Personalised medicine

Key Terms

- **Genome:** An organism's complete genetic information, coded in DNA, found in all cells.
- **DNA:** The chemical that carries genetic information, made up of 4 chemical units: A, T, G and C.
- **Gene:** A chunk of DNA which usually contains information to build proteins or to regulate cell's functions.
- **Genome**: The order of the units of the DNA is a code for the chemical units in a protein.
- **Mutation**: A change in the DNA sequence, which can have a positive, negative or neutral effect.
- **Sequencing:** Reading the code of a genome.
- **Genomics:** The study of how the genome works.

What's all the fuss about?

Imagine doctors taking a sample of your saliva to uncover your genetic profile, finding out what diseases you are at greater risk of developing, and prescribing you personalized preventive treatment, at exactly the right dose for your specific profile. This may be possible in the future thanks to advances in genomics. The more we understand the genome, the more we can use this information in healthcare. But this brings up important ethical, social and legal questions. Who should have access to our genetic profiles? How should personalised medicine be funded? And how much information is too much information?

Statistics

- **1 month**: the time taken to obtain a person's complete genome sequence
- **2000**: the number of genetic tests currently available which can inform doctors about how to use drugs to treat a patient
- **25 000**: the estimated number of genes in a human being

Science Q&A

How could our genome affect our health?

Every cell in our body contains genetic material with instructions on how to build every single part of our bodies.

These instructions are coded in DNA, a code made up of four chemical units (bases) that we represent by the letters: A, G, T and C. The order in which these letters appear in the DNA determines the information. A chunk of DNA that contains the instructions to build one protein or to regulate cell's functions is called a gene.

This genetic material varies from one individual to the next, and is inherited from our parents (50% from our mother and 50% from our father). This is what determines genetic differences between us, like eye colour. Identical twins share the same genetic profile, and the differences between them shows the environment also plays a role on determining how we are.

Some DNA code combinations could lead to disease. So by studying a person's genetic profile, there are ways to see how likely that person is to suffer from certain diseases. Our genetic profile also determines how well we will react to different types of medicines.

What is the idea of personalised medicine?

Many drugs do not work effectively for a large number of patients. Medicine used to adopt a "one size fits all" approach. But personalised medicine aims to apply individualised treatments depending on the genetic characteristics of the patients. To do this, it is convenient to have a thorough knowledge of their genome (their genetic information).

Personalised medicine divides patients into groups based on their individual genetic profiles. In this way, it is possible to adapt the treatments, giving each patient the minimal effective dose to avoid side effects and prevent toxic effects.

This can mean identifying genes that make drugs effective or harmful and developing new medicines or vaccines based on the differences between groups in how susceptible they are to infectious diseases.

How can we test to see if a person is likely to develop a medical condition?

Genetic testing can be a way of finding out whether you are carrying a particular genetic mutation that could cause a medical condition. This testing is only effective if research has shown that a medical condition is caused by a specific genetic mutation.

We know, for example, that a specific gene causes **spinal muscular atrophy** (an illness that causes muscle weakness and leaves patients increasingly unable to move). A genetic test on a sample of blood can show whether this gene is present and therefore if the person is likely to develop the illness.

How are genetic tests performed?

Genetic tests can be performed on a sample of blood or hair, or a cotton swab could be used to collect a sample of cells from the inside of the cheek, for example. A laboratory receives the sample and technicians look at the DNA, the chromosomes or the proteins it contains. The laboratory reports the test results to the patient's doctor, and patients have a discussion with a healthcare professional who is trained in the science of human genetics in order to discuss and interpret the results.

Not all genetic conditions have genetic tests to diagnose them – at the moment there are only tests for around 1000 genetic disorders. Most tests are used to diagnose rare conditions like Fragile X Syndrome and Duchenne Muscular Dystrophy.

In the future it may be possible to get an instant result from a genetic test, or even buy a genetic test at a pharmacy to perform at home.

How do patients get advice about genetic conditions?

Patients at risk of genetic conditions usually receive some form of genetic counseling.

Family history is usually very important. Seeing who may have had the condition in the past will help indicate what genetic tests may be appropriate. Geneticists can explain the results of any tests or examinations and help patients decide how to progress.

