NHS
Central and South
Genomics

Genomics: From Niche to Normality

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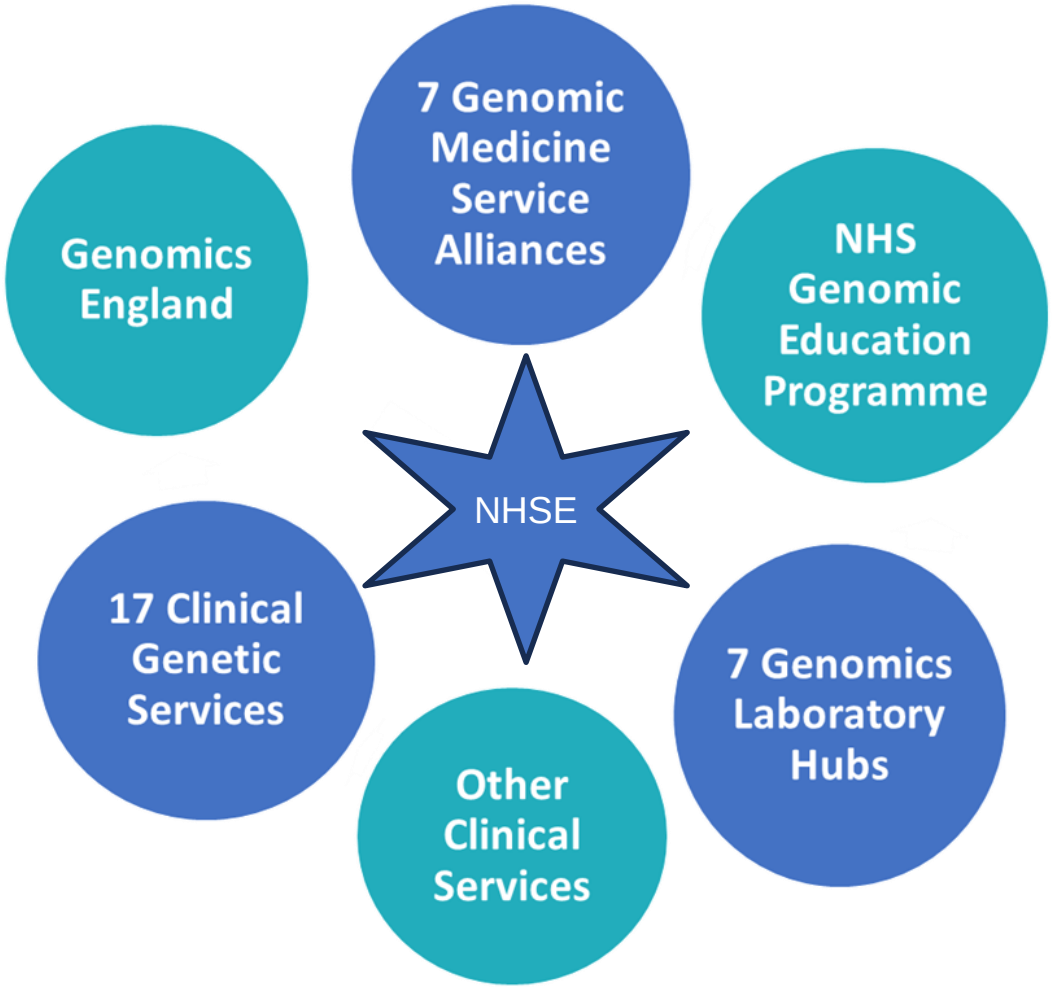
Ass. Director N&M

https://centralsouthgenomics.nhs.uk/
@CaS_Genomics

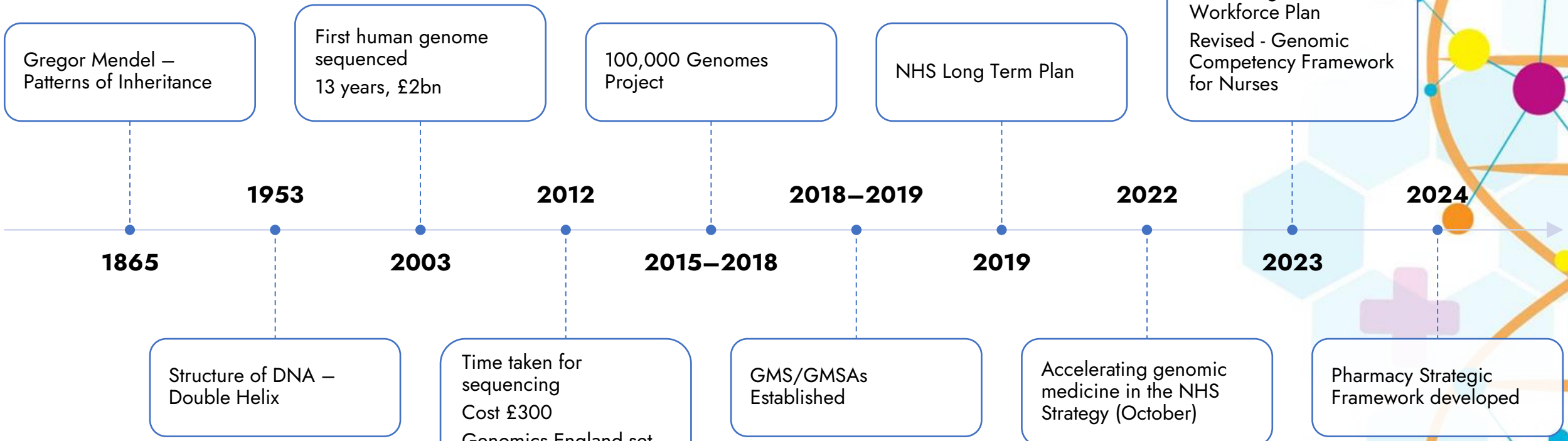
Outline

- GMS
- Basics
- In the News
- Why is genomics relevant
- Cancer
- Genomic Tests
- EDI
- Education and Resources
- Support

Components of the NHS Genomic Medicine Service



Timeline


1865
Gregor Mendel – Patterns of Inheritance
1953
Structure of DNA – Double Helix
2003
First human genome sequenced
13 years, £2bn
2012
Time taken for sequencing
Cost £300
Genomics England set up
2015–2018
100,000 Genomes Project
2018–2019
GMS/GMSAs Established
2019
NHS Long Term Plan
2022
Accelerating genomic medicine in the NHS Strategy (October)
2023
NHS Long Term Workforce Plan
Revised - Genomic Competency Framework for Nurses
2024
Pharmacy Strategic Framework developed

