NHS
Central and South Genomics

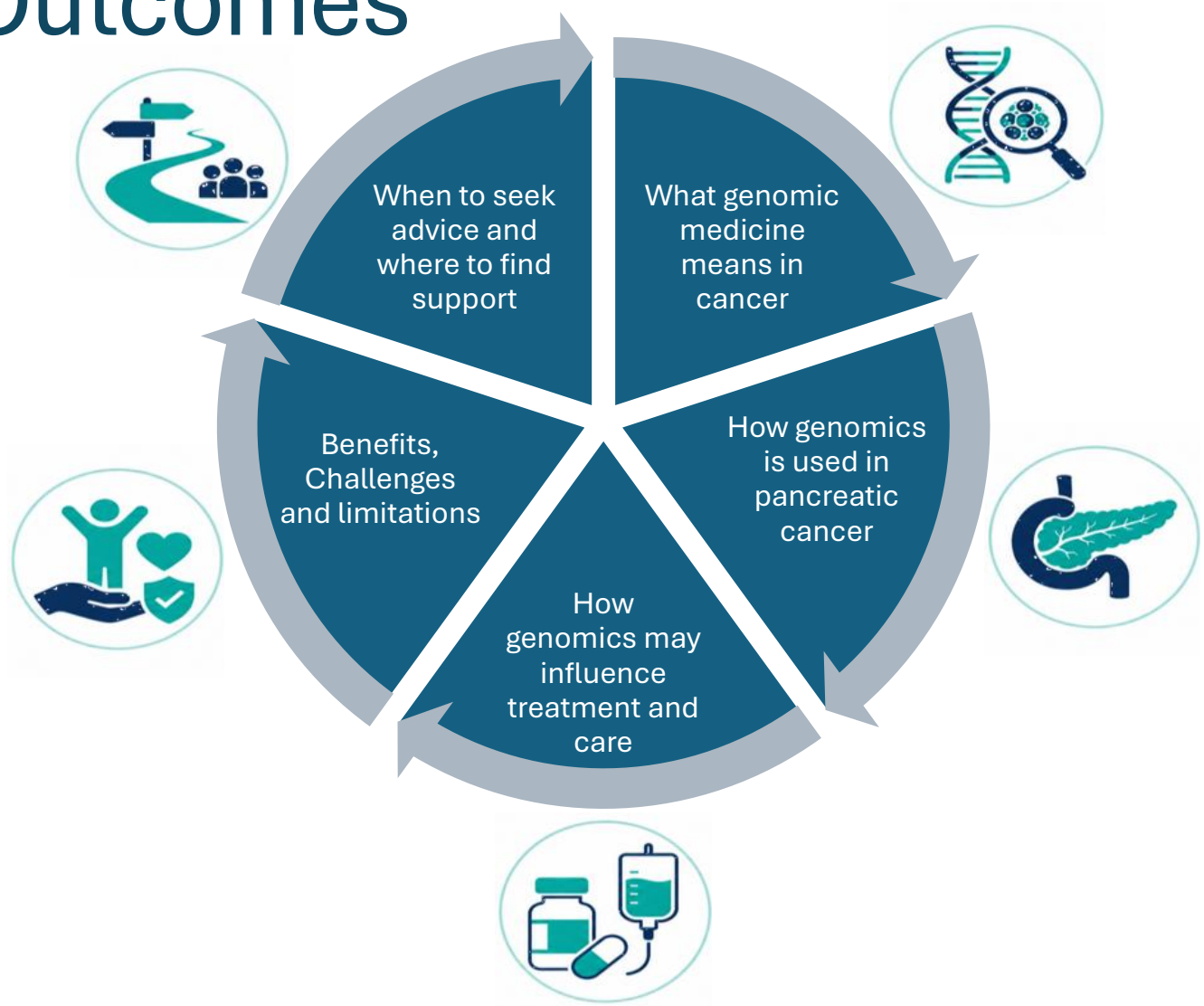
An introduction to Genomic Medicine in Pancreatic Cancer

Philandra Costello
Lead Genomics Nurse
Central & South Genomic Medicine Service

https://centralsouthgenomics.nhs.uk/

- When you hear the word **GENOMICS**, what comes to mind?

Learning Outcomes



Genomic Medicine: From Specialist Testing to Everyday care

The changing role of genomics

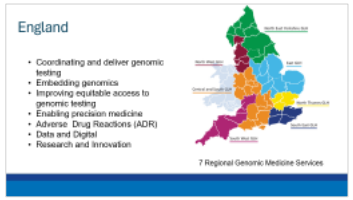
Past
- Mainly specialist services
- Rare inherited conditions
- Limited genetic testing

Today
- NHS Genomic Medicine Service
- Testing embedded into cancer pathways
- Tumour profiling and inherited risk

