



yourgenome.org

Olly and Lily are expecting their first child. The baby, currently known as 'Bump', is scheduled to be born by caesarean section on 1st March 2026. Angie, Lily's midwife, is running through the paperwork in preparation for the birth and asks the couple if they would like Bump's genome sequenced.

Angie explains that along with being weighed, measured and having a heel prick test, a simple swab taken at birth will reveal Bump's genetic background, indicating the various conditions he or she may or may not be at risk of developing. The results will be presented on an ID card and will form part of Bump's medical records. Olly thinks it's a great idea but Lily is concerned that the information revealed could lead to discrimination and inequality later in life.

Should Olly and Lily have their baby's genome sequenced at birth?

More and more parents are opting for the test. It means that parents can be prepared for, and avoid, any health issues that may develop later in life. Lots of mums worry about it, but it's perfectly safe and painless.

What if the test shows that there are problems? If they need to show this ID card for everything, my poor child might get shunted out. If you were going to give out a university place or a job to someone, you would choose the healthiest one wouldn't you? Not the one that you know might develop a disease.



Lily
(Bump's mum)

Olly
(Bump's dad)

It's a great idea! It will revolutionise healthcare. Doctors can just check the patient's records and know every kind of treatment they might need further down the line. It will also mean we will be aware of any issues our child might face in the future and make better decisions to overcome them.



Should the test be compulsory? If some people choose to opt out will they become an excluded minority?

Should the data be available for use by researchers or pharmaceutical companies, and if so, should they have to pay for access?



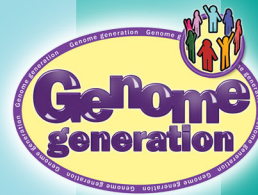
Should the personal data be made available to the police, insurance companies and employers?
Could it lead to a genetic underclass?

New born babies cannot give consent and they cannot receive genetic counselling. Is it acceptable to involve them in something so significant that they have no say over? At what age should they be informed of the results?



If an increased risk to a certain disease is found, how will it change Bump's lifestyle and upbringing?

What are the implications for the health service?
Is it a good use of resources?



yourgenome.org

The human genome

The human genome contains all the information required to make a human. Although any two human beings are more than 99% alike in their DNA, the differences in the other 1% of DNA are what make us unique.



How good are genetic tests at predicting risk?

Genetic tests are a useful tool for predicting disease risk, where family history or other factors have already shown that a person's risk is higher than normal. However, there is concern that these tests will be much less accurate when used for everyone, regardless of their family history. It could result in many people getting false negative or false positive results.



yourgenome.org



EMBL-EBI

WELLCOME
GENOME
CAMPUS
PUBLIC
ENGAGEMENT

wellcome trust
sanger
institute

yourgenome.org



EMBL-EBI

WELLCOME
GENOME
CAMPUS
PUBLIC
ENGAGEMENT

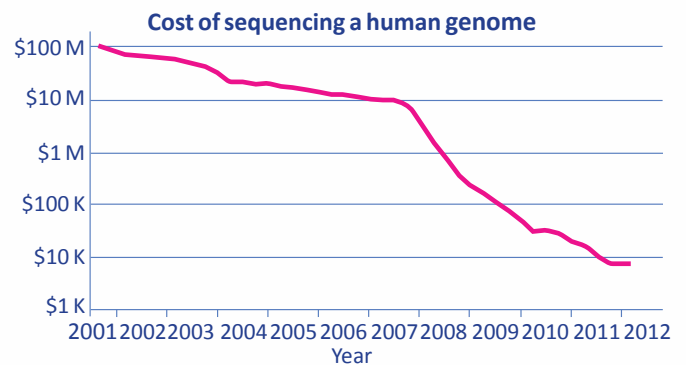
wellcome trust
sanger
institute

yourgenome.org



Affordable genome sequencing

The cost of sequencing a whole human genome is plummeting and it could soon become affordable for individual genomes to be sequenced as part of routine healthcare. Each person's genome sequence differs in a possible 3-4 million places. Some of these variations will be relevant to the development of diseases, and others could reveal how people are related, but for the vast majority the clinical relevance is uncertain.



Source: National Human Genome Research Institute



Impact on the health service

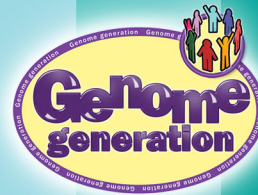
Genomics generates a vast amount of data. Each human genome is made up of a sequence of 3 billion chemical 'letters'. Storage of one person's genome sequence could require up to 1.5 gigabytes of space. Information about medical histories would also need to be stored. It will be costly to produce the computer infrastructure to store, analyse and provide access to this data. It will require substantial investment to develop the expertise and facilities needed.

EMBL-EBI

WELLCOME
GENOME
CAMPUS
PUBLIC
ENGAGEMENT

wellcome trust
sanger
institute

yourgenome.org



How much can genetics tell us?

Our personalities, behaviours and disease predispositions are the result of complex interactions between our genes and our environment. This complexity makes it very difficult to make solid predictions about an individual's risk of illness and disease.



yourgenome.org

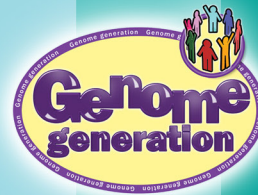


False positive tests

Genetic tests are used to predict disease but they are not 100% accurate. If evidence of genes that can cause disease is found, the result is said to be positive. If the relevant genes are not found, the result is said to be negative. But occasionally, false results occur when genes are present but not detected (false negatives) or shown to exist when they do not (false positives).



yourgenome.org



The National DNA Database

The UK's National DNA Database contains DNA data from 5.5 million people who have been arrested for a criminal offence. The information is limited to the 20 specific points in a person's DNA that are used in DNA profiling. Although it is highly useful for catching offenders, the storage of data from those who were not charged with an offence has been controversial.



yourgenome.org

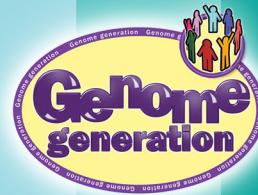


Genetic discrimination in the UK and Europe

The UK has no specific legislation against genetic discrimination. However, use of genetic tests in employment is covered by the Employment Act 2010 which restricts what employers can ask about in pre-employment medical checks. Discrimination based on genetic characteristics is also prohibited in the European Union under the terms of the Lisbon Treaty.



yourgenome.org



Genetic discrimination in the USA

The United States of America has prohibited the use of genetic information in health insurance and employment through the Genetic Information Nondiscrimination Act (GINA) of 2008. Healthy individuals with a genetic predisposition to a particular disease cannot be prevented from getting health insurance or charged higher premiums simply because of their genetics. It also stops employers from using genetic information when making recruitment, promotion or dismissal decisions.



yourgenome.org



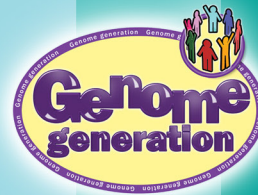
Our DNA

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.