Genomics

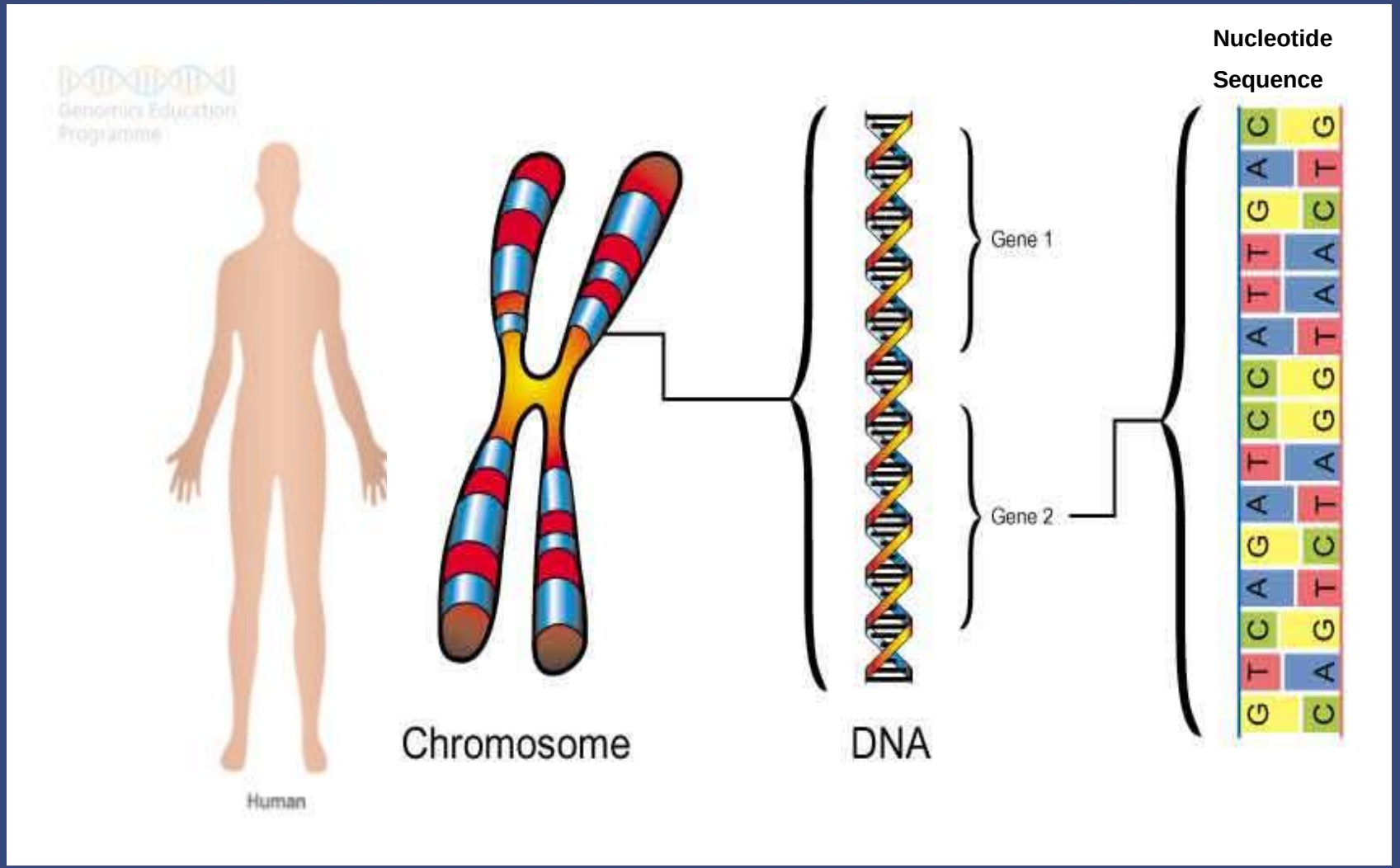
- The study of an organism's complete set of genetic information.
- The genome includes both genes (coding) and non-coding DNA.
- 'Genome': the complete genetic information of an organism.

vs

Genetics

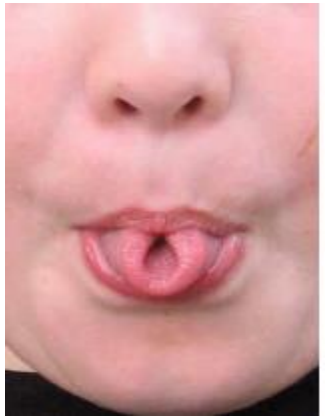
- The study of heredity
- The study of the function and composition of single genes.
- 'Gene': specific sequence of DNA that codes for a functional molecule.

Your GENOME contains all the instructions for you to grow throughout your lifetime.
These instructions are passed down from your mother and father.



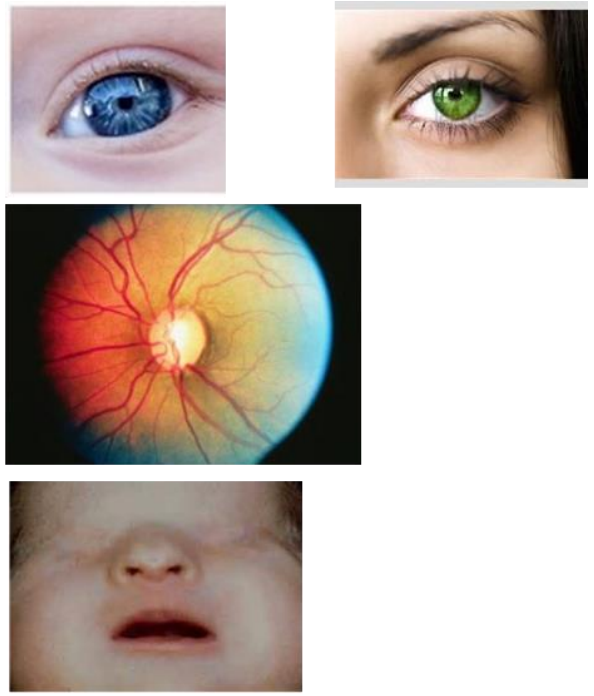
Not all variation in the genome is pathogenic

- Some differences in our DNA determine physical characteristics, such as eye colour. Others can influence our susceptibility to develop a disease.




What effect do variants have?

- No effect at all
- Effect on appearance - eye colour
- Slight effect on health – slight increased risk of glaucoma
- Severe effect on health - single gene disorder (‘pathogenic’ or ‘disease causing’)



Genomics Fun Facts

Genetic differences

Your genome is only around

0.1%

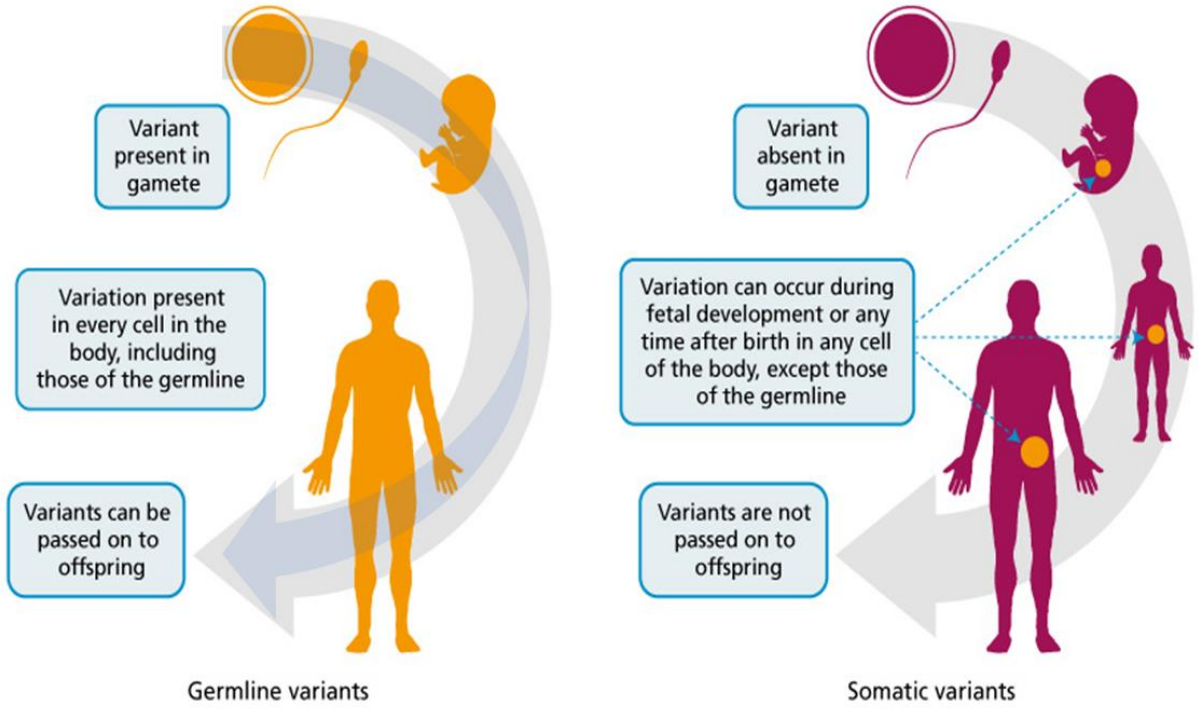
different from any other person's,

but that equates to

3 million

differences in your DNA

Cancer



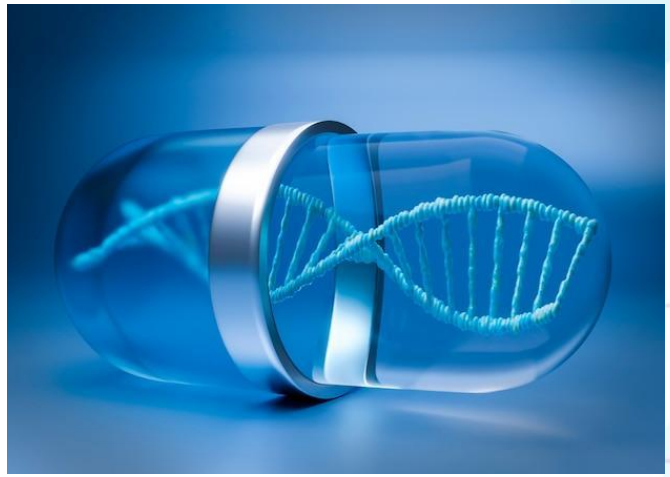
- Genomics and precision medicine is changing practice rapidly

