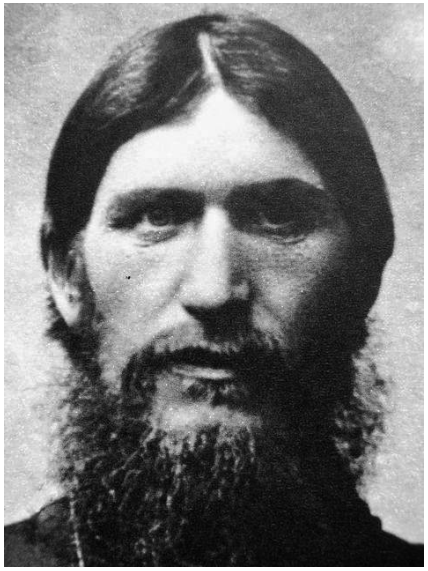
GENETICS

The science of inherited disease



The 100,000 Genomes Project
What's a genome?

Human Genome Project
First draft sequence 2000

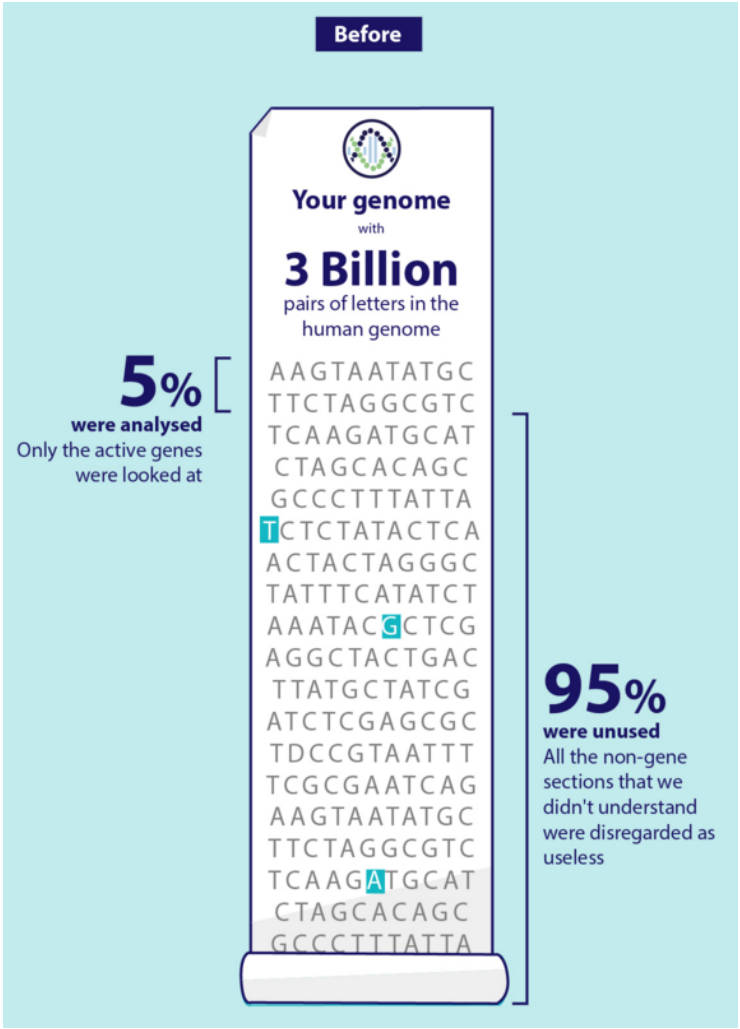
- They thought they'd find a minimum of 100,000 genes
- They found 20,000
- That's the same number as a starfish

How much of your DNA is genes?

- 80%
- 50%
- 20%

Genes: less than 5% of DNA

- Rest of the DNA used to be ‘junk’, wasn’t thought to be important.
- Now know it’s important for regulation, switching genes on and off at the right time.
- Still much more to learn about what it all does.



