Genomics Education Programme

NHS
Health Education England

Personalised medicine: innovations and interventions to improve patient care

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Genomics Education Programme

Developing people
for health and
healthcare
www.hee.nhs.uk

Overview of presentation

- Genomics – use in personalised medicine
- How do nurses play their part?
- Rare disease
- Cancer
- Infectious disease
- Learning opportunities

What do we mean by personalised medicine?

"A move away from a 'one size fits all' approach to the treatment and care of patients with a particular condition, to one which uses new approaches to better manage patient's health and target therapies to achieve the best outcomes in the management of a patient's disease or predisposition to disease"

- Not a new concept
  - Tailor care to the individual
- New technologies allow us to do this at a greater level
  - Genomics

Future or now?

Genome revolution targets treatments for common cancers

STEVE CONNOR
SCIENCE EDITOR

NHS patients in England will be the first in the world to participate in an ambitious government-led programme to sequence 100,000 genomes as part of a "paradigm shift" in healthcare focusing on the genetic causes of disease.

First genome project diagnoses give hope to two four-year-olds

Pair are first to benefit from the 100,000 Genomes Project, launched in 2014 to improve diagnosis and treatment of rare genetic disorders and cancer

What is genomics?

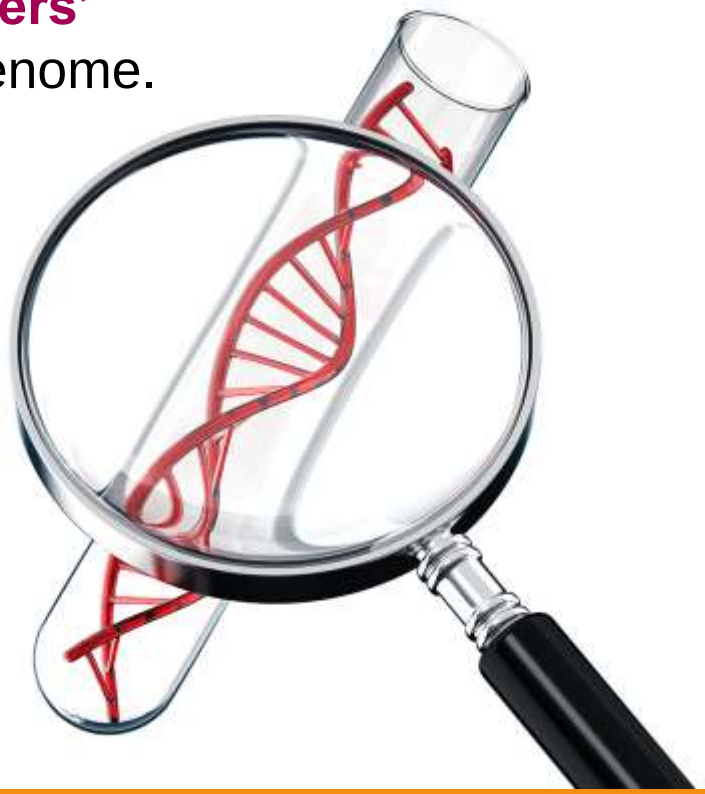
- Genomics is the study of the **whole genome** and how it works.
- Your genome is **1** whole set of **all** your genes **plus** all the DNA between the genes.
- Put very simply, **genomics = all your DNA**;

  genetics = effect of 1 gene
- There are **20,000** genes – and they make up just **2%** of your genome.
- There is a **copy of your genome** in almost every cell of your body.

Don't forget, humans are not the only organisms that have a genome, ***pathogens (eg bacteria and viruses) have their own genome.***

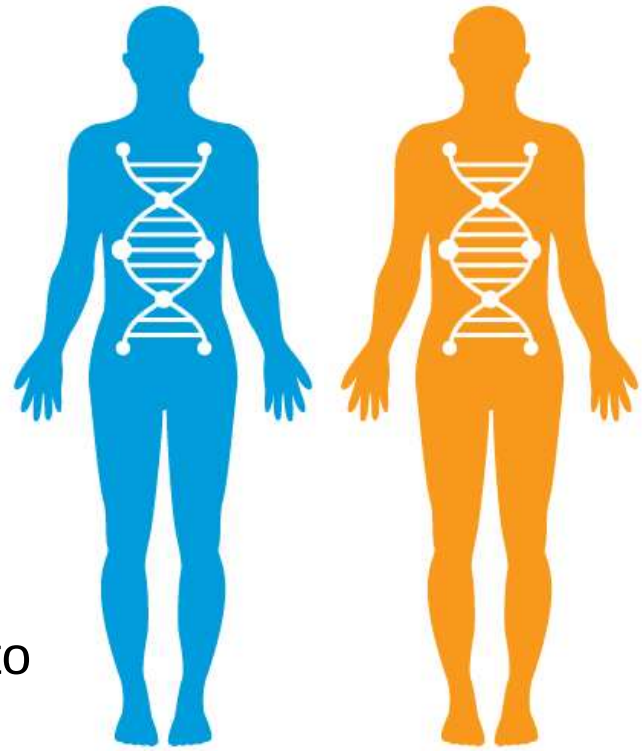
How can we look at our genome?