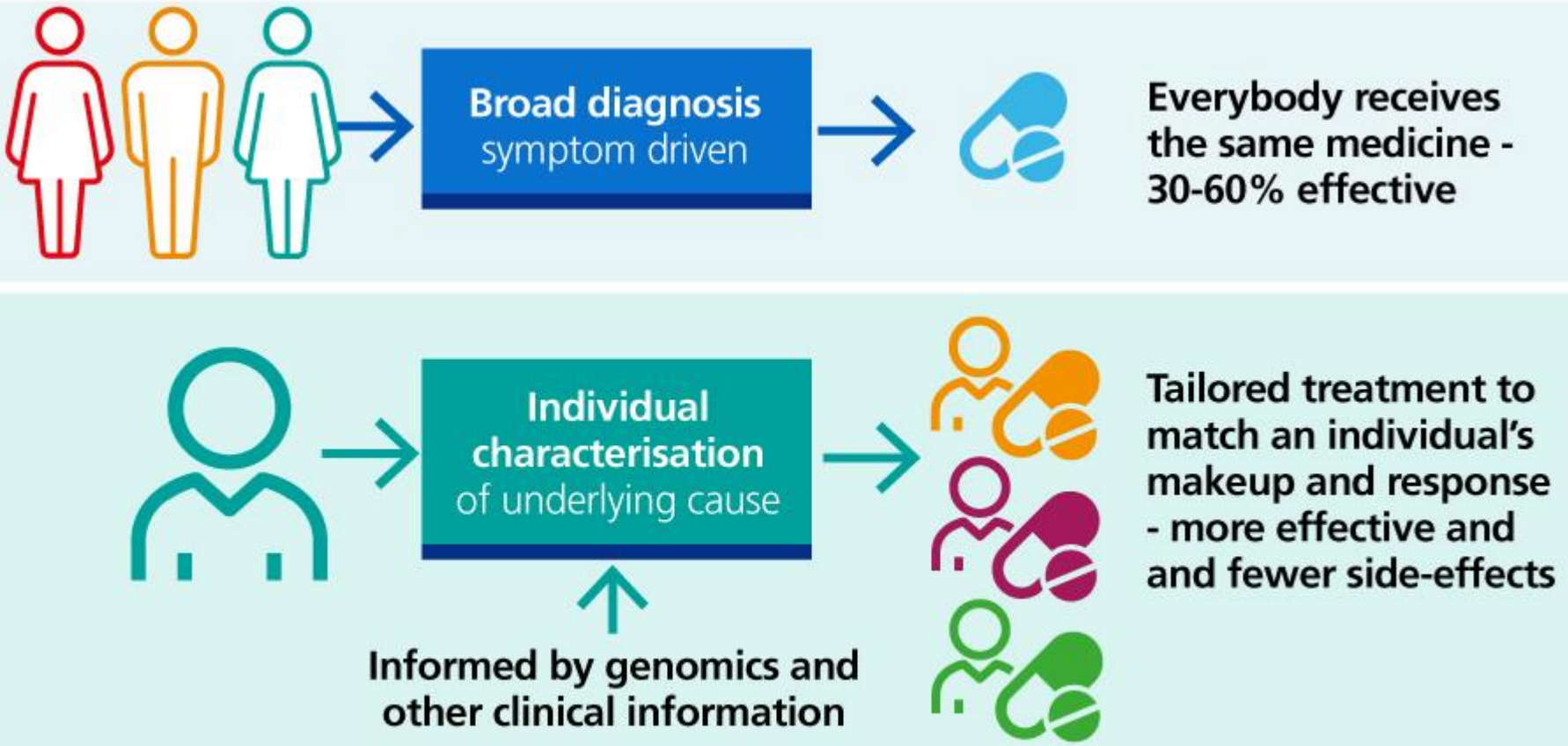
- Advances have led to an increased variety of testing to categorise cancer and create targeted treatments

Pharmacogenomics

The study of how genes affect a person's response to drugs.

It can be used to help develop effective, safe medications and doses that will be tailored to a person's genetic makeup.




Broad diagnosis
symptom driven
Everybody receives the same medicine - 30-60% effective
Individual characterisation of underlying cause
Informed by genomics and other clinical information
Tailored treatment to match an individual's makeup and response - more effective and and fewer side-effects

Royal College of Physicians | BRITISH PHARMACOLOGICAL SOCIETY

Personalised prescribing

Using pharmacogenomics to improve patient outcomes

A report from the Royal College of Physicians and British Pharmacological Society joint working party

NICE National Institute for Health and Care Excellence

Search NICE... Sign in

Guidance | Standards and indicators | Life sciences | British National Formulary (BNF) | British National Formulary for Children (BNFC) | Clinical Knowledge Summaries (CKS) | About

Home > News

NICE launches second consultation on genetic testing to guide treatment after a stroke

People could be offered a genetic test in the immediate period after having a stroke to help identify the most suitable treatment to reduce their risk of further strokes.

03 April 2024

NICE National Institute for Health and Care Excellence

Search NICE...

Guidance | Standards and indicators | Life sciences | British National Formulary (BNF) | British National Formulary for Children (BNFC) | Clinical Knowledge Summaries (CKS)

Home > News

NICE final draft guidance recommends olaparib for early breast cancer and metastatic prostate cancer following new commercial deals

NEWS

Home | Cost of Living | War in Ukraine | Coronavirus | Climate | UK | World | Business | Politics | Tech