Future
- Earlier detection
- Prevention and prediction
- More personalised care

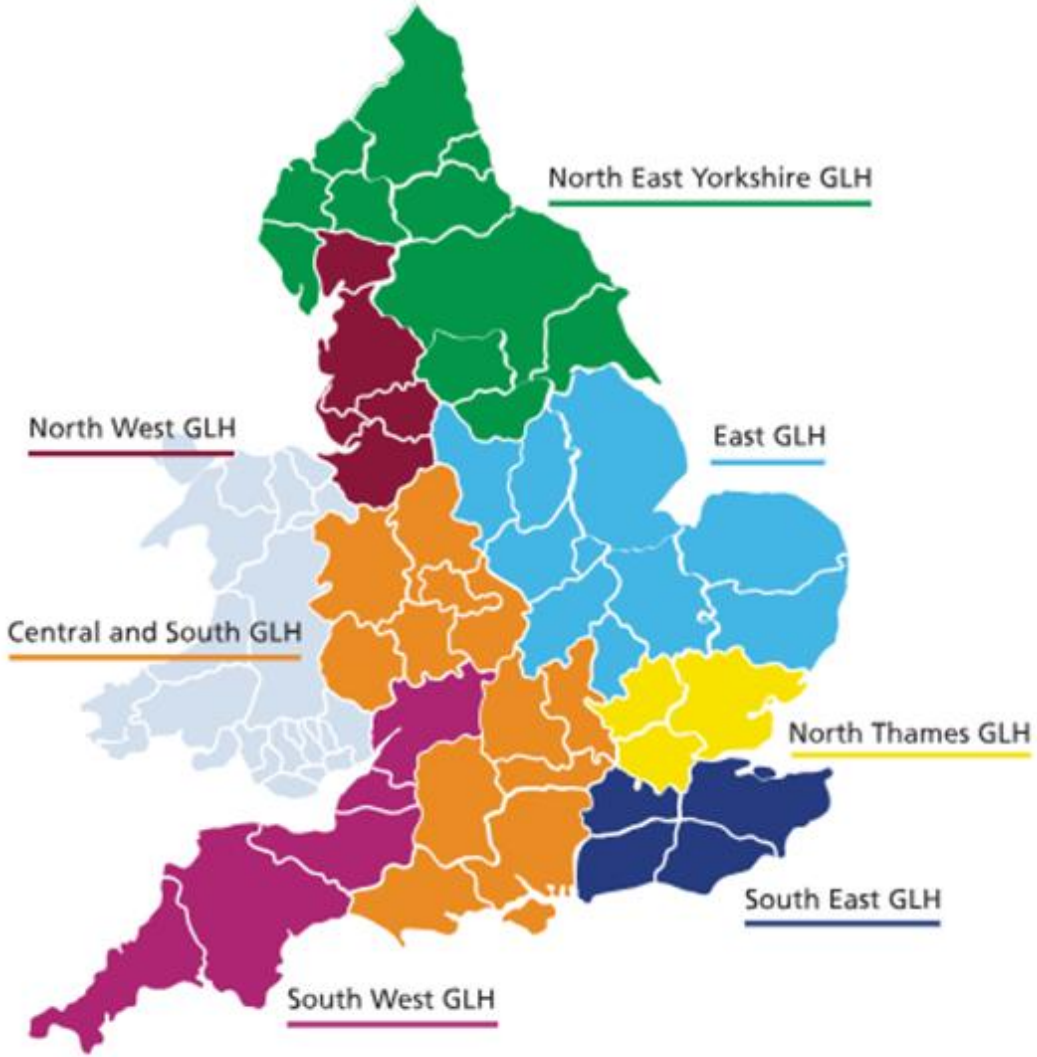
Harnessing genomics, data and innovation to transform care

[10 Year Health Plan for England: fit for the future - GOV.UK]



