

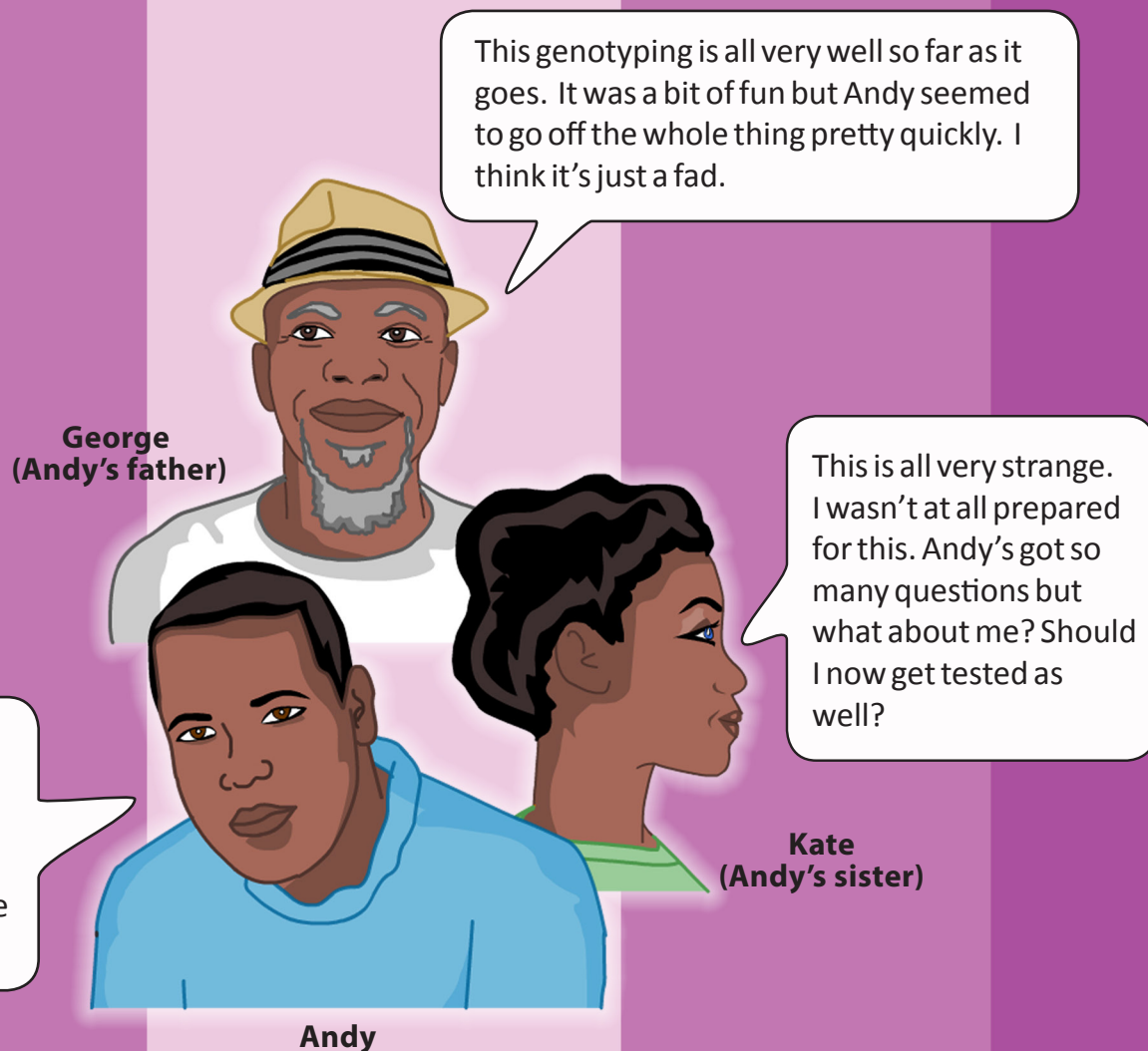


yourgenome.org

Last year (2024) Andy bought himself an online genotyping kit. The results were kind of fun; no big shocks, just a couple of things that made his friends laugh. Last Christmas Andy decided to have some more fun and bought a testing kit for his father, George. Andy had to take his father through the whole genotyping process and was surprised how long it took to explain the basics.