Health

NHS to use test that prevents babies going deaf

9 hours ago

Awareness, Knowledge and Education

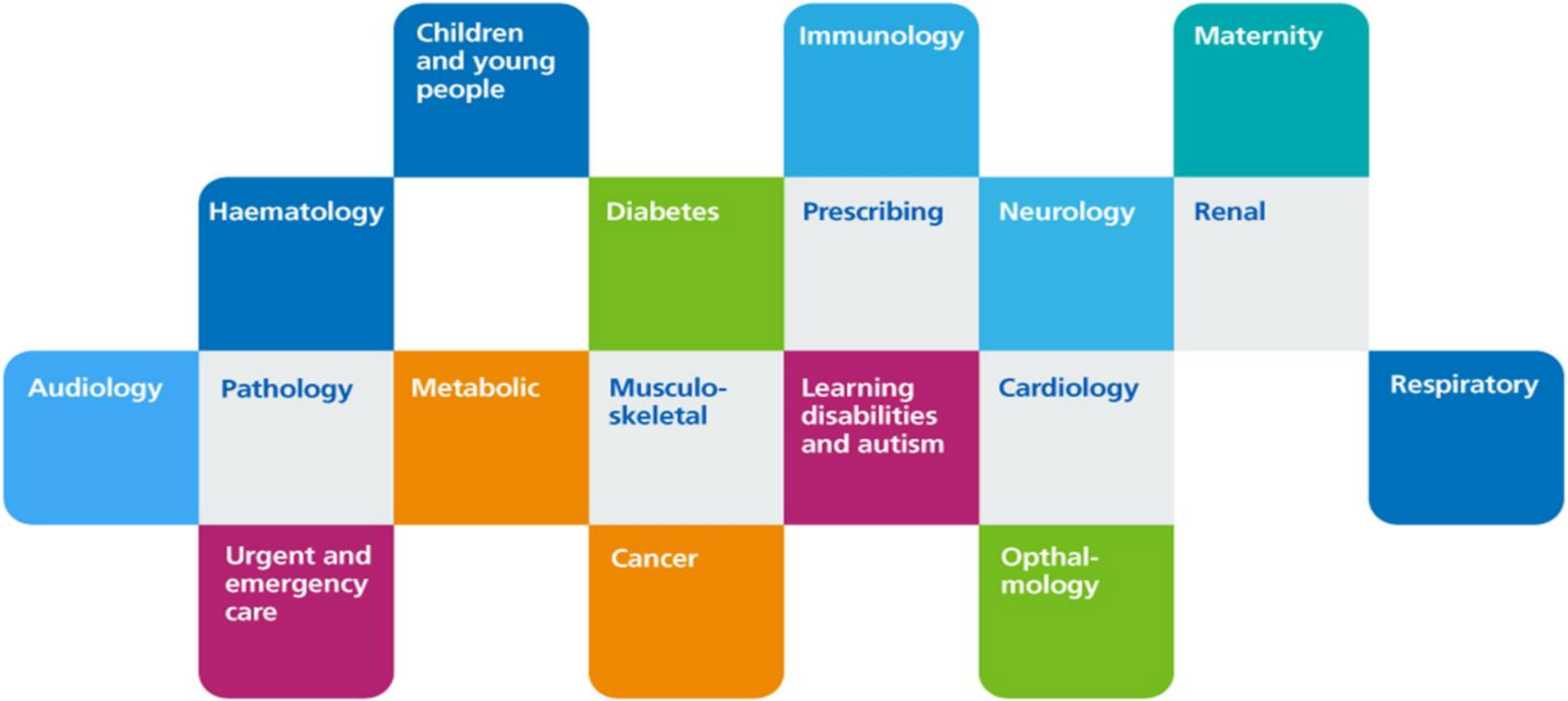
Why genomic education for nurses?

- Minimal genetics/genomics education
- Do not feel comfortable talking about genomics, fear of genomics

- ***Without genomics knowledge how do you know what is important?***

- Widened and more equitable access to genetic testing
- Better patient treatment outcomes
- Identification of more genetically “at-risk” individuals; screening, prevention and early detection

Clinical services impacted by genomics



PROSTATE CANCER UK

TRANSFORM TRIAL

Biggest prostate cancer screening trial in 20 years, could save the lives of thousands of men each year

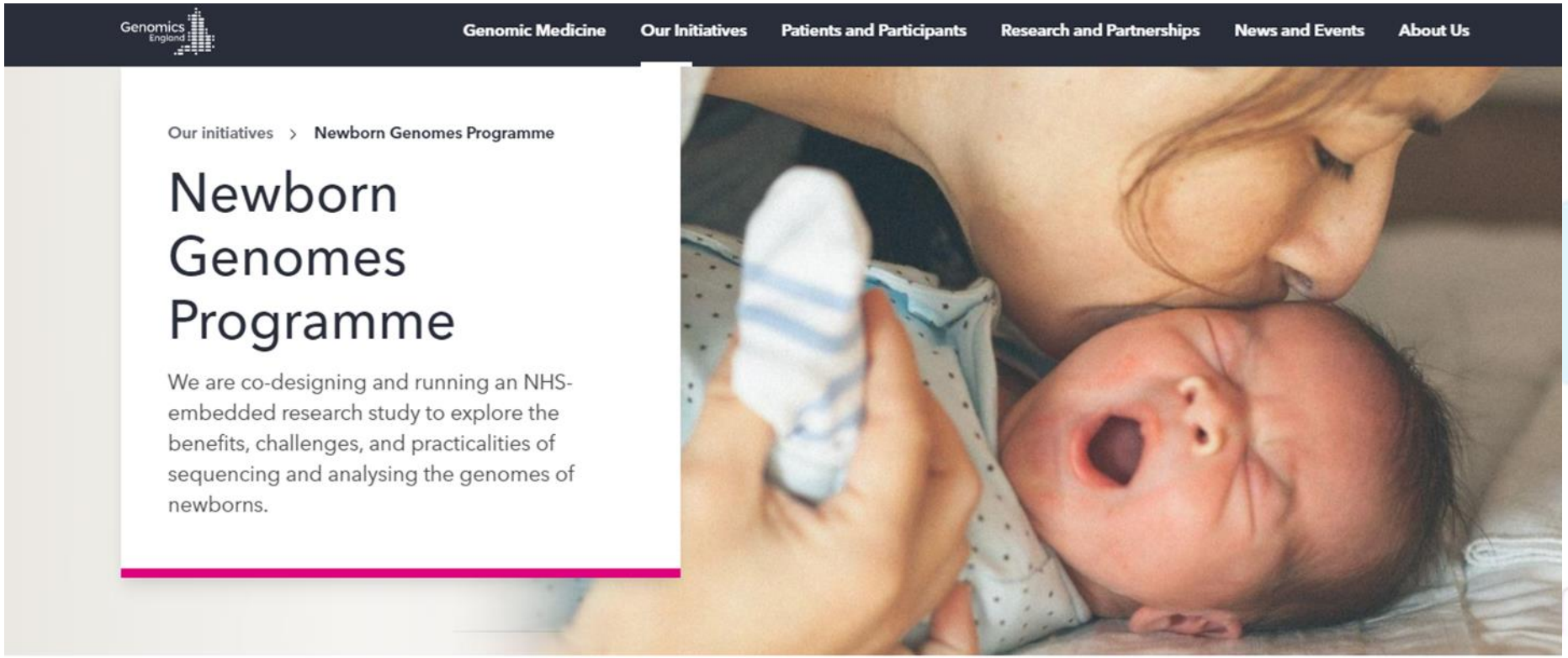
This research will transform how prostate cancer is diagnosed.

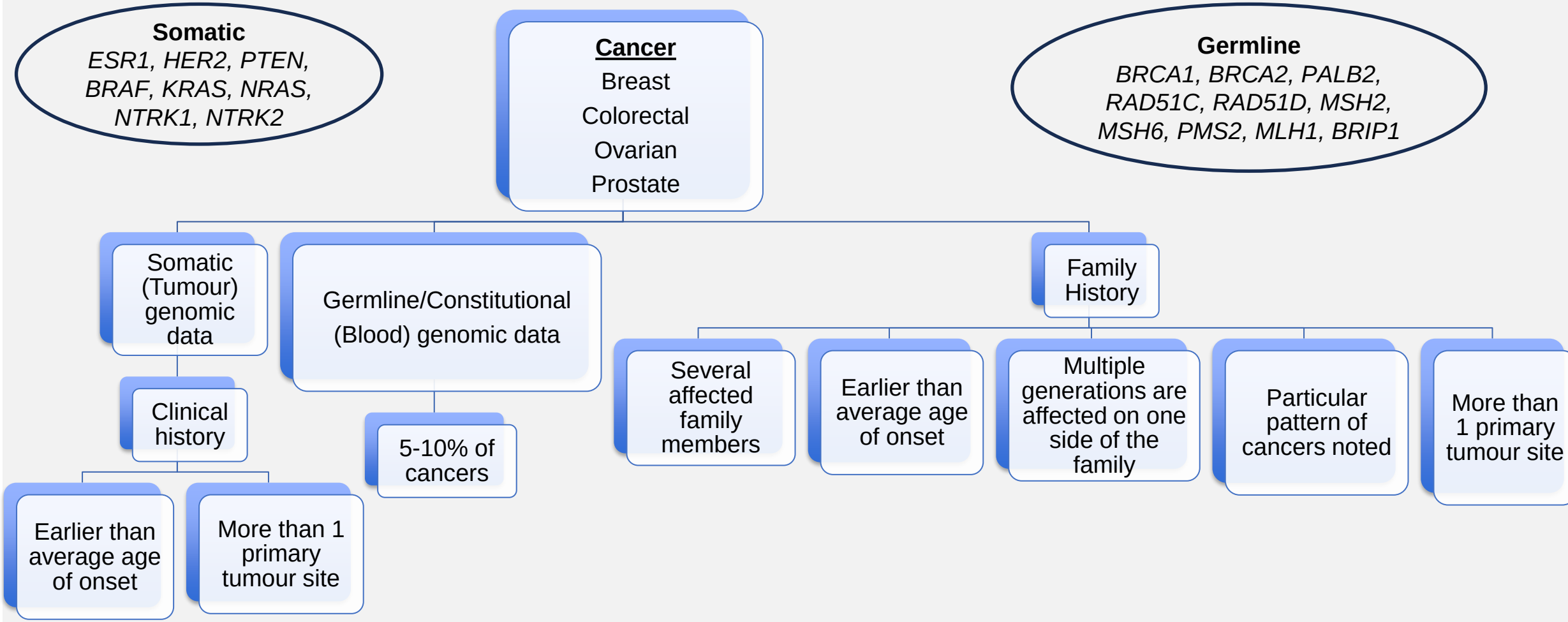
Men over 50 – or over 45 if you are Black or have a family history of prostate cancer