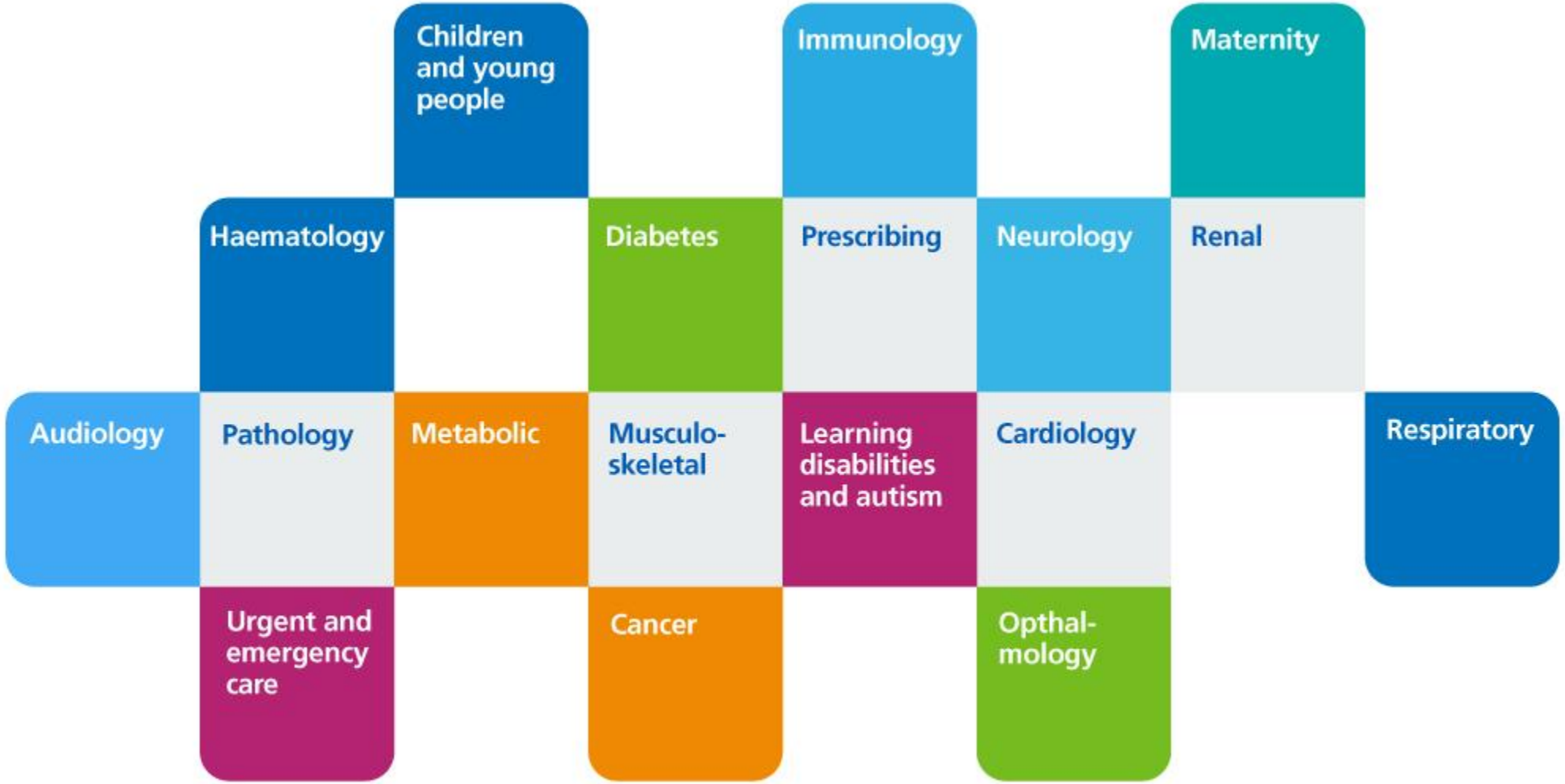
England

- Coordinating and deliver genomic testing
- Embedding genomics
- Improving equitable access to genomic testing
- Enabling precision medicine
- Adverse Drug Reactions (ADR)
- Data and Digital
- Research and Innovation