Going through the results together, Andy noticed that there were some major differences between markers on the Y chromosomes of the two men. This was a shock, as the only explanation is that George is not Andy's biological father. Andy asked his sister, Kate, about this and she just got really angry. Naturally, George didn't spot the differences and wouldn't recognise their importance if he did. Andy's mum died a few years ago, so he can't ask her about it.

Should Andy tell his father the result?





If Andy reveals that George is not his biological father, what impact will that have on the family?

What if George has a fertility issue and is not Kate's biological father either?



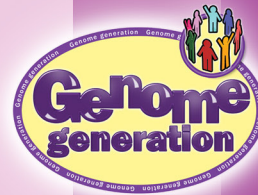
Should online genotyping organisations share their data with researchers? Who else should have access to the data?

Should people be allowed to buy genotyping tests for others, as gifts, without prior consent?



If genotyping is available commercially, is it fair that only those who can afford it get to find out about any preconditions they may have? Should it be available for free through health services?

Should genetic testing be offered over the counter without genetic counselling?



yourgenome.org

What is genotyping?

Genotyping is a process by which a number of genetic variants are determined. These variants do not provide a diagnosis, but are associated with an increased or decreased risk of developing a disease. This type of genotyping cannot provide a complete picture as it does not look at all of the genetic variants possible in a person's genome. Many diseases and characteristics are influenced by multiple genes working in combination, along with environmental factors.

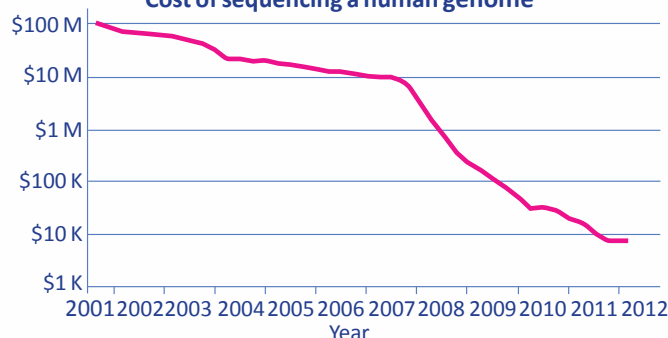


yourgenome.org

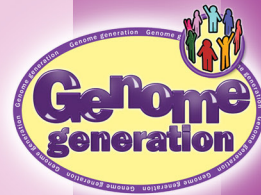
What is genome sequencing?

Genome sequencing looks at the entire sequence of a person's DNA to find ALL of the genetic variants a person may possess. It took the Human Genome Project over 10 years (across many labs) to sequence the 3 billion 'letters' of the reference human genome. However DNA sequencing technologies are developing very quickly and it will soon be possible to sequence our genomes in a short period of time and at a more affordable price.

Cost of sequencing a human genome



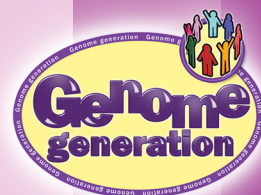
Source: National Human Genome Research Institute



yourgenome.org

Does online genotyping already exist?

It is already possible to get yourself genotyped online for about \$200 (as of February 2012). Subscribers send a sample of their saliva and await their results, which can be viewed online. The testing kits can also be purchased as a gift. It is possible that recreational genotyping will become more widespread in the future.



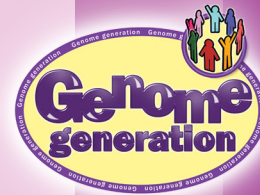
What can online genotyping tests tell us?