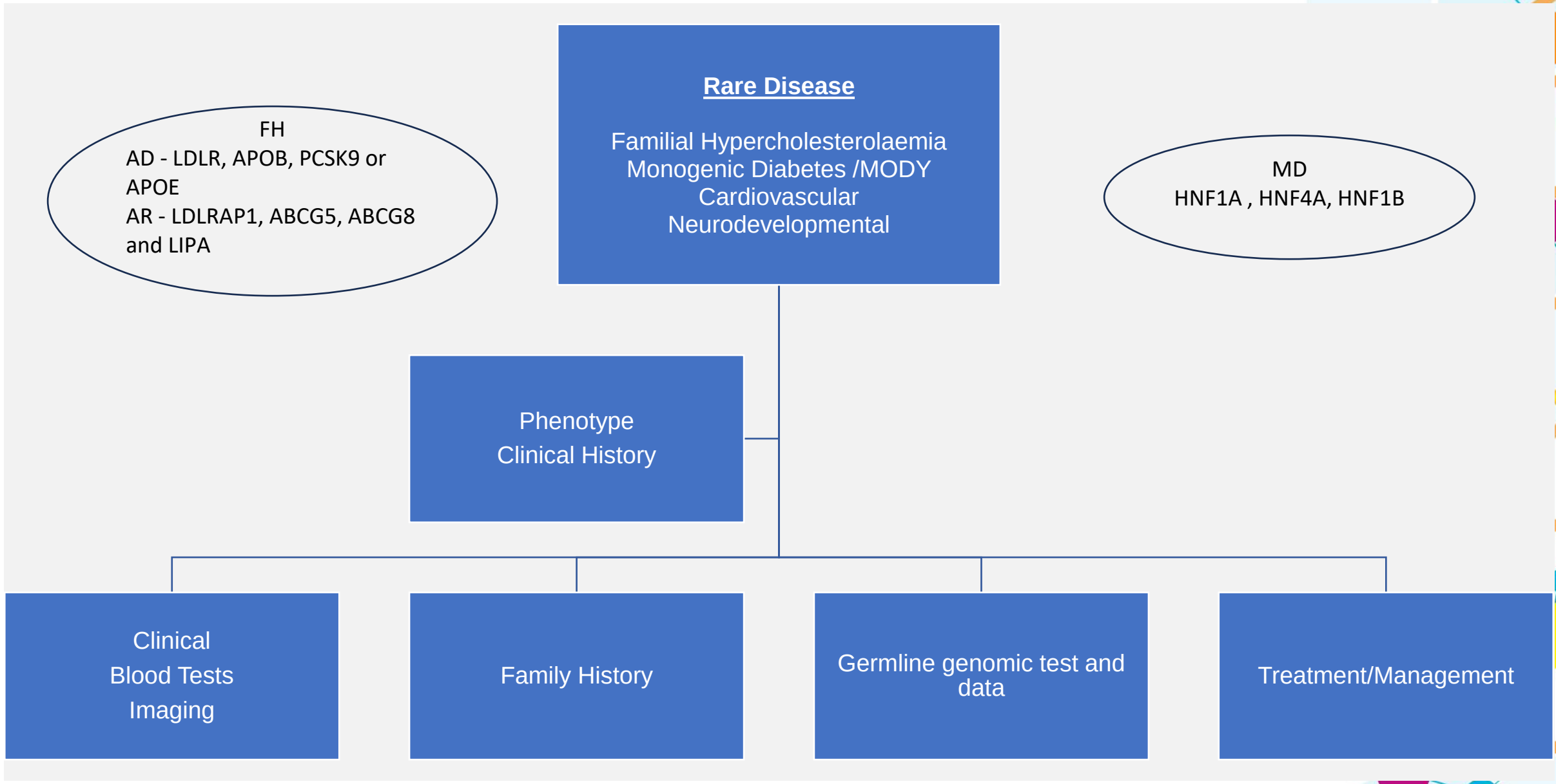
Lung Cancer – ctDNA

- Identify genetic variants in a tumour through a blood test
- Faster diagnosis
- Targeted treatment earlier

