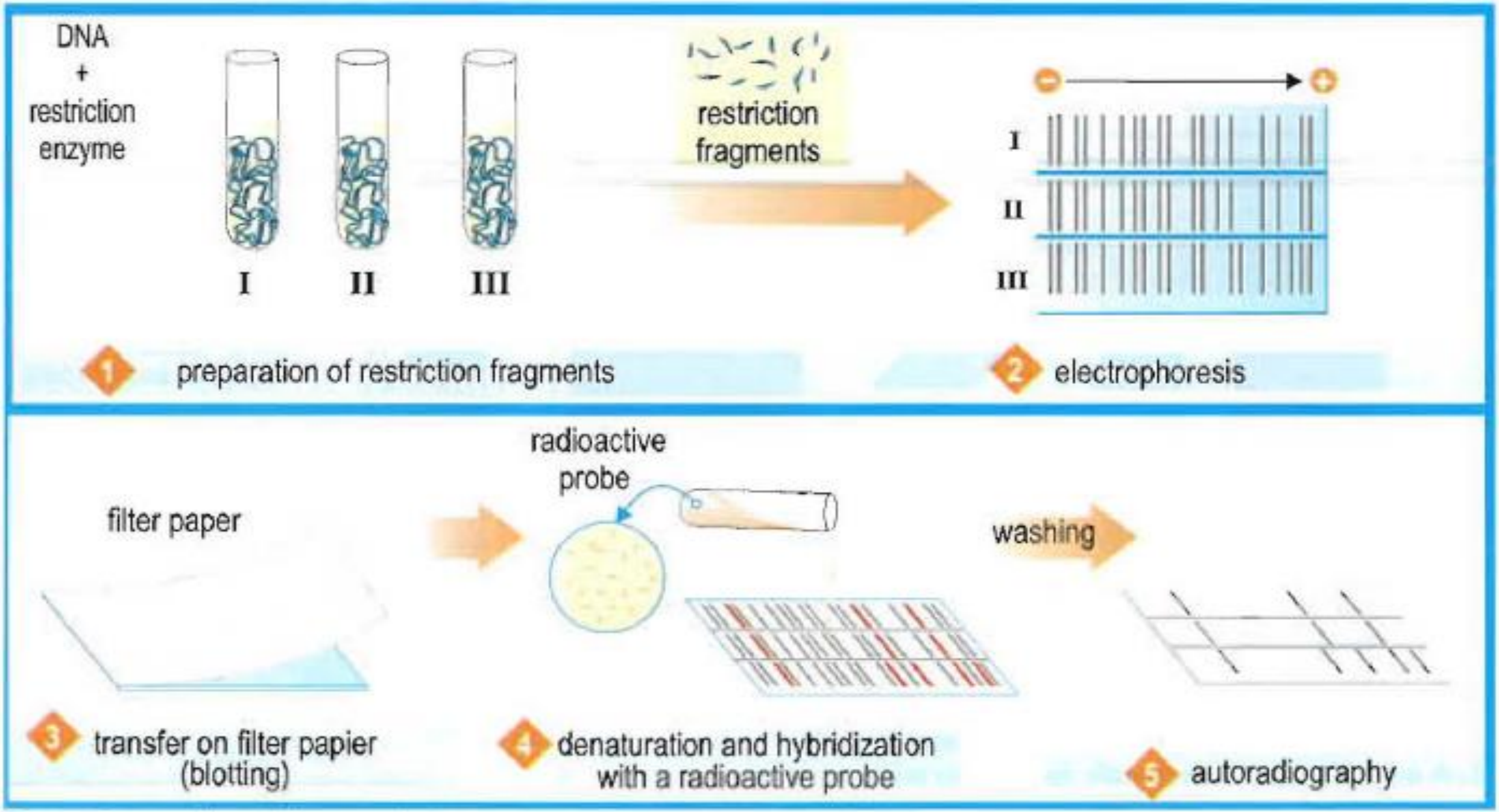
For the sake of simplicity, suppose an individual has only 500 genes (rather than 30,000) with two alleles (rather than several alleles) for each gene. The possible allelic combination is 3 for each gene. Thus each individual could have 3500 possible combinations of the different genes. The likelihood of two individuals to have identical genotype is virtually nil, except of course, for identical twins.

The uniqueness of the genotype can be noted when DNA fingerprints of two individuals are examined by the Jeffreys technique

Jeffrey's technique

This technique consists of five steps:

1. DNA is first cut with a restriction enzyme.
2. DNA fragments are separated by electrophoresis.
3. Fragments are then transferred (blotted) and fixed onto a solid membrane (filter paper).This technique is called "Southern blotting".
4. On the membrane, the denatured fragments are hybridized to a radioactive labeled ^{32}P-DNA probe. This probe is complementary to a DNA sequence that occurs frequently throughout the genome (i.e. a repetitive sequence or multi-locus probe).
5. The hybridization of the DNA sequence with the probe is visualized by a technique known as autoradiography, The resulting pattern of bands for each individual is referred to as a DNA fingerprint, and is uniquely characteristic for that individual (Doc. d).



Doc.c Steps of the Jeffreys technique.

4- DNA fingerprint in paternity

The uniqueness of our DNA fingerprint can be used to determine parenthood. Doc. e shows the DNA fingerprints in two paternity cases.

Each band of the child's DNA fingerprint should be shared with either the father's or the mother's DNA fingerprint, as in case 1.