7 Regional Genomic Medicine Services

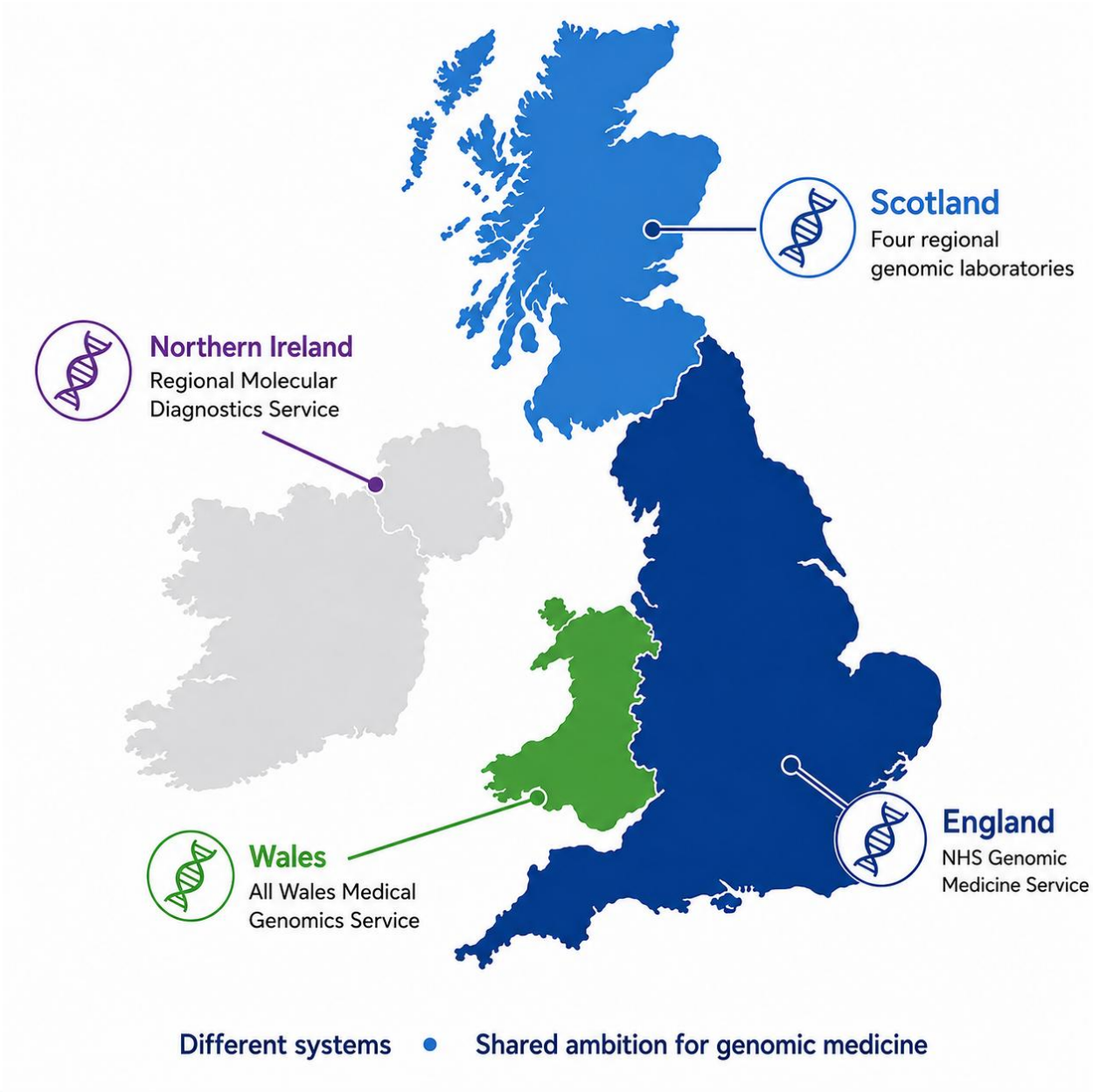
Where it applies



Genomics Test Directory

- The primary source for all commissioned genomic tests in NHS England is the **National Genomic Test Directory**. This exists in two parts:
  - **Cancer Test Directory**
  - **Rare & Inherited Disease Directory** – for (germline) tests

UK

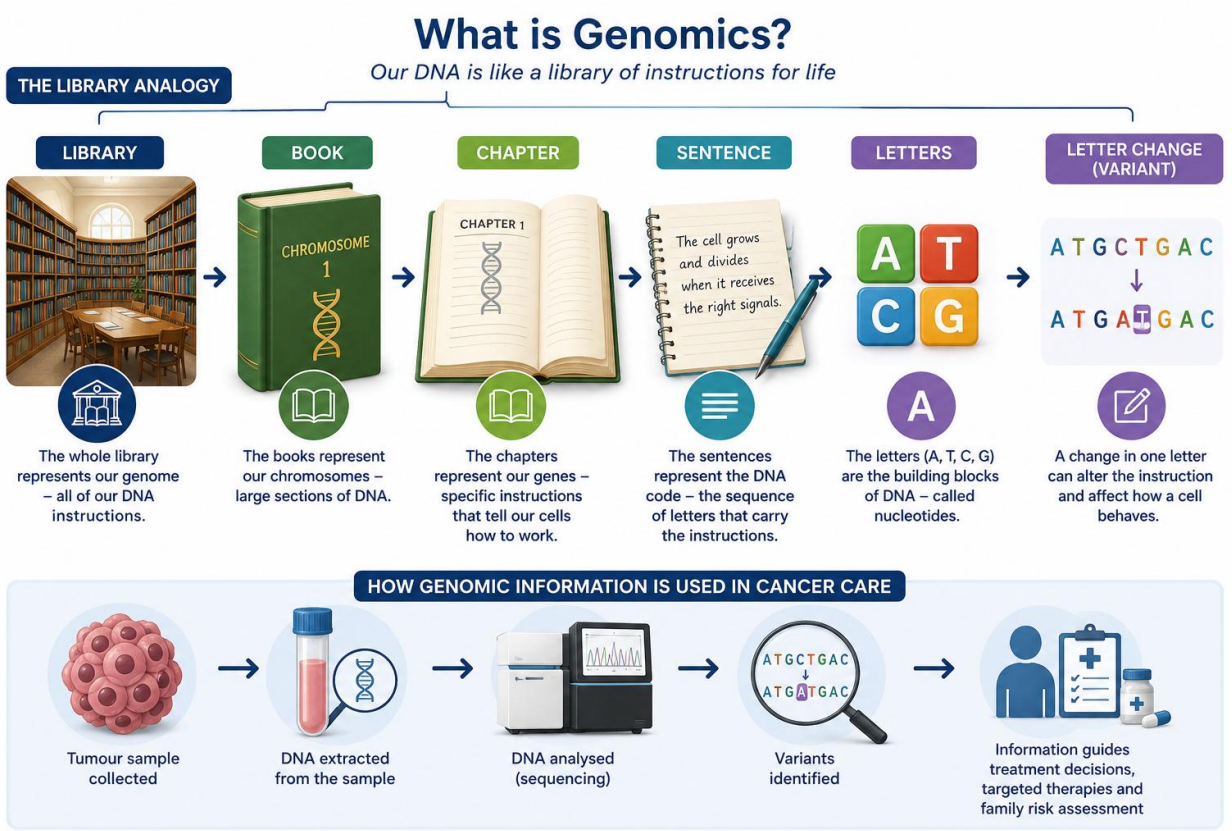

Scotland
Four regional genomic laboratories
Northern Ireland
Regional Molecular Diagnostics Service
Wales
All Wales Medical Genomics Service
England
NHS Genomic Medicine Service
Different systems
Shared ambition for genomic medicine

What is Genomics?