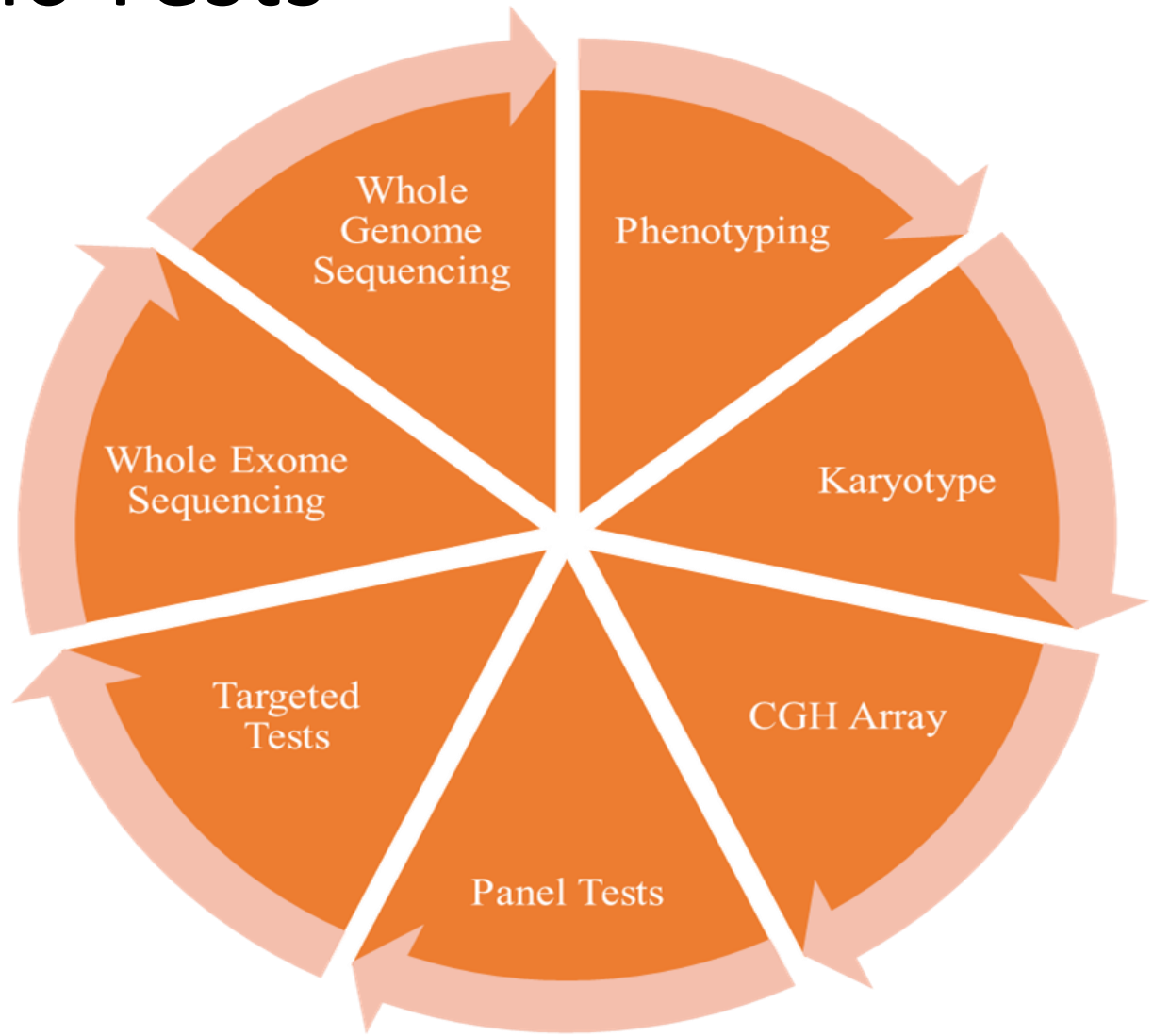
Kat Robinson, 33, Dorset

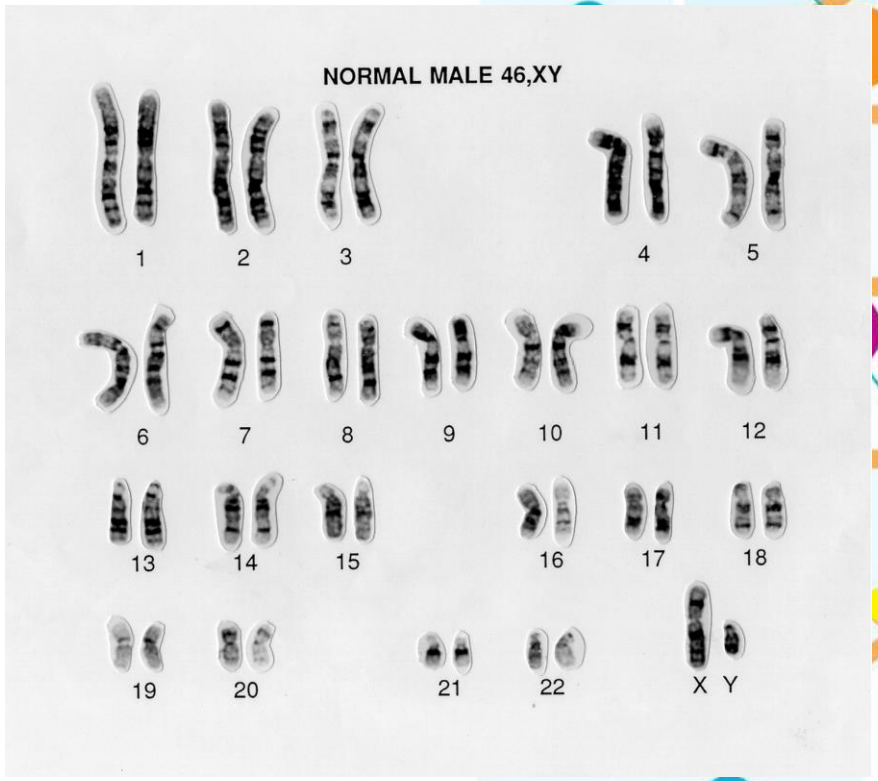

Genomics England
Genomic Medicine
Our Initiatives
Patients and Participants
Research and Partnerships
News and Events
About Us
Our initiatives > Newborn Genomes Programme
Newborn Genomes Programme
We are co-designing and running an NHS-embedded research study to explore the benefits, challenges, and practicalities of sequencing and analysing the genomes of newborns.


Somatic
ESR1, HER2, PTEN, BRAF, KRAS, NRAS, NTRK1, NTRK2
Cancer
Breast
Colorectal
Ovarian
Prostate
Germline
BRCA1, BRCA2, PALB2, RAD51C, RAD51D, MSH2, MSH6, PMS2, MLH1, BRIP1
Somatic (Tumour) genomic data
Germline/Constitutional (Blood) genomic data
Family History
Clinical history
5-10% of cancers
Several affected family members
Earlier than average age of onset
Multiple generations are affected on one side of the family
Particular pattern of cancers noted
More than 1 primary tumour site
Earlier than average age of onset
More than 1 primary tumour site


Rare Disease
Familial Hypercholesterolaemia
Monogenic Diabetes /MODY
Cardiovascular
Neurodevelopmental
FH
AD - LDLR, APOB, PCSK9 or APOE
AR - LDLRAP1, ABCG5, ABCG8 and LIPA
MD
HNF1A , HNF4A, HNF1B
Phenotype
Clinical History
Clinical
Blood Tests
Imaging
Family History
Germline genomic test and data
Treatment/Management

Genomic Tests





National Genomic Test Directory

- https://www.england.nhs.uk/publication/national-genomic-test-directories/

Part I. Acutely unwell children

R14 Acutely unwell children with a likely monogenic disorder 23

Part II. Cardiology

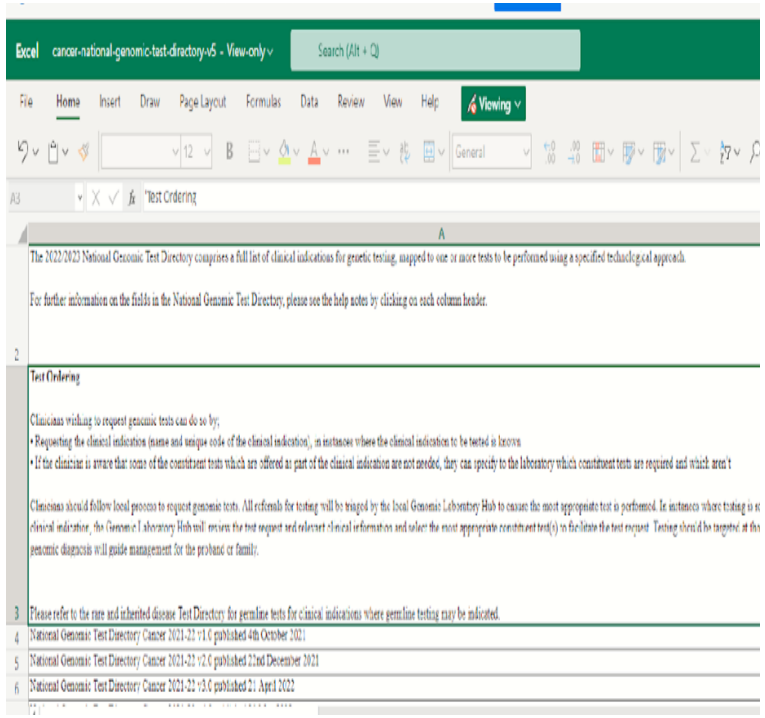
R137 Congenital heart disease - microarray 24
R125 Thoracic aortic aneurysm or dissection 25
R127 Long QT syndrome 26
R128 Brugada syndrome and cardiac sodium channel disease 27
R129 Catecholaminergic polymorphic VT 28
R130 Short QT syndrome 29
R131 Hypertrophic cardiomyopathy 30
R132 Dilated and arrhythmogenic cardiomyopathy 31
R391 Barth syndrome 33
R133 Arrhythmogenic right ventricular cardiomyopathy 34
R135 Paediatric or syndromic cardiomyopathy 35
R136 Primary lymphoedema 36
R138 Sudden unexplained death or survivors of a cardiac event 37
R328 Progressive cardiac conduction disease 38
R384 Generalised arterial calcification in infancy 39
R140 Elastin-related phenotypes 40

