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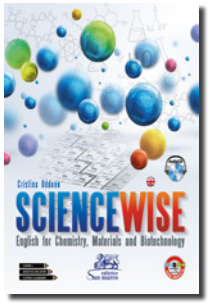
SCIENCEWISE
English for Chemistry, Materials and Biotechnology



Genetic testing

Genetic testing (also called DNA-based tests) is a type of medical test that identifies changes in chromosomes, genes or proteins. It involves direct examination of the DNA molecule taken from cells in a sample of blood, hair, skin, or other tissue. For example the procedure called "buccal smear" uses a small brush to collect a sample of cells from the inside surface of a cheek. Genetic testing is among the newest and most sophisticated techniques used to determine gene variants associated with a specific disease or condition; it is also used for paternal testing and in forensics[1].

In the clinical setting, individuals may wish to be tested if they have a family history of one specific disease, if they show symptoms of a genetic disorder or if they are concerned about passing on a genetic problem to their children. As for **genetic testing before birth**, pregnant women may decide to undergo amniocentesis or chorionic villus sampling, so to check for genetic problems and determine the sex in the child. The most widespread[2] type of genetic testing is **newborn screening**: infants are tested just after birth in order to identify genetic disorders that can be treated early in life; one of the most common types of genetic disorder tested in babies is phenylketonuria. **Diagnostic testing** is used to confirm a diagnosis concerning a specific genetic condition. **Carrier testing** is used by couples whose families have a history of genetic disorders and who are considering having children: if both parents are tested, information can be provided about their risk of having a child with a genetic problem. A common carrier test is the one for cystic fibrosis. **Preimplantation testing** (or preimplanatation genetic diagnosis, PGD) is used to detect genetic changes in embryos in the case of assisted reproductive techniques or in-vitro fertilisation. Much of the current excitement about genetic testing, however, centres on **predictive gene testing**: these tests can determine whether a person will develop a genetic disorder before any signs or symptoms appear. The results can help the person make decisions about medical care. At the moment predictive gene tests are available, among the others, for Huntington's disease and haemochromatosis. **Forensic testing** uses DNA sequences to identify crime or catastrophe victims, implicate a crime suspect or establish biological relationships between people. The physical risks of the gene test itself are minimal, as they usually require only a blood sample or buccal smear. For prenatal testing the procedures can carry a small risk of miscarriage[3]. The limitations of gene testing involve the fact that the tests give only a probability for developing the disorder: one person with a given disease may develop the disease, while another person remains healthy. One of the negative sides of genetic testing is therefore the psychological impact: for people who have already experienced the tragedy of a genetic disease in their families, the news that they can carry that gene themselves can elicit[4] depression or even despair. When considering genetic testing it is important to consult a counsellor, so that the benefits, risks and limitations of that particular test can be made clear in view of the decision.

GLOSSARY

1 scientific tests or techniques used in the investigation of crimes
2 widely diffused
3 spontaneous abortion
4 to give rise to

Amniocentesis is a prenatal test in which a small amount of amniotic fluid containing foetal tissues is sampled from the amniotic sac, and the foetal DNA is analysed for genetic abnormalities such as Down syndrome. It is usually done when a woman is between 16 and 18 weeks pregnant. Generally women choose to have this test if they are at increased risk for genetic problems or if they are over 34, as the chances of having a child with a chromosomal problem increase when a woman is older.

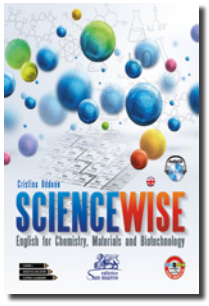
Chorionic villus sampling, usually performed between the 10th and the 12th weeks of pregnancy, is a prenatal test to determine chromosomal or genetic disorders in the foetus. A small piece of the placenta tissue (chorionic villi) is removed from the uterus and tested to check for genetic defects. As chorionic villus sampling is an invasive test, there is a small risk that it can induce a miscarriage.