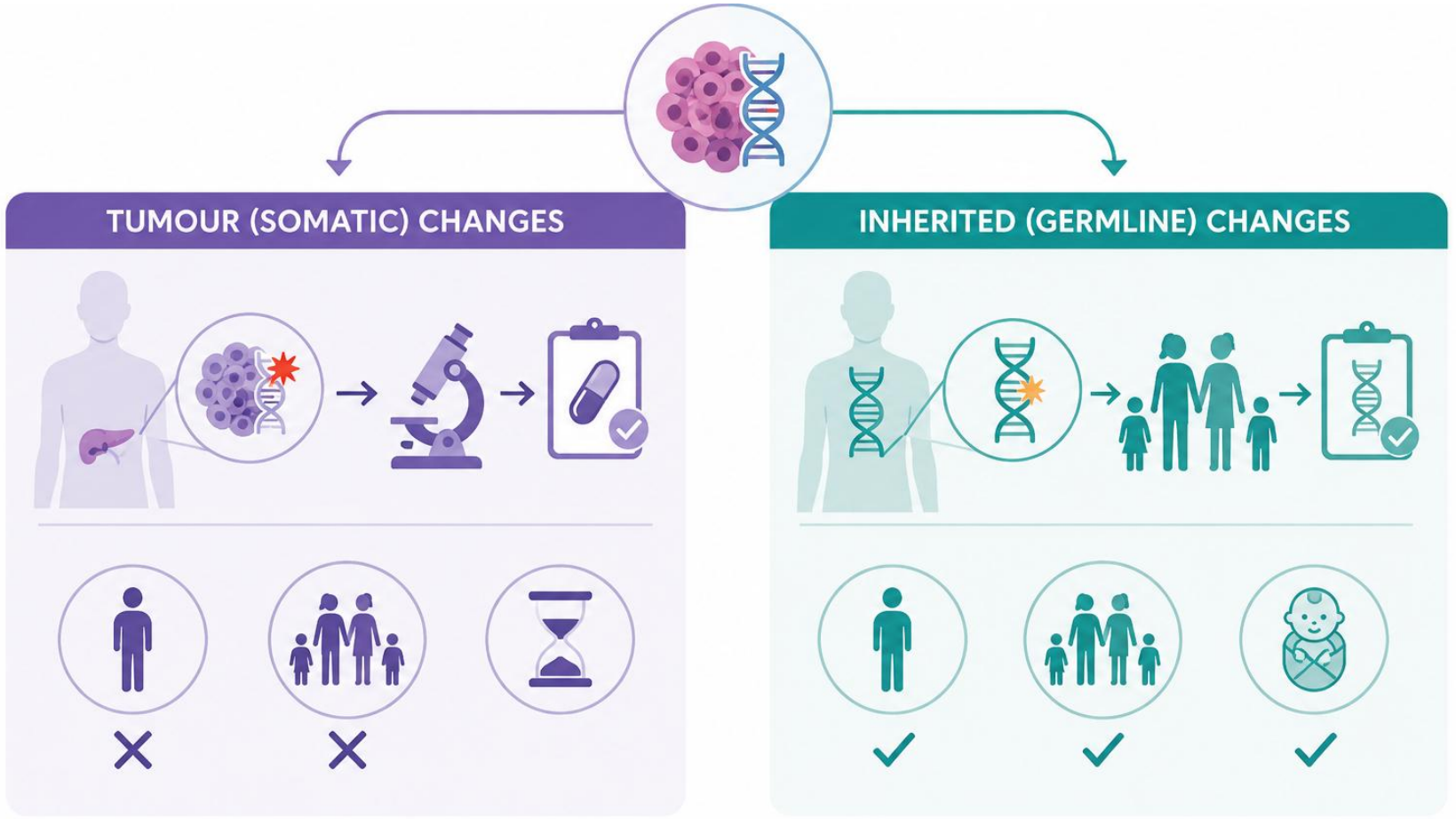
Genomics vs Genetics

- 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.

- 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.