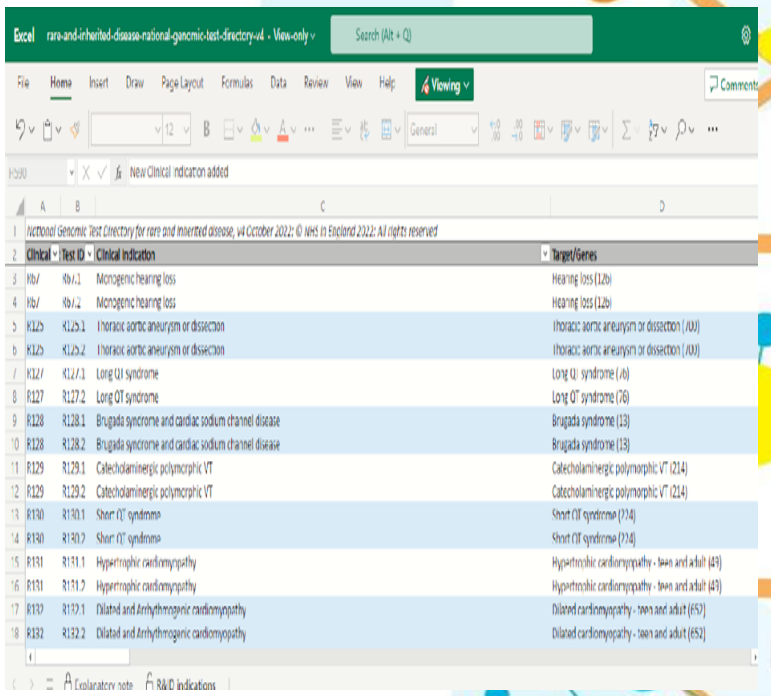
Part III. Developmental disorders

R26 Likely common aneuploidy 41
R27 Congenital malformation and dysmorphism syndromes - microarray and sequencing 42
R28 Congenital malformation and dysmorphism syndromes – microarray only 43
R29 Intellectual disability - microarray, and sequencing 44
R377 Intellectual disability - microarray only 45
R47 Angelman syndrome 46
R48 Prader-Willi syndrome 47
R53 Fragile X 48
R69 Hypotonic infant 49
R312 Parental sequencing for lethal autosomal recessive disorders 50

Part IV. Endocrinology

R402 Premature ovarian insufficiency 51

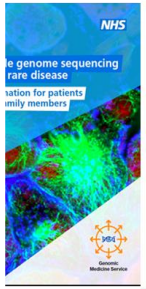




Order WGS Test

- **Test Order Form**
- Complete all sections
- Common sections not completed
  - Family Test
  - Age of Onset
  - Disease Penetrance
  - Parents Details
  - Phenotype for all – absent/present to be ticked

https://www.england.nhs.uk/genomics/genom-res/



Patient Information Leaflets

https://www.genomicseducation.hee.nhs.uk/wp-content/uploads/2019/11/Guide-to-requesting-WGS-RD-Nov-20.pdf

- Can be given to patient at any point
- Easy read version available
- 10 Different Translations available

https://www.genomicseducation.hee.nhs.uk/wp-content/uploads/2020/07/Top-tips-for-discussing-genomic-research.pdf

Record Of Discussion

- Confirmation that a discussion has occurred and all the relevant information has been given to patient
- A place to document the research offer choice of the patient
- Remote consent permitted – no patient signature required
- All sections to be completed
- Once signed and saved electronically – cannot be edited
- Must have a HCP signature

shc-tr.WRGLdutyscientist@nhs.net
bwc.centralsouthglh@nhs.net

Genomic Test Results

The results from a genomic test may show:

- A change in genes which are known to cause a health condition
- No change in genes which are known to cause a health condition
- Variant of Unknown significance - it's not clear what the results mean for health (but doctors may have a better understanding of the results in the future)

Validation of DTC results is not funded by the NHS unless the patient would otherwise meet criteria for germline genomic testing.

Why do you want the test?

Imagine receiving a result you are concerned about

Have you read all the information and small print about the test?

Could your decision to have a test affect your family?

Have you told your family that you are thinking about having a genetic test?

Are you happy with what the DTC company might do with your data?

- Trust
- Understanding (Translation/Literacy)

Research (NGRL)

Equality

Diversity

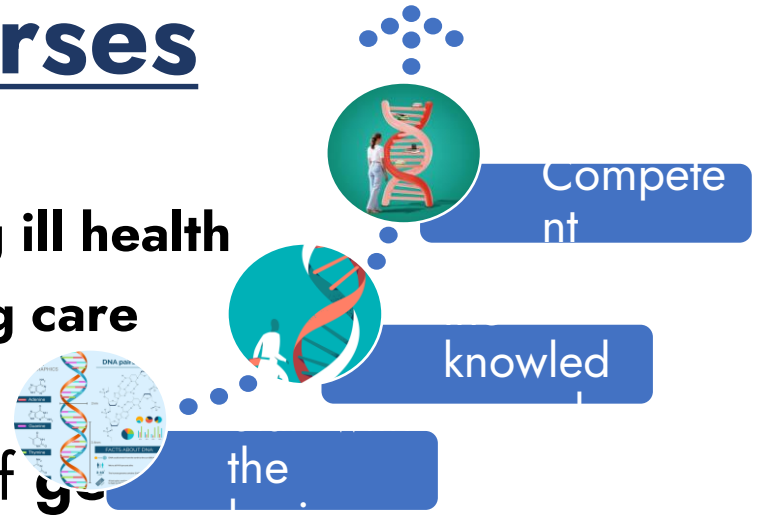
Inclusion

Equity of Access

NMC Standards of Proficiency for Registered Nurses

2.2 Promoting Health and preventing ill health

3.2 Assessing needs and planning care



Demonstrate and apply knowledge of **ge**

Competency Frameworks

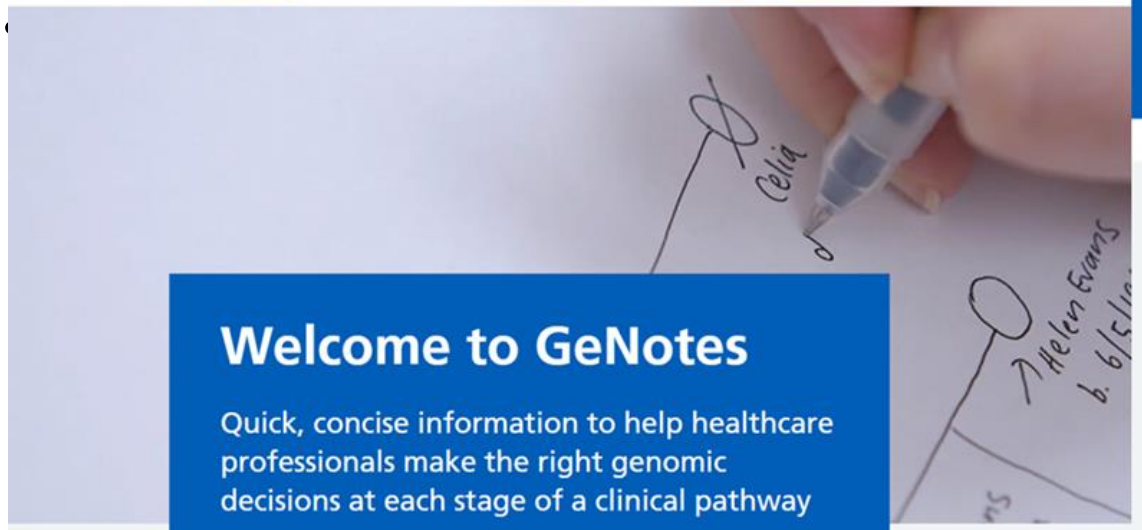
•www.genomicseducation.hee.nhs.uk/consent-a-competency-framework/

Education Resources

Create networks – share experience and knowledge

Home In the Clinic Knowledge Hub

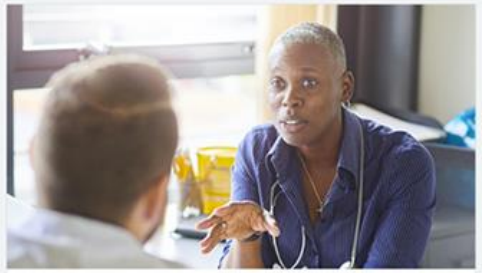
Public beta This website is in public beta – please give your feedback.