Results are presented as an elevated risk, decreased risk or typical risk for developing a number of diseases (in comparison to the average population). The results also present:

- details about a person's likely drug response to a series of medications
- whether a person has carrier status for a variety of genetic disorders
- details of the genetics behind benign physical characteristics, such as likely eye colour, male pattern baldness and earwax type.



yourgenome.org



Chromosome inheritance and gender

Our chromosomes are inherited in pairs – one chromosome in each pair comes from each of our parents. The sex chromosomes 'X' and 'Y', determine our gender. A female has two X chromosomes, while a male has both an X and a Y chromosome. The chromosome we inherit from our mother will always be an X, whereas we can inherit either an X or a Y from our father.



yourgenome.org



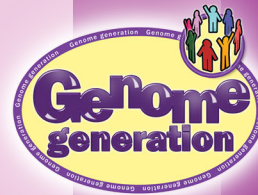
The unique Y chromosome

The Y chromosome is unique because:

- It is something that only males have. This means that the Y chromosome (and all of the genetic markers upon it) is always inherited directly from father to son.
- Most of the Y chromosome is inherited as a complete unit. Therefore, if a male has a clinical disorder linked to his Y chromosome, ALL of his sons will inherit it.



yourgenome.org



Y chromosome microdeletions (YCM)

YCM are a family of genetic disorders caused by a missing gene, or genes, on the Y chromosome. One example is azoospermia, which is associated with having very low levels of fertility, or even infertility. The condition is characterised by the lack of any measurable level of sperm in a person's semen.



yourgenome.org

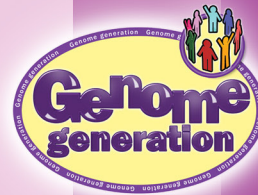


Genetic counselling

There is no telling how an individual may react to the discovery of a predisposition to a genetic disease. Individual genetic tests, carried out by medical geneticists, usually involve genetic counselling. Genetic counsellors work with patients and their families to prepare them for the outcome and to discuss the choices available to them. Online genotyping, carried out in the absence of a genetic counsellor, could lead to anxiety and stress.



yourgenome.org

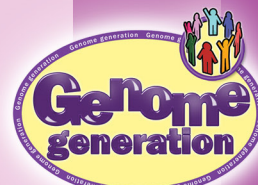


Genetic counselling – where does it end?

Genotyping produces a vast amount of data. On average, each person's genome is likely to contain around 100 different things of interest. Ethicists have calculated that counselling someone through all of this would take around 5 hours per person.



yourgenome.org



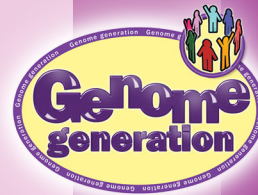
Our DNA

Our bodies are made up of around 50 trillion cells. Most of these cells contain a person's genetic information within a long molecule called DNA (deoxyribonucleic acid). This DNA contains the genetic information that we inherit from our parents.

DNA is made up of specific sequences of letters or bases that encode instructions on how to make proteins. These bases [Adenine (A), Guanine (G), Cytosine (C) and, Thymine (T)] are lined up along two strands of DNA. The DNA is coiled into a double helix structure that resembles a twisted ladder.



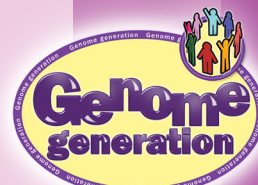
yourgenome.org



yourgenome.org

Gene mutations

A mutation is a permanent change in the DNA sequence. Mutations come in a number of different forms and can change the functioning of our genes. Some mutations may lead to a difference in the amount, or structure, of a protein produced by specific genes. Some mutations may switch genes on at the wrong time, preventing a protein from doing its job, or changing the way it does its job.



yourgenome.org

Our genes

Most of the DNA in human cells is packaged into 46 chromosomes, arranged in 23 pairs. Our genes are arranged along these chromosomes and carry the instructions to make molecules, such as proteins. To function properly, each cell depends on thousands of proteins doing the right job, in the right place, at the right time. Our complete set of DNA, containing all the instructions required to make us, is called a genome.