All cancer is genomic, but not all cancer is inherited



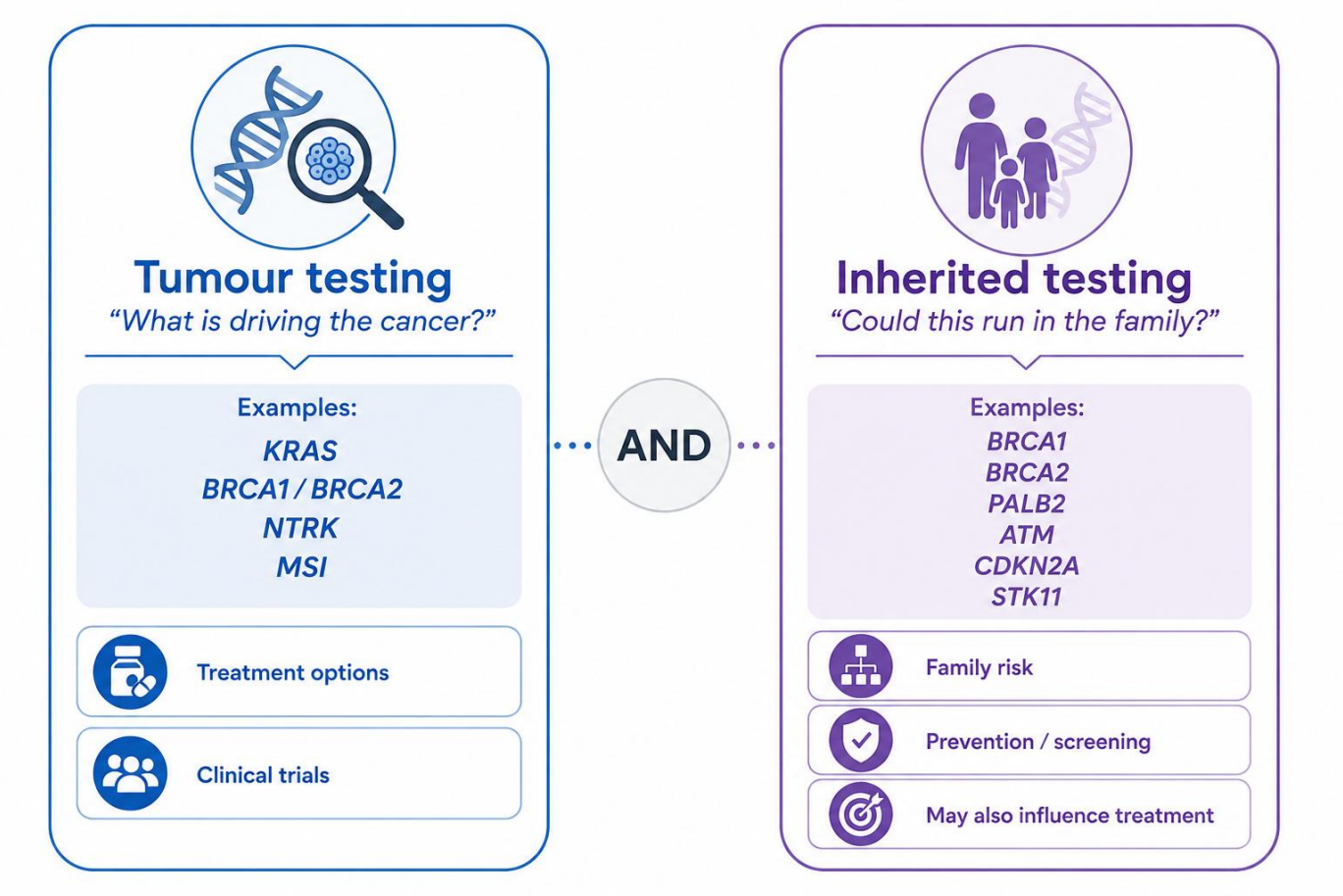
Tumour/Somatic

- ✓ Occur during life
- ✓ Found only in cancer cells
- ✓ Guide Treatment decisions

Inherited/Germline

- ✓ Present from birth
- ✓ Can run in families
- ✓ Can influence treatment, prevention and screening

Genomic testing in pancreatic cancer: different questions, different tests



Meet David: Where does genomics Fit?