- Whole genome sequencing is a technique that reads the entire DNA sequence, the **3 billion 'letters'** (A, T, C and G), that make up the human genome.
- The first whole genome took **13 years** and **£2 billion** to read every letter - today it takes around **2 days** and **£1,000.**
- Looking at the whole genome can help us to **understand** how disease develops and which treatments will be most **effective**.



How do changes in DNA affect health?

- The DNA sequence of any two individuals is around **99.9% identical**.
- The remaining **0.1% variation** may:
  - have **no effect** or be the cause of **normal variation** (e.g. hair colour);
  - alter control of **genes** or action of gene products (**proteins**);
  - result in different responses to **drugs**; or
  - **cause** a genetic condition (e.g. cystic fibrosis) or an **increased susceptibility** to a particular disease (e.g. diabetes).



Genomics is being used to…

- ✓ **Help with the prediction and prevention of disease**
  - Genomic information can identify those at risk of having a genetic condition. It can also help with the identification and development of new treatments to reduce the burden of disease.
- ✓ **More precise diagnosis**
  - Many conditions have the same clinical signs and symptoms, but are caused by different genetic variants.
- ✓ **Targeted and personalised interventions**
  - Identifying genomic variants can help to select effective treatment (based on condition itself) and predict how a person will react to the medication.
- ✓ **Track epidemics**
  - Identify specific pathogens and whether an outbreak originated from a single source.

How do nurses play their part?

1. **Identify individuals who have, or could have, a genetic condition**
   Nurses may be involved in identifying people who could have a genetic condition by:
   - Their involvement in **screening programmes**:
     - Early in life = newborn screening (blood spot test)
     - Later in life = Bowel cancer screening programme
   - Being **observant**:
     - recognising signs and symptoms of genetic conditions
   - Taking a **family history**:
     - identifying conditions that run in the family

How do nurses play their part?

2. **Make a referral for additional clinical input or social /community services**
   Nurses may refer individuals and their families (where appropriate) to:
   - Other clinical services (e.g. GP, specialist services)
   - Social and/or community services
   - Support groups

How do nurses play their part?

3. **Receiving consent from individuals**
   Nurses may receive consent from individuals with regard to:
   - Genetic/genomic tests
   - Other clinical tests
   - Treatment
   - Research projects (including clinical trials)

How do nurses play their part?

4. **Provide support for the family**
   Nurses will be able to support individuals and families by:
   - Providing ongoing support to parents going through the '**diagnostic odyssey**'
   - **Clarifying** information about genomics and genomic testing, and **directing** them to additional information sources
   - Noticing if people are **struggling to cope** with or **understand new information**, such as a new diagnosis
   - **Supporting** parents whose child has special needs

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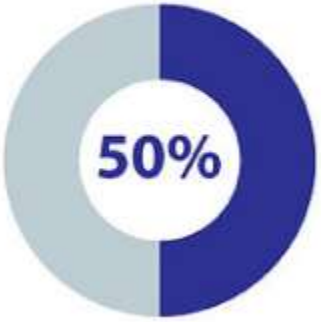
Rare disease

Rare disease: facts and figures

- Affects **less than 1 in 2,000** people
- Collectively affects **1 in 17 of the population**

