

GENOME GENERATION

Guide to scenario 6

SHOULD A BABY HAVE ITS GENOME SEQUENCED?



This scenario is set in the future. It assumes that we are at a point where whole genome sequencing can be carried out cheaply and quickly.

Summary: In this scenario expectant parents are asked whether they would like to have their child's genome sequenced after it is born. The information would form part of the child's ID card.

Initial question: Should the parents have their child's genome sequenced?

Key issues: Incidental findings; Access to data; Impact on families; Would you want to know?

SENSITIVE ISSUES & GUIDANCE

This scenario may be more suited to A-level students.

This scenario deals with the impact of whole genome sequencing and the potential information contained within the genome. It also raises the issue of data storage and who should have access to our data. It is an opportunity to explore the wider social issues of the personal genome.

QUESTIONS TO DISCUSS

What are the implications of a child having its genome sequenced at birth?

What if the baby has a genetic condition, revealing that one or both of the parents is also a carrier of a condition?

If the child has a predisposition, they can opt to make the right lifestyle choices and undertake monitoring, which could improve their prognosis.

If a child has a worrying predisposition, will this cause anxiety for the parents and the child in later life?

Will anxious people with certain genotypes become vulnerable to commercial organisations wishing to sell them interventions or preventative medications that may not be necessary?

Issues of consent

If a child is genotyped at birth, they cannot give their consent or receive genetic counselling. Should parents be allowed to take this decision for their child?

Will a genetic ID card lead to discrimination?

Should everyone be genotyped? If the results are stored on an ID card – should it be compulsory for all?

How should the results be used? What if people are expected to provide their ID card and sequence results in order to secure employment, insurance, mortgages, university placements etc.? Will those with certain genotypes suffer discrimination? Or will those who opted out of testing be at a disadvantage?

Cost implications

Who should pay? The impact on the NHS in terms of building the infrastructure for the widespread testing and storage of this data is considerable. Conversely, is it unfair for only those who can afford it to opt in?

Who should be allowed access to the genetic data?

Should researchers and pharmaceutical companies be allowed to access the data? Widespread genotyping could help the development of effective personalised medicines and interventions.

FURTHER INFORMATION – WHAT THEY MIGHT NEED TO KNOW

The \$1000 dollar genome

Early in 2012, two rival companies announced advances to their sequencing machines that meant a human genome could be sequenced within a day at a materials cost of \$1000. \$1000 had been seen as the breakthrough target that would open the door to routine genome sequencing for healthcare. Although the cost and time to sequence a human genome has decreased dramatically, the opposite has occurred for analysis and interpretation of the information which is now the major bottleneck for whole genome sequencing.

Who is thinking about the issues surrounding genome screening programmes?

The UK government has set up advisory boards to address the issues around whole genome sequencing. In response to the 2009 House of Lords Inquiry into genomic medicine, the Government set up the Human Genomics Strategy Group. Their remit is to monitor advances in genetic and genomics research to evaluate their benefit to healthcare services in the NHS, and develop a vision for genomics in the NHS.

What can we learn from a genome?

The genomes of apparently healthy people, as well as those with a genetic disease, will contain around 3-4 million sequence variants. These can be broadly classified into three groups:

- Those that have no effect (i.e. neutral variation)
- Those that have an effect on the normal phenotype (e.g. height, eye colour)
- Pathogenic variants or mutations which either cause, or predispose to, disease.

FURTHER READING

The following news story can be used in addition to the Info cards to provide extra background information to help students understand the major issues raised in this scenario.

Parents 'want child gene tests'

www.bbc.co.uk/news/health-13099090

FURTHER INFORMATION FOR STUDENTS

Students can find out more information about whole genome sequencing from the following websites:

PHG Foundation

The PHG Foundation is a UK charity that aims to influence public policy on the use and implementation of biomedicine and genomics in health care settings.

www.phgfoundation.org/project/wgs

Article by Katherine Harmon in Scientific American

This article discusses whether whole genome sequencing can be used to predict your risk of developing disease.

www.scientificamerican.com/article/whole-genome-sequencing-predict-disease/

Genetic profiling of newborns: ethical and social issues

An article discussing the ethical and social barriers to the introduction of genetic profiling of babies at birth as a public health service.

www.nature.com/nrg/journal/v7/n1/full/nrg1745.html

FOLLOW UP ACTIVITY

1. Organise a screening of the film **GATACCA** for your year group. This can be followed by a group debate on the topic "This house believes that genome sequencing is the future for public healthcare".
2. Ask students to prepare a report on the possible advantages and disadvantages of introducing genome sequencing for all babies at birth. Make sure they consider the impact this information has on the individual and society.

They can choose from one of the following styles for their report:

- website/blog
- magazine article
- TV chat show.