

# HUMAN GENOMICS

Applications, benefits & concerns of human genomics

## GENOME ANALYSIS



### RESEARCH

- Genome variations in populations
- Functions of genes
- Disease causing genes
- Genome & environment interaction
- Developing clinical and other applications of DNA sequencing technologies



### HEALTHCARE

- Diagnostic testing
- Predictive testing (e.g. BRCA genes)
- Newborn screening
- Carrier testing/screening
- Prenatal testing
- Pharmacogenomic testing



### CONSUMER

- Genetic tests are sold online by companies. Some of these tests are unreliable. Companies offer:
- Ancestry testing
  - Disease predisposition testing
  - Pharmacogenomic testing



### FORENSIC

- Kinship determination (e.g. paternity tests)
- Analysing crime scene DNA using forensic databases
- Using crime scene DNA to determine features of an unidentified suspect

## GENE EDITING



### RESEARCH

- Studying gene functions
- Learning about the development of diseases (e.g. cancer)
- Understanding embryo development (from research on embryos)
- Developing clinical applications



### HEALTHCARE

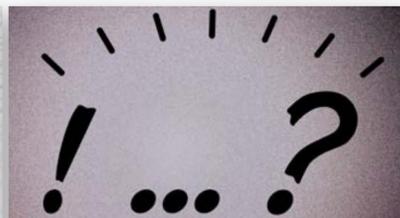
- No gene editing treatments are available yet. But some are in clinical trials. For example, treatments for:
- blood diseases
  - HIV
  - muscular dystrophy
  - cystic fibrosis

## IMPACTS



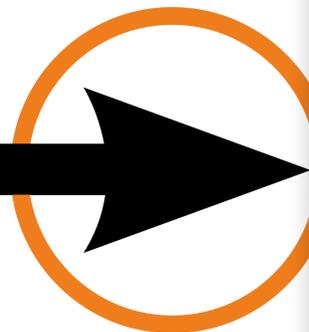
### BENEFITS

- Earlier and/or faster diagnosis
- Prevention of diseases
- Increased treatment options
- Tailored treatment (pharmacogenomics)



### RISKS

- Genetic discrimination
- Misinterpretation of results
- New eugenics (selecting people with(out) certain traits)
- Instrumentalization of embryos
- Unintended social changes (e.g. less compassionate society)
- Adverse impacts on disabled people



### PEOPLE AFFECTED

- Patients with genetic disease
- People with higher risk of genetic disease
- People taking certain medication
- Newborns
- Pregnant women
- Suspects of criminal offenses
- Donors of research samples/data
- Participants of clinical trials
- Consumers of genetic tests
- Relatives of people undergoing genetic tests

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Technology, ethics and human rights

**We will help address some of these issues by:**

- Analysing the legal, ethical, social & economic impacts of human genomics
- Connecting with citizens to understand their concerns via surveys in 11 countries & panels in 5 countries
- Consulting experts and discussing ethical & human rights issues
- Developing ethical codes & protocols

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