Genomics

The science of

- **all** the DNA in the genome
- And how it's sequenced, analysed and interpreted

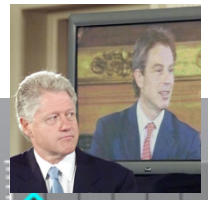
How different are our genomes?

- More difference between individuals than between different races
- We are 99.8% identical
- But 0.2% is still 3 million differences. Its what makes you you

The first human genome sequence

- 26th June 2000 - Cost $3.2 billion and took 13 years

Cost of sequencing



Today it takes 24 hours costs less than $1000

The 100,000 Genomes Project

Genomics england

1. To bring benefit to NHS patients

2. To create an ethical and transparent programme based on consent

3. To enable new scientific discovery and medical insights

4. To kickstart the development of a UK genomics industry

The 100,000 Genomes Project

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…….in a nutshell

- The 100,000 Genomes Project is a research project, a clinical service and an NHS transformation project in one
- 100,000 whole genomes from 70,000 patients and their families with rare disease or cancer, completion by 2018
- Based on consent
- Recruitment through 13 NHS Genomic Medicine Centres

The 100,000 Genomes Project

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……in a nutshell

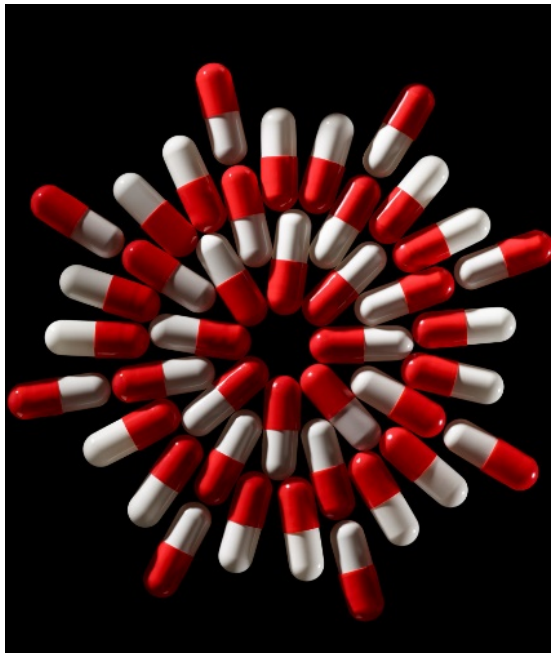
- Sequence data linked to health data to diagnose
- Sequence data, health data and lifelong medical record data put into dataset available for research including access by drug companies
- NHS will be first health system in the world to embed genomic medicine in mainstream healthcare
- At the cutting edge of science and very, very hard.

The 100,000 Genomes Project

The beginning of something amazing

Personalised Medicine

New era of personalised medicine



THE 4 Ps:

1. **Prediction** and **prevention** of disease
2. More **precise** diagnosis
3. Targeted and **personalised** interventions
4. A more **participatory** role for patients

The future
- starts with beauty

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GENEU®

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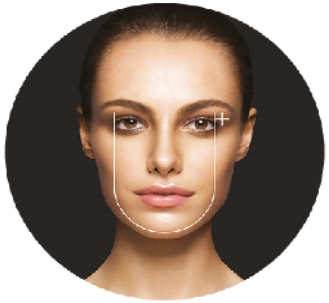
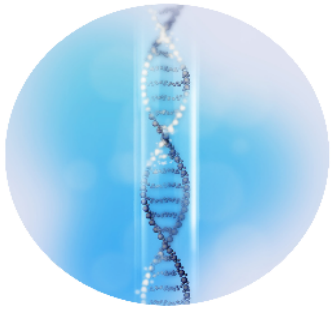
OUR GENOMICS REVOLUTION

The search for the secret to eternal youth is centuries old, but GENEU has turned to the latest in science for the answer. Our scientific research recognises that 60% of your skin ageing can be attributed to inherited genes, and 40% to lifestyle factors*. This is why our unique service starts by offering customers a DNA & Lifestyle Test to look at the two key genes responsible for skin ageing, and includes a lifestyle assessment. Image of test This allows us to determine your skin's profile based on your genes and lifestyle, which forms your U* skin profile.