50% of newly diagnosed cases of rare diseases are in children



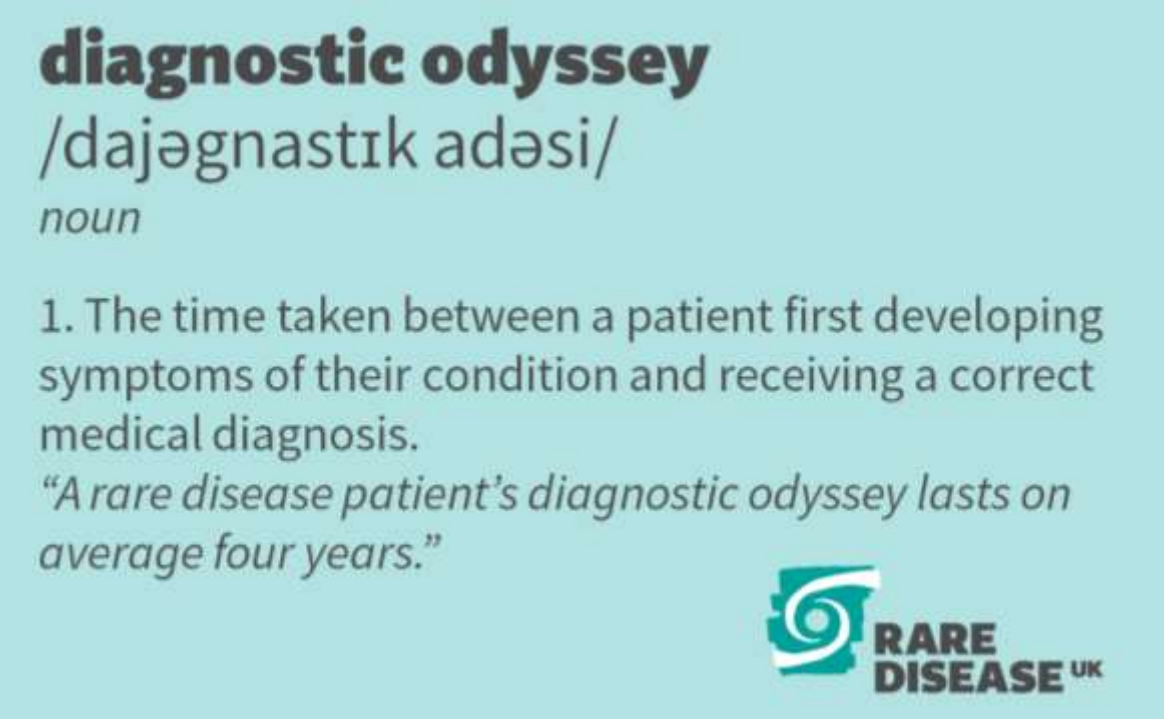
There are between **5,000** and

8,000

different rare diseases...

… and **80%** of them have a known **genetic origin**

The diagnostic odyssey



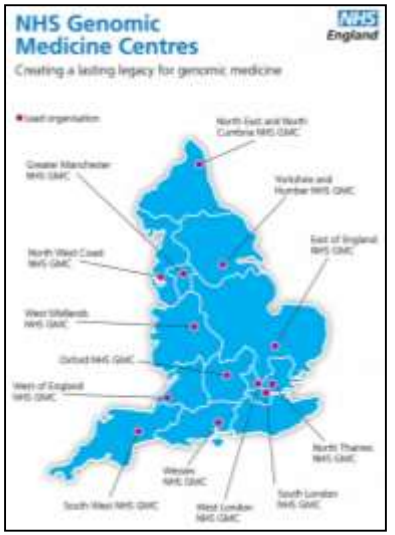
Friday 27 April, celebrating Undiagnosed Children's Day

Jessica's story

Jessica has a rare disease.

- Epilepsy and affects her movement and general development
- Enrolled on the **100,000 Genomes Project**
- 100,000 Genomes Project run by Genomics England
- Delivered in NHS through 13 Genomic Medicine Centres

Genomics england



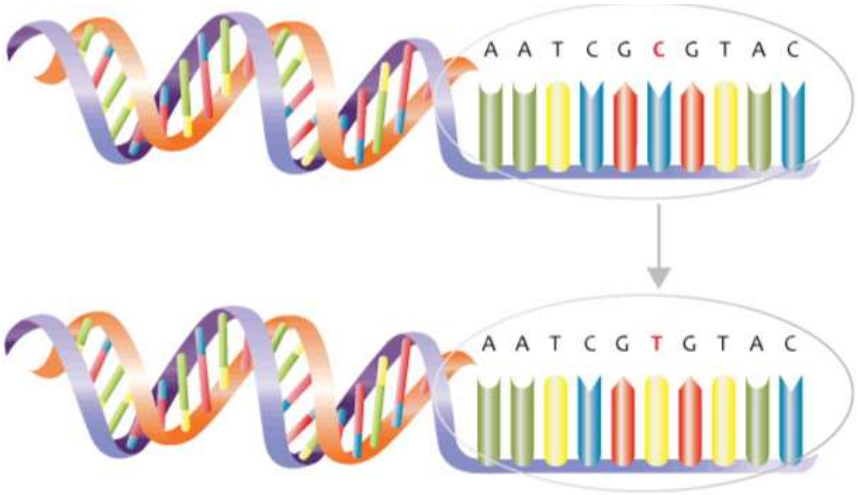
www.genomicsengland.co.uk

Using genomics to find an answer?