Alternatively, a geneticist may be involved in assessing the risk of developing a particular condition. For example, if you have a strong family history of cancer, a geneticist will assess your risks and discuss this with you. They can help you to decide whether

Genetic testing and Herceptin

One of the first drugs to require a genetic test was Herceptin, which is a treatment for breast cancer. Before giving it to patients, doctors first need to do a genetic test to check the levels of a gene called HER2 in the tumour tissue, to see if the Herceptin will be effective.

you would like to have cancer screening or other further tests.

Healthcare professionals are obliged to keep genetic information confidential. It is covered by the patient's right to privacy, which is a fundamental human right, and especially because this information could be used for corporate interests instead of for medical objectives.

How can we use genetics to make sure medicine works properly?

Personalised medicine is also having an impact in the way drugs are developed. Increasingly, when a new drug is developed, researchers also develop a genetic test to go with it. This test will give the doctor information on the effectiveness of a drug according to the patient's genetic profile. In this way, when the patient takes the genetic test, the doctor can have a better idea of whether the drug will be effective and choose the most appropriate medication for that case.

Discussion Continuum

This activity is designed to facilitate debates about the ethical, legal and social aspects of research into genomics. Groups of 8-12 students discuss the issues raised by each statement and choose where each card should go between 'agree' and 'disagree'. Larger groups could use the resource to have a free discussion of the topic or you could use formats that require the students to work more formally or in smaller groups.

Contents:

The resource consists of:

- An AGREE and a DISAGREE card
- 8 Discussion Cards, which include a statement on some aspect of genomics

Gameplay:

1. Players form small groups, between 4 and 12 per group. Each group is given an AGREE and DISAGREE card and 8 discussion cards.
2. Within each group, the AGREE card and DISAGREE card are placed on the floor/table about one metre apart, to represent the two extremes of the continuum. The space in between is where the discussion cards will be placed.
3. The first player reads the first discussion card to the rest of the group. The player should check everyone understands the card.
4. The first player then decides to what extent s/he agrees with the first card. S/he places the card face up, anywhere on the discussion continuum, closer to AGREE or DISAGREE as s/he chooses. This is entirely the choice of the individual player, and is not discussed by the group. The player can give a reason, if s/he wishes.
5. Each player in turn then reads a card, checks that everyone understands, and chooses individually where to place it on the continuum in a similar way.
6. When all the cards have been read, understood and placed on the continuum, the discussion begins. The aim is to place the cards between AGREE and DISAGREE in an order that most of the players agree on. Players should pick a card for discussion, and debate whether to move it.
7. At the end of the discussion, each group should have a continuum which they mostly agree with.
8. If several groups are playing at the same time, the facilitator may wish to bring the different groups' results together. Are they similar? Can someone from each group explain their choices on particular cards.

Discussion continuum developed by Ecsite, in collaboration with Barcelona Science Park, in the context of the Xplore Health project.

Thanks to At-Bristol for the development of the discussion continuum format: www.at-bristol.org.uk

Agree

Disagree

Discussion Card 1

"If I don't want to know what my genetic profile says about me, I **should never be obliged to have a test**."

Discussion Card 2

"If I can afford **expensive private healthcare**, I should expect to have **more access to personalised medicine** than someone who has only the most basic healthcare."

Discussion Card 3

"For research into personalised medicine, **more money should be spent on the diseases which affect the most people** in each country. The rarer the disease, the less research should be funded."

Discussion Card 4

"Personalised medicine should be **funded by public money more than for-profit companies**, to ensure it is developed focusing more on need than on profit."

Discussion Card 5

"When testing patients to see what diseases they are at risk of in the future, doctors should **never test for diseases like Alzheimer's where there is no cure**."

Discussion Card 6

"The police **should not be allowed to keep a genetic database** of all citizens in order to facilitate their investigations."

Discussion Card 7

"If we identify a gene which relates to a predisposition for baldness, a couple receiving fertility treatment **should be able to choose** whether or not they want their child to inherit that gene."

Discussion Card 8

"Genetic tests for to find out whether men are infertile should **never be available over the counter** in pharmacies. This type of test must be accompanied with guidance from a doctor."

Discussion Card 9

"If a genetic test reveals the risk of a serious condition, **doctors should be obliged to tell the immediate family** of the patient, as they share genes with the patient and may be at risk."