What makes this further unique, is that it's actually a very quick and effective process. Once your DNA sample is collected and received by us, our scientists analyse it in our quality-assured gold standard London laboratory, and using our unique algorithm we combine the results with your lifestyle assessment.





Significant scientific research has been conducted in collaboration with researchers at Imperial College, London to understand the link between the variations we detect in the two ageing genes and the lifestyle factors and how they relate to the optimal active ingredients and the best concentrations to use in our personalised serums. This research forms the scientific basis of our skin care. These relationships have been used to create the algorithm we use, into which we provide the genetic results and answers to your lifestyle questionnaire. The algorithm provides the result that creates your recommended U* skin profile. This lab-process only takes 2 working days, so very quickly, we'll have your results and can invite you to have a private results consultation with our scientific advisor, who will personally-prescribe and provide for you two recommended GENEU serums, each with the right concentration of advanced active ingredients for your skin. Your two prescribed serums will provide you with the anti-ageing support you need, to delay the appearance of the formation of wrinkles and fine lines, whilst optimising your skin's health through hydrating benefits.

Prediction & Prevention

Familial Hypercholesterolaemia (FH)

- Of the 120,000 people in the UK with FH only 15% – less than 1 in 5, know they have the condition
- At least 28,000 children in the UK have FH but only 600 of these are known