DNA from Jessica → Sequenced the genome ↓

What's different in Jessica's genome? ↓

6.4 million variants



From 6 million to 1

Identifying which variant(s) relate to the condition

↓

Filtering – bioinformatics

Use information such as:

- Clinical data – genes associated with signs and symptoms
- Scientific data – changes disruptive to gene function
- Compare Jessica's sequence to that of her parents

↓

One variant: *SLC2A1* gene – **Glut1 deficiency syndrome**

Ketogenic diet can reduce number of seizures

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What this means for Jessica

"The outcome has taken the uncertainty out for us and the worry of not knowing what was wrong. Now that we have this diagnosis there are things we can do differently straight away. A special diet means her medication can decrease and her epilepsy be more easily controlled."

Improving outcomes through personalised medicine, NHS England

Image courtesy of Genomics England

Introduced into the NHS – equity of access to testing

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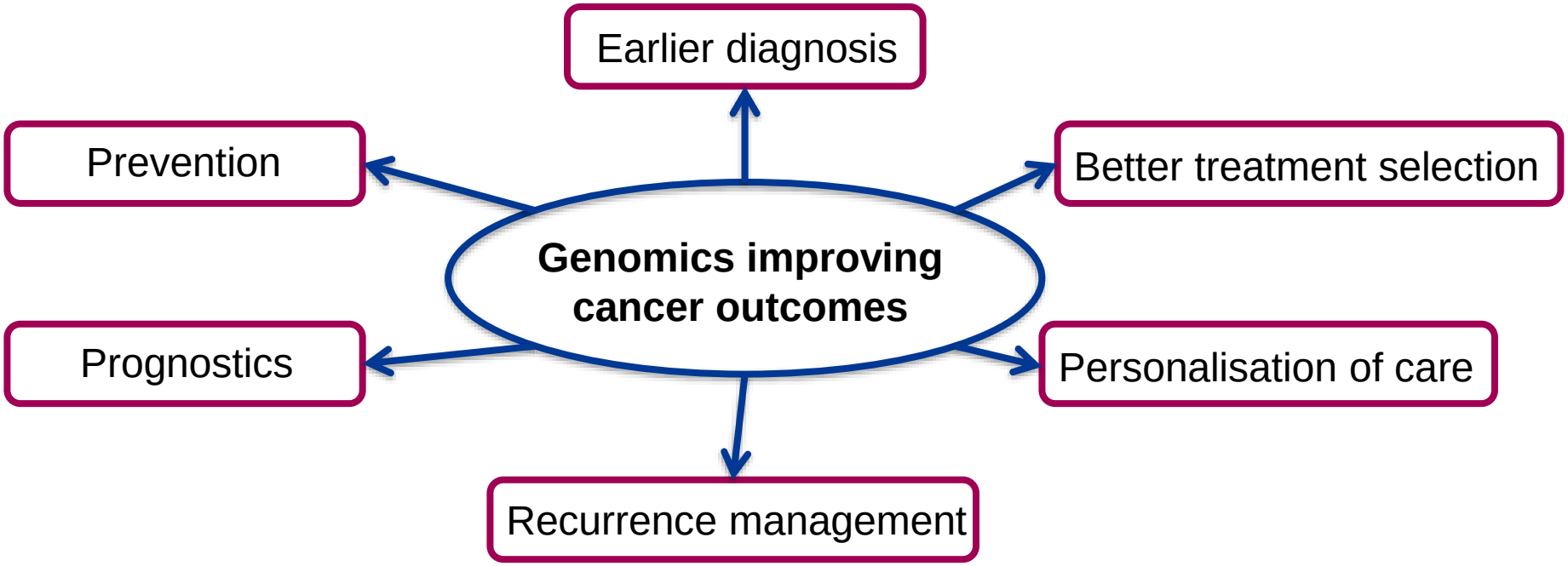
Cancer