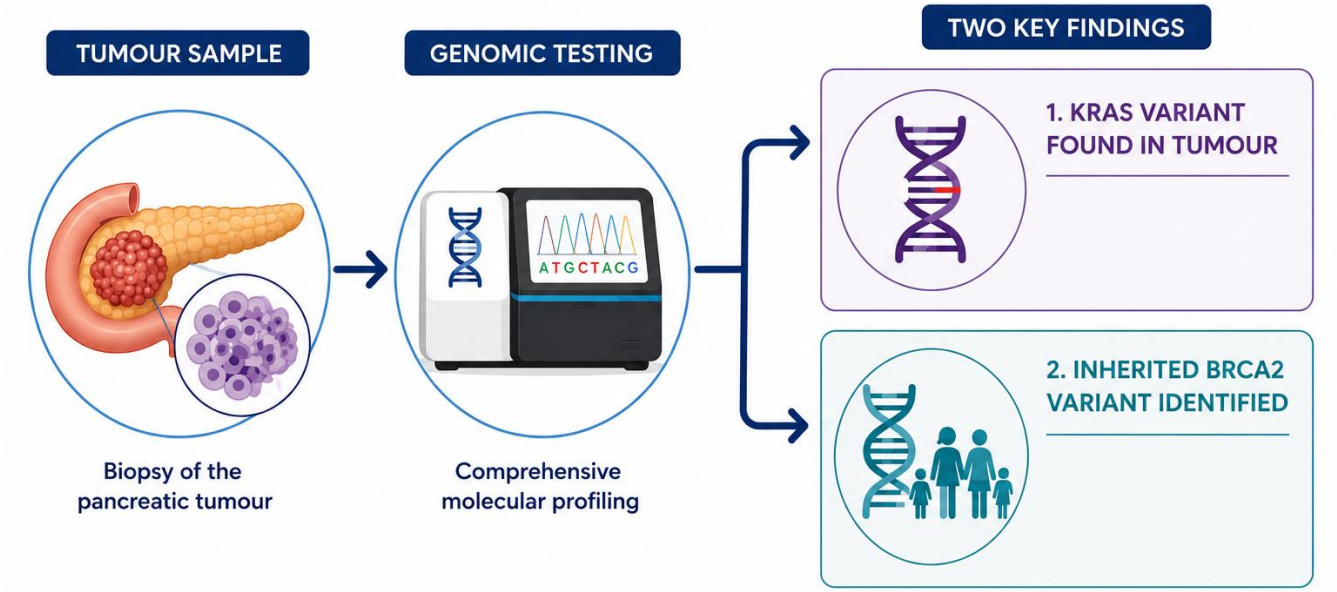
 David, 58

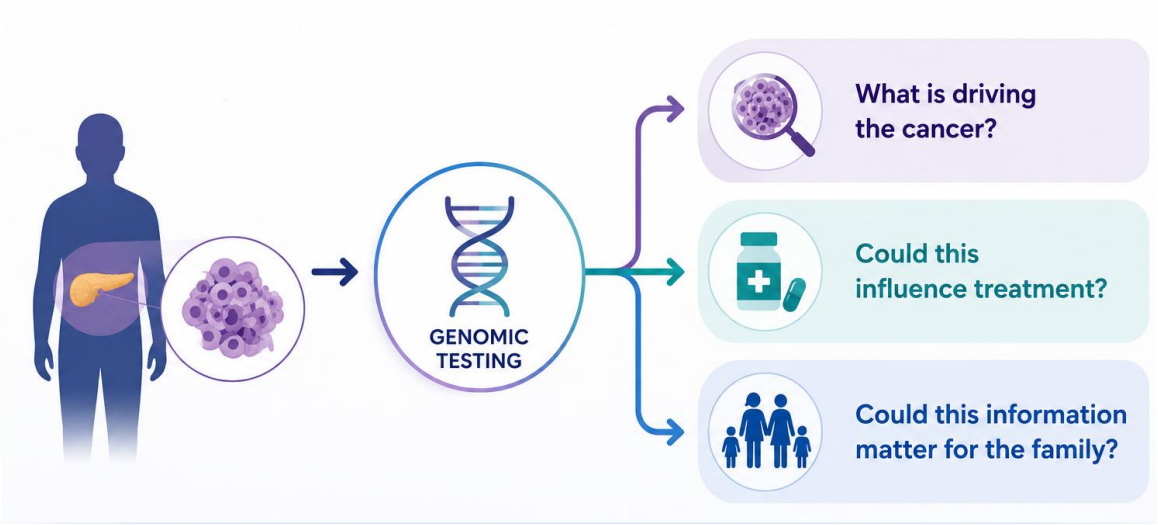
Diagnosed with advanced pancreatic cancer

Questions:

- What is driving my cancer?
- Could genomics affect my treatment?
- What does this mean for my family?

David’s genomic results





David's tumour testing identifies a *KRAS* variant.

Does this mean his children have inherited an increased cancer risk?