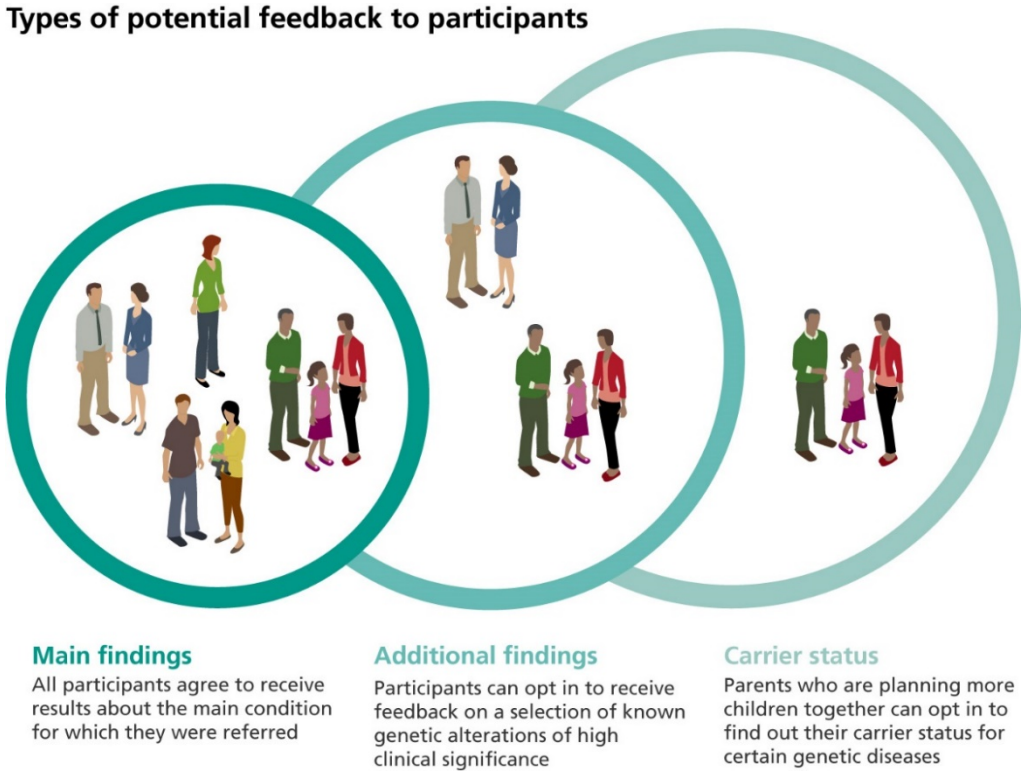


Image courtesy of Health Education England

More Precise Diagnosis

The right diagnosis first time

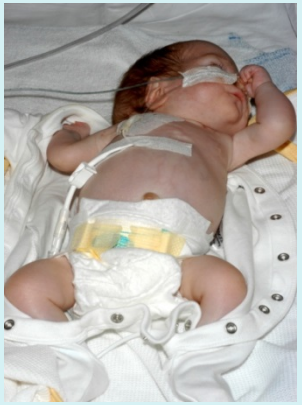
Five babies: all the same symptoms

***KCNJ11* p.V59M**
Permanent diabetes and developmental delay

Sulphonylurea therapy

EIF2AK3* p.E371
Wolcott Rallison Syndrome

Liver Transplant



***FOXP3* c.227delT**
IPEX syndrome

Bone Marrow Transplant

***GATA6* c.1448-1455del**
Syndromic pancreatic agenesis

Insulin and exocrine supplements

STAT3 p.T716M
Multi-organ autoimmune disease

? STAT3 inhibitor

The right treatment first time

Pharmacogenomics

- 8000 NHS beds a day occupied by people who have had adverse drug events (6.5% of total beds)
- 70% of the way that drugs are metabolised is through the cytochrome p450 pathway
- Many drugs affected by genetic variants e.g. codeine, abacavir, warfarin
- Right antibiotics first time

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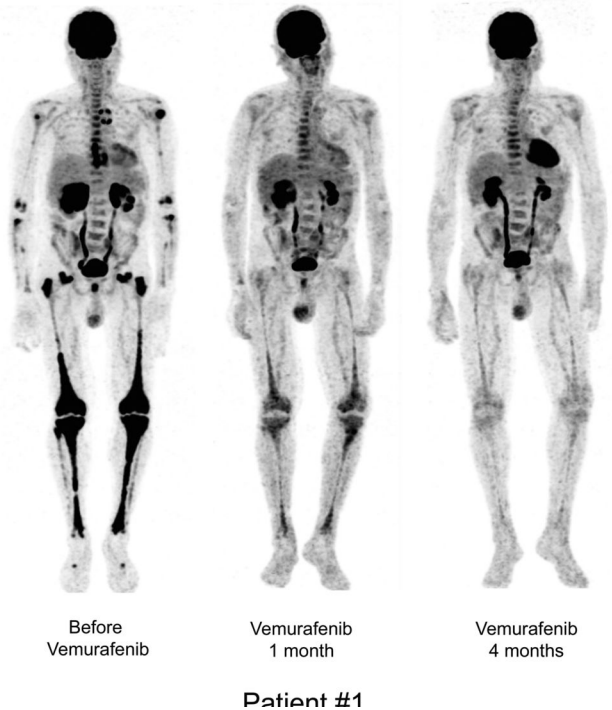
Personalised & targeted

Cancer care

- Highly targeted
- Panels or WGS? Jury is out.
- Expensive drugs only for those for whom it will work
- If resistance develops, re-sequence
- A baseline if cancer returns
- Provides dataset for future clinical trials



A Participatory role for patients

- Wellness rather than illness
- Approach with caution

Ethics

Where will this technology take us?

- Whole genome sequencing at birth
- What about before birth?
- What do we want to know?
- What do we want NOT to know?
- How sure do we have to be?
- Over-diagnosis?

What the future is not

- Genomics in genetics departments

The future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DIAGNOSTICS
Our innovative testing pipeline will provide ultra-high sensitivity and low cost diagnostics for use anytime, anywhere.
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