Cancer: a disease of the genome

- We accumulate changes in our DNA over time
- Some changes lead to development of cancer

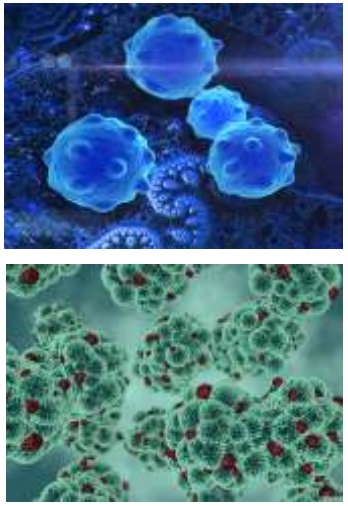


Identifying changes in tumour DNA

Tumour DNA *and* Constitutional DNA

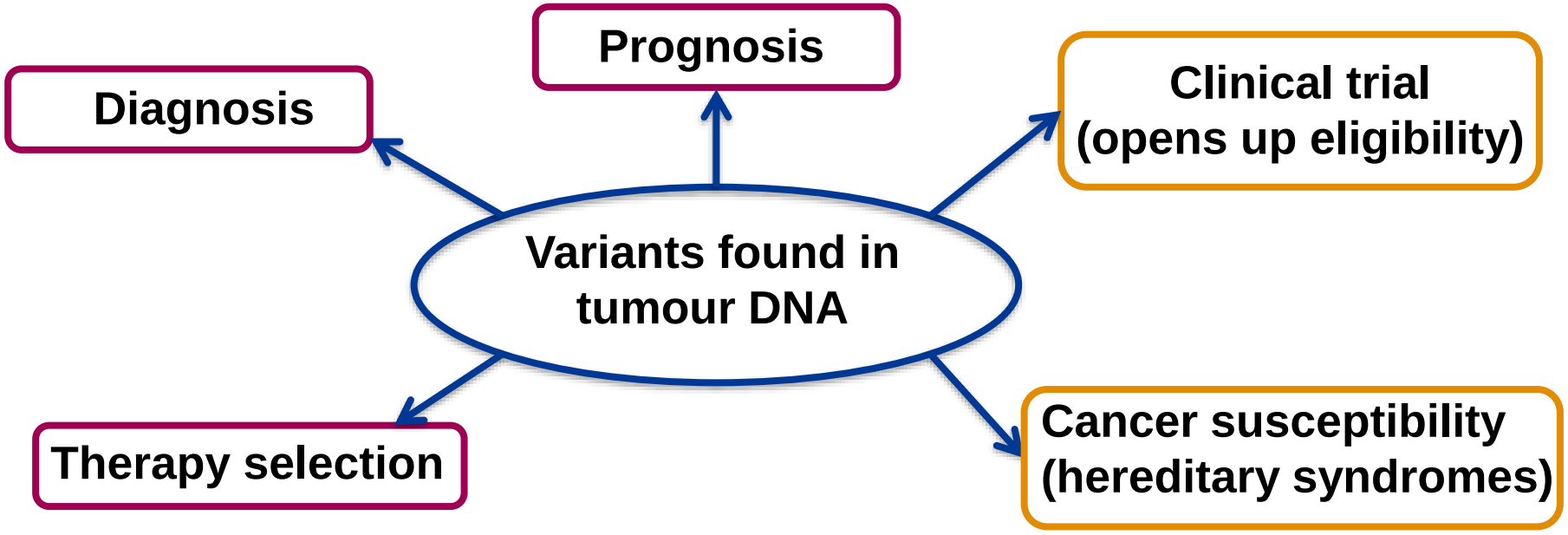
↓ ↓

Sequence Sequence



- **Compare the sequences** – looking for changes in the tumour DNA not present in the constitutional DNA
- **Looking for patterns** of ‘mutations’, which are known as molecular signatures

What can we do now? Actionable information for cancer patients



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Infectious disease

Genome = pathogen

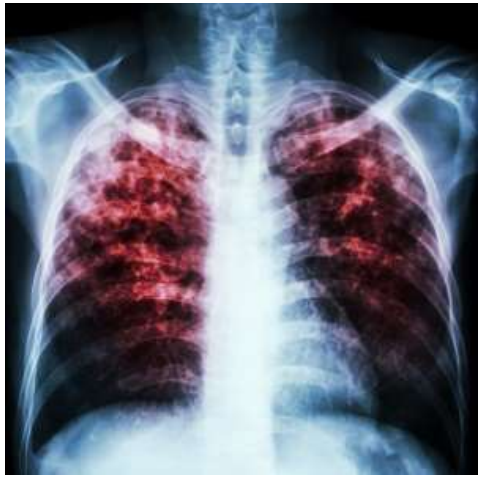
Pathogens have their own genome. Application of genomics include:

- Diagnosing an infection
- Investigating outbreaks
- Identifying most appropriate treatment
- Monitoring antimicrobial resistance
- Developing intervention, including vaccines

Tuberculosis

Aim of testing symptomatic (asymptomatic) individuals:

- **Diagnosis** – identifying mycobacterial species
- **Ensuring correct treatment** – 'drug resistant' strains
- **Minimise spread** - detecting source of outbreak and spread amongst cases



Traditional testing ~8 weeks end-to-end, and reports are issued at each stage of the workflow.

Use of genomics in TB management

- First time whole genome sequencing being used as a diagnostic solution at this scale.
- Allowing **faster** and **more accurate** diagnosis
  - Identify species by comparing against a catalogue of sequences
  - Predict drug susceptibility identifying specific variants
  - Link to previous cases so that can infer origin of outbreak
- Time frame **< 7 days** post liquid mycobacterial culture with a single report issued.
- Slowing the spread of disease and boosting fight against anti-microbial resistance.

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Learning opportunities

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How can nurses find out more?

HEE Genomics Education Programme

- The GEP was established to ensure ***all*** NHS staff have the right knowledge, skills and experience to deliver the genomic medicine service.
- Offers a broad range of **educational and training opportunities**, from formal taught courses, to short online courses and ‘just in time’ educational resources.
- Funded to March 2020

www.genomicseducation.hee.nhs.uk

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Useful resources for nurses

► Public health masterclass

- Filmed event featuring 5 expert speakers covering key genomics topics including prenatal testing and ethics
- Suitable for all healthcare professionals
- Available on Vimeo and YouTube

► Family history films

- Based on three patient scenarios, these films show the questions that can be used to gather a family history, and demonstrate how the genetic pedigree is constructed.
- Available on Vimeo and YouTube

► Genomics 101 series

- Series of 8 short online modules aimed at health professionals with limited or no genomics knowledge.
- Designed to take the learner from genes and proteins, to genomics in clinical practice.
- First course **launching Spring 2018**

► Genomics Game

- Board game for face-to-face interactive group learning activity
- Developed primarily for nursing workforce
- Aligned to current curricula
- 100 games distributed to NHS GMCs and HEIs

The Genomics Education Programme

A funded NHS initiative that ensures healthcare professionals have the genomics knowledge they need for the future.

Training existing and future NHS healthcare professionals

Supporting and enhancing the 100,000 Genomes Project

Bringing better care and lasting benefits to patients

Genomics vs Genetics

Genomics

- The study of an organism's complete set of genetic information.
- 'Genome'- the complete genetic information of an organism.
- The genome includes both genes and non-coding DNA.

Genetics

- The study of heredity
- The study of the function and composition of single genes.
- 'Gene'- specific sequence of DNA which codes for a functional molecule.

Key partners

Department of Health | Public Health England | NHS England | NHS Health Education England

Genomics England | NHS Genomic Medicine Centres | GeCIP domains

Our educational resources

Master's in Genomic Medicine

Multidisciplinary qualifications that can be applied to research and clinical practice.

Short online courses

Engaging and interactive courses that are tailored for professional development.

Training tools

Digital training to directly support the 100,000 Genomes Project.

Multimedia

A collection of informative videos, images and animations that assist in genomics learning.

Events

Our team facilitates a series of genomics workshops and events that run alongside the education programme.

> *"At times it feels like we are learning a new language, but it is definitely worth it."*
>
> - Katherine, Nurse

Genomics is for everybody

Highly specialised workforce

- Clinical and laboratory genetics
- Molecular pathology
- Molecular haematology
- Bioinformatics

Specialised clinical workforce

- Cancer surgery and medicine
- Cardiovascular, diabetes and neurology specialist teams

Wider clinical workforce

General practice and other healthcare professionals

Raising awareness

Managers, commissioners, patients and public

> *"The programme is part of a bigger movement across the country, genomics and personalised medicine is the future."*
>
> - Brenda, Masters Student

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