Phenylketonuria is a genetic disorder which does not allow the body to process part of a protein called "phenylalanine". It is caused by a mutation in a gene that helps create the enzyme needed to break down phenylalanine. If untreated it can cause problems with brain development, mental retardation, seizures. If diagnosed early, a baby can grow up with normal brain development, but only by controlling the levels of phenylalanine through diet and medication.

Cystic fibrosis is a genetic disease of the secretory glands which make mucus and sweat. With cystic fibrosis, mucus becomes thick and sticky, causing infections that can severely affect the lungs. It can also affect the pancreas and the intestine. Furthermore the sweat becomes very salty, causing problems such as weakness, heat stroke, dehydration, decreased blood pressure.

Huntington's disease is a neurodegenerative genetic disorder that causes certain nerve cells in the brain to waste away. It typically becomes noticeable in mid-adult life. Early symptoms are uncontrolled movements, clumsiness or balance problems. Later the disease can take away the ability to walk, talk or swallow. Medicines can help manage some of the symptoms, but at the moment there is no cure and the disease cannot be stopped.

Haemochromatosis is a genetic disease which causes the body to absorb and store too much iron. The extra iron can damage the body's organs. If haemochromatosis is not treated early, it can eventually lead to serious problems such as arthritis, liver and pancreas disease, heart abnormalities. Treatment consists of getting rid of excess iron by removing a certain quantity of blood, and then maintaining the right level by giving a specific amount of blood every two to four months for life.



ACTIVITIES

1 *Complete the following sentences filling in the gaps.*

1 Another name for genetic testing is ..... .
2 Genetic testing analyses ..... taken from human tissue.
3 Besides the clinical setting, genetic testing is used in ..... .
4 Phenylketonuria is one of the most common types of genetic disorders tested just after ..... .
5 If a couple with genetic problems in their family is considering ....., they can undergo carrier testing.
6 ..... is used if you are trying to have a baby using assisted reproductive techniques.
7 One of the few physical risks in genetic testing is a low percentage of ..... in prenatal testing.
8 A ..... may be consulted before undergoing genetic testing.

2 *Find questions to the following answers.*

1 ..... ?
It is called buccal smear.
2 ..... ?
Amniocentesis and chorionic villus sampling.
3 ..... ?
When you want to confirm a diagnosis about a specific genetic disorder.
4 ..... ?
This test identifies people who are at risk of getting a disease before any symptoms are visible.
5 ..... ?
The identification of catastrophe or crime victims, the implication of a crime suspect, establishing biological relationships among people.
6 ..... ?
The psychological impact.

3 *Read the text again and find the words or expressions corresponding to the following definitions.*

1 A strand of DNA in the cell nucleus that carries the genes in a linear order. .....
2 A part of an organism consisting of a large number of cells having a similar structure and function. .....
3 The fleshy part of either side of the face below the eye. .....
4 A sign or an indication of disorder or disease. .....
5 Carrying a foetus within the womb. .....
6 An organism in the early stages of growth. .....
7 A process by which egg cells are fertilised by sperm outside the body. .....
8 A large-scale disaster, a horrible event. .....
9 Someone who is under suspicion of committing violation of the law. .....
10 A mood disorder characterised by the inability to experience pleasure and feelings of sadness and helplessness. .....

4 *Write a summary of the text basing on the following guidelines.*

1 What is genetic testing?
2 How are genetic tests done?
3 What are the types of genetic tests?
4 What are the risks and limitations of genetic testing?

5 *Match each keyword to the sentence referring to it.*

1	Amniocentesis	A	Among its possible consequences there is mental retardation
2	Chorionic villus sampling	B	It involves accumulation of too much iron in the body
3	Phenylketonuria	C	It is done with a sample of the placenta tissue
4	Cystic fibrosis	D	It is done with a sample of the amniotic fluid
5	Huntington's disease	E	One of its early symptoms is clumsiness
6	Haemochromatosis	F	This disease can cause serious problems to the lungs