

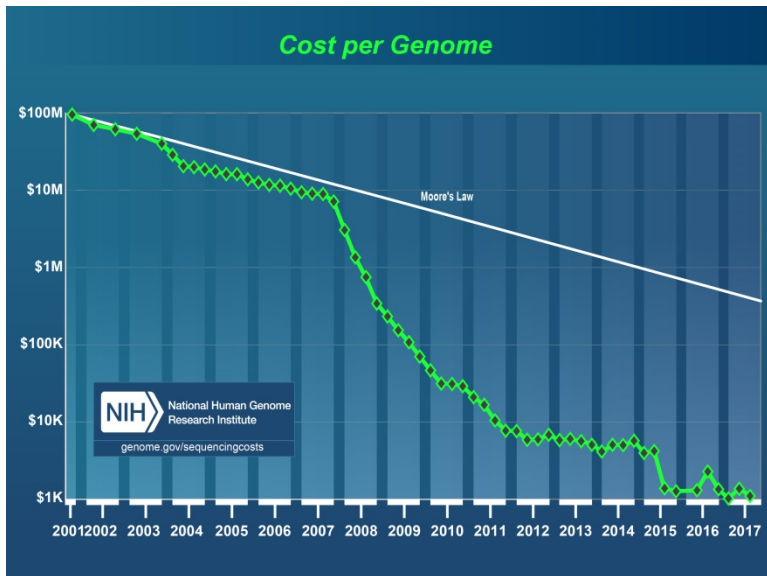
Ethical Issues in Genomic Medicine

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Ethical Issues in Genomic Medicine

- Context
- Importance of Ethics
- Ethical Issues
 - Consent
 - Additional Findings
 - Variants of Uncertain Significance
- The Future of Ethics



Importance of Ethics

The screenshot displays two articles from The Guardian. The top article, 'Gene therapy for the unborn', features a photograph of a family and a headline that reads 'Gene therapy for the unborn'. The bottom article, 'Privacy and the 100,000 Genome Project', is categorized under 'Science' and 'Political science'. Its headline asks, 'Privacy and the 100,000 Genome Project' and includes a sub-headline: 'As the Department of Health starts to draw a map of thousands of genomes, will it keep its promise to anonymise our data? Edward Hockings and Lewis Coyne investigate'. The website's navigation bar includes sections like 'UK', 'world', 'politics', 'sport', 'football', 'opinion', 'culture', 'business', 'lifestyle', 'fashion', and 'environment'. The Guardian logo is prominently displayed in the center of the page.

Gene therapy for the unborn

Privacy and the 100,000 Genome Project

As the Department of Health starts to draw a map of thousands of genomes, will it keep its promise to anonymise our data? **Edward Hockings** and **Lewis Coyne** investigate

Data and computer security

When data gets creepy: the secrets we don't realise we're giving away

We all worry about digital spies stealing our data - but now even the things we thought we were happy to share are being used in ways we don't like. Why aren't we making more of a fuss?

Ben Goldacre

Marketing Manager | **Floral Designer**

Ethical Issues in Genomic Medicine

Case Study

- Results of genomic investigations in a patient investigated for neuro-developmental delay reveal a mutation in a gene that increases the chance of a brain tumour.
- The relative risk that this mutation appears to confer is very high but the absolute risk is <15%.
- Records of consent taken at the time of testing reveal no mention of the possibility of tumour/cancer risks being found.

Questions to consider

- What is the role of consent?
- Are the researchers obliged to disclose AFs?
- Does this change if the researcher is also a clinician?
- How can the clinical significance of AFs be determined?
- What significance level should determine disclosure?
- What should happen if adult-onset risks are found in children or pre-natally?

Consent

Valid consent

1. **Informed** (the patient is provided with relevant information to make the decision)
2. **Competent** (the patient is competent (has the capacity) to give consent)
3. **Voluntary** (the patient is able to make a free choice, i.e. no undue influence or coercion is applied to the decision that the patient makes)

Consent

- What is relevant information in this context?
- Communicating and understand of uncertainty.
- Widespread background belief that genetics is clear-cut and highly deterministic.
- The possible need to investigate relatives needs to be incorporated.
- Obtaining adequate consent to disclose an AF for which there is no prior suspicion is problematic.

Additional Findings

- Pertinent findings = findings that have been sought with the purpose of answering a particular question
- Additional findings = findings that may/may not have potential health implications, that are discovered during the course of investigation, but are beyond the aims of the original test or investigation
- No consensus about disclosing AFs, or about how participants can be helped to make decisions about and give consent to receive them



Image: Elin Svensson

Case Study

- A mutation in the *BRCA1* gene was thought to confer a high risk for breast and ovarian cancer.
- More recently, evidence suggests that it is a benign variant and that the surveillance and interventions offered were therefore wrongly directed.
- This evidence has come to light through international efforts and database linkages.
- Although those with the variant have previously been advised they are at high risk they, their relatives and future individuals can now receive more up to date clinical advice.

Variants of Uncertain Significance

- Findings in genomic medicine may be scientifically significant, or not.
- ‘Scientifically significant’ - there is robust statistical evidence of a relationship between a genetic variant and a particular phenotype.
- If there is insufficient evidence to support a genotype-phenotype relationship then the finding is often referred to as a variant of uncertain significance (VUS).

Large Cohort Databases

“New data which is added will mean that we understand more about the genomic factors behind illnesses and more about what we can do to help patients and improve outcomes. As a patient, I could be getting my diagnosis ‘live’ from a comparison of all genomic data available.....The more people have contributed their data, the better the results for any one individual. So use of data in this way benefits all patients both as a group and individually.”

Chief Medical Officer (2016)



Questions to consider

- Is health-system learning research or clinical practice and does it matter?
- Should patients be expected to contribute and share their healthcare information for the mutual benefit of themselves, other patients and the wider population?
- Should genomic data be routinely collected for reasons of a public good?
- Should the overemphasis in policy and regulation on privacy and ownership of data be challenged to enable data sharing for patient benefit?

The Future

- New challenges from technological advance in healthcare
- 20th Century ethics was not formed to address these intense challenges
- New forms of research present unprecedented problems for the core assumptions and practices of ethics as currently understood
- This requires a paradigm shift and change of scale in ethics research and a closer more constructive relationship between ethics and science

Examples of new challenges to ethics

- The excessive primacy given to consent
- Inability to deal with risk and probability
- Lack of clear concept of collective responsibility
- Responsibility and good ethical practice in context of health uses of machine learning

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