Welcome to GeNotes

Quick, concise information to help healthcare professionals make the right genomic decisions at each stage of a clinical pathway

Navigate



In the Clinic

Focused on the point of patient care, these short scenarios look at when to consider genomic testing and what you need to do.

Knowledge Hub

Extend your learning with this encyclopaedia of resources, designed to support your understanding of genomics in medicine

Genomics Education Programme

GeNotes
Genomic notes for clinicians

NHS England

Home In the Clinic Knowledge Hub About GeNotes

Public beta This website is in public beta – please give your feedback.

Home › In the Clinic

In the Clinic

Focused on the point of patient care, these short scenarios look at when to consider genomic testing and what you need to do.

Endocrinology

Key information for clinicians accessing genomic testing for their patients

Fetal and Women's Health

Clinical presentations from pre-pregnancy to antenatal care

Neurology

Clear, concise guides to genomic testing in patients presenting with neurological symptoms

Oncology

Short, useful summaries to support your clinical decisions when considering genomic testing

Paediatrics

Built around patient scenarios, these articles outline key information about testing and results

Pharmacogenomics

A handy guide to when and why genomic testing is of relevance when prescribing and dispensing

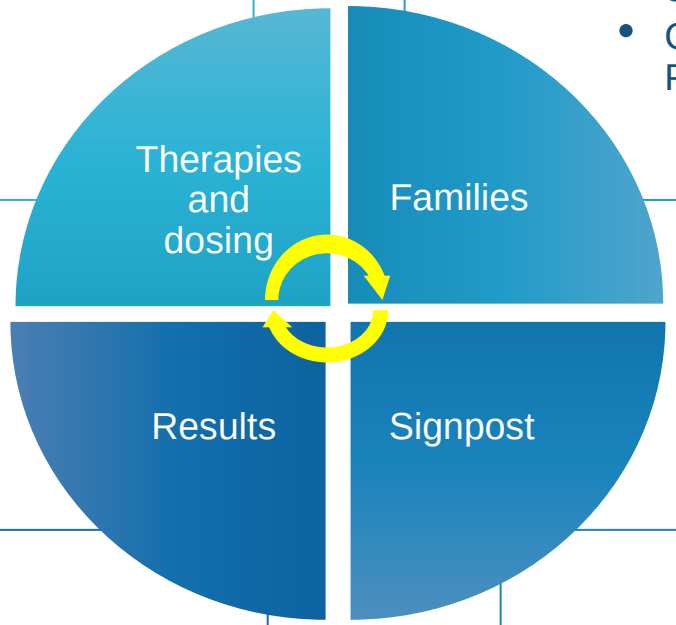
Primary Care

From first presentation to receiving results, these step-by-step summaries help support your practice

Support

- Pharmacists

- Clinical Genetic Services
- Genomic Practitioners



- Genomic Laboratory Hubs (GLH)

- Genomic Medicine Service

Clinical requests for whole genome sequencing should be sent to bwc.centralsouthglh@nhs.net

West Midlands Clinical Genetics Service
For general enquiries: genetics.info@nhs.net
Telephone: 0121 335 8024

Oxford Clinical Genetics Service
For general enquiries: orh-tr.churchill-clinicalgenetics@nhs.net
Telephone: 01865 225931

Wessex Clinical Genetics Service
For general enquiries: UHS.GeneticsTeam@nhs.net
Telephone: 023 8120 6170

For Central and South Genomic Medicine Service enquiries, GMSAAdmin@uhb.nhs.uk.

Genomics Learning Passport

Genomics Learning Passport for Editing

Your personal record of your genomics learning.

Genomics plays a fundamental role in nursing practice. The ever-evolving field of genomics is transforming patient care across various aspects, including diagnosing illnesses, predicting disease risks, and tailoring treatments to enhance efficacy while minimizing adverse effects. It is imperative that all nurses possess a solid understanding of genomics, enabling them to confidently integrate genomics into patient care, as outlined in the NMC Standards of proficiency for registered nurses[1]. The NHS recognises the significance of genomics education for healthcare professionals and there are many strategies [2,3,4] addressing this importance.

However, embarking on this genomics journey can sometimes be overwhelming. To assist you on this path, we have developed a genomics learning passport as a guide and evidence of your learning. It is not a competency document however it can help you to acquire the necessary knowledge to meet the competencies outlined in the nurses competency framework (the updated framework is due for publication soon). This tool is designed to navigate you through the wealth of available resources while also documenting your genomics learning journey and can also support your revalidation process.

Whether you are just starting to explore the core concepts or are ready to delve into master's level studies, there are education and training resources available to suit your needs. Towards the end of this log, we've provided a list of websites and additional sources of information to facilitate your self-directed learning, including the links to the national strategies

Should you have any inquiries or require further guidance, please feel free to contact : Philandra Costello, Lead Genomic Nurse, Central & South Genomic Medicine Service Alliance, Philandra.costello@uhs.nhs.uk

Guide to completing log:

The following table lists the available online genomic learning resources relevant to nursing. Not all modules will be relevant to your current sphere of practice at this time, however all nurses should be aware of the basic concepts covered in the Genomics 101 modules.

The log is based on the NMC CPD activity log so that you can use this record of learning for your revalidation. See also www.revalidation.nmc.org.uk/download-resources/guidance-and-information .

Name		NMC number	
Job Title		Date revalidation next due	
Place of work			

Date completed	Learning Resource [Including method of study] • Online learning • Course attendance • Independent learning	Overview of resource content / subject area	Topic(s): Provide an overview of the learning activity, its relevance to your scope of practice, the knowledge gained and how you have integrated this into your practice. Alternatively reflect on the connection between the learning activity and your practice and your ability to apply what you have learned	Link to Code: Please identify the part or parts of the Code relevant to your CPD. • Prioritise people • Practise effectively • Preserve safety • Promote professionalism and trust	Link to Standards of proficiency: Please identify the part or parts of the relevant standards that you used to inform your CPD. Align to Genomic Nursing Competency Framework To ensure that the knowledge you gain is targeted, relevant and effective in your practice	Number of hours:	Number of participatory hours:

The Genomic Education website has a wide range of resources from short films on core concepts to information about studying at master's level. There are too many courses to list below so we have just included some of the key resources that are most relevant to nursing practice to get you started on your genomics learning journey. You can also add any additional learning at the end of the log.

Where to begin? A good place to start is the resource developed specifically for nurses at Genomics in Nursing (hee.nhs.uk). We suggest that you start by reviewing this content and record that you have completed this activity for your revalidation below.

Date Completed	Learning Resource and method of study	Overview of resource content / subject area	Topic(s):	Link to Code:	Link to Standards of proficiency Genomics Competency Framework	Number of hours:	Number of participatory hours:
	Genomics in Nursing – Genomics Education Programme (hee.nhs.uk) Online /independent learning						
Not sure of the difference between genomics and genetics? The following module revisits the basics.							
	What is genomics? Online learning						
Need more help with terms or words - the following glossaries might help							
Glossary Talking Glossary							

Genomics Education Programme: Genomics in the NHS

The Genomics in the NHS - eLearning for healthcare (e-lfh.org.uk) programme is a collection of online courses developed by NHS educational teams to support and educate healthcare professionals about genomic medicine and the benefit it will bring to patient care.

The following modules are available at E-Learning for Health and are free to NHS staff once registered onsite at Home - eLearning for healthcare (e-lfh.org.uk)

Genomics 101: A series of 9 short 30–40-minute online modules explaining the core concepts of genomics and how to apply these in your clinical practice. Certificate of completion available for each module. All nurses should be aware of the content covered in these modules. Access via links below or via E-LFH.

	Genomics in healthcare						
	From Genes to Genome (Nursing competency 1,4,6)						
	From gene to protein (Nursing competency 1,4,6)						
	Inheriting genomic information (Nursing competency 1)						

https://forms.gle/EdS8GsV2TWn33wYeA

Thank you!

Phil & Charlotte

Charlotte.Hitchcock@uhb.nhs.uk