A. Yes

B. No

C. Unsure

KRAS: Understanding the tumour

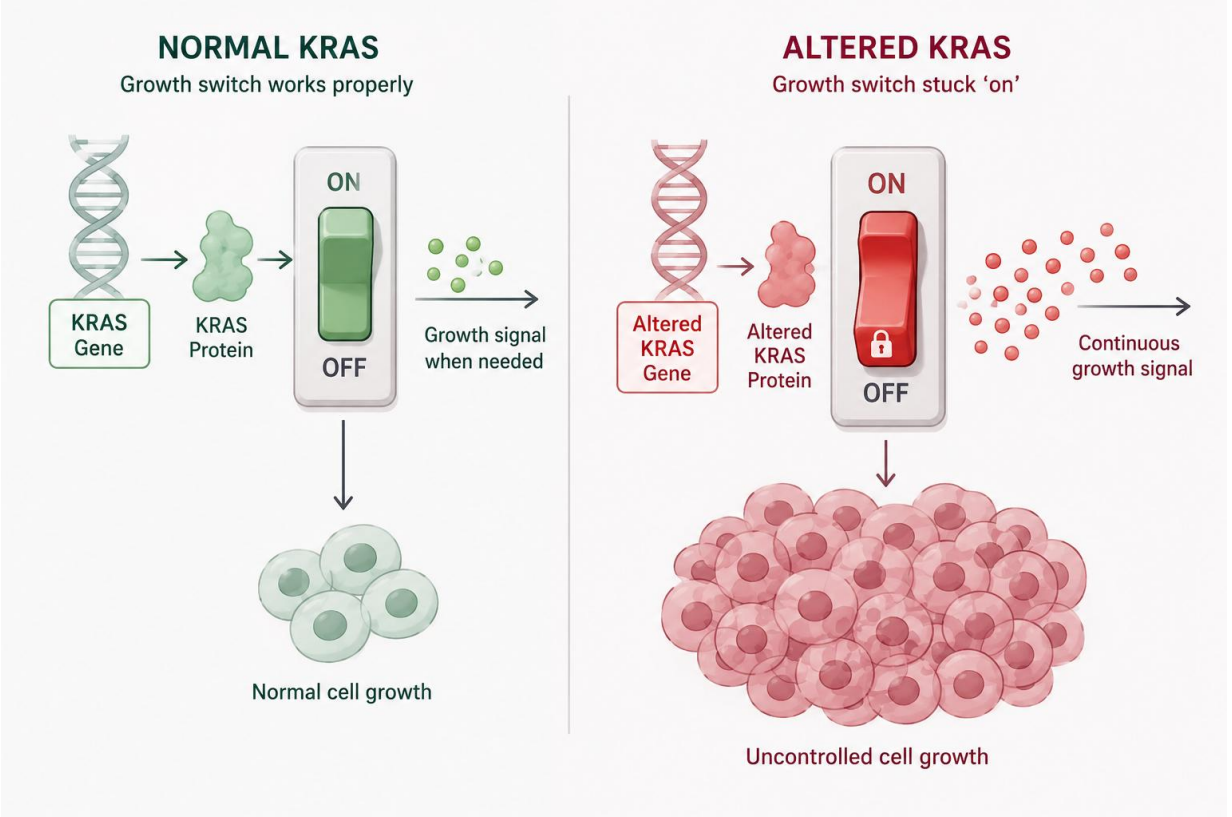
***KRAS* = growth switch**

Normal:
ON ↔ OFF

KRAS variant:
Stuck ON

Tumour change

Future:
KRAS-targeted therapies
Clinical trials



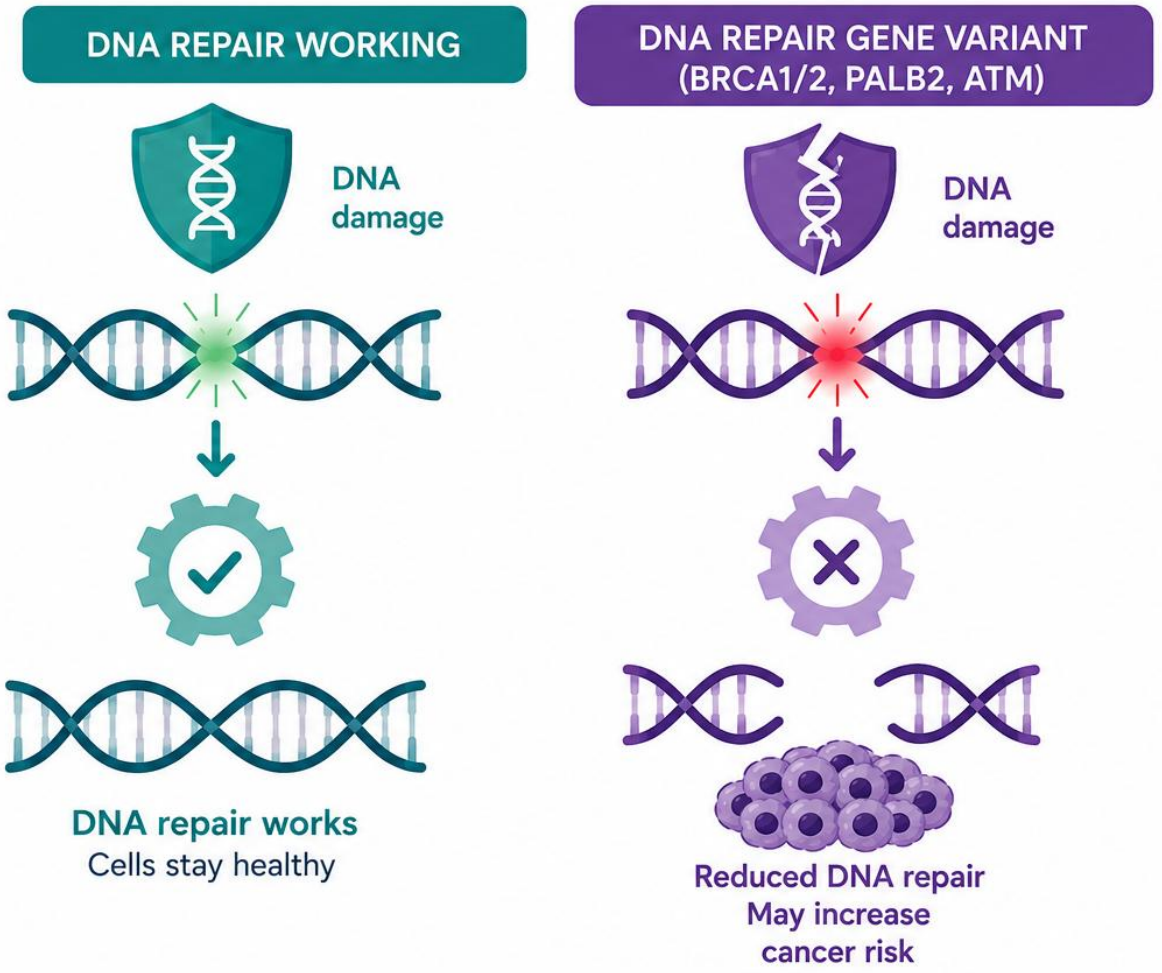
Inherited genomic findings: treatment and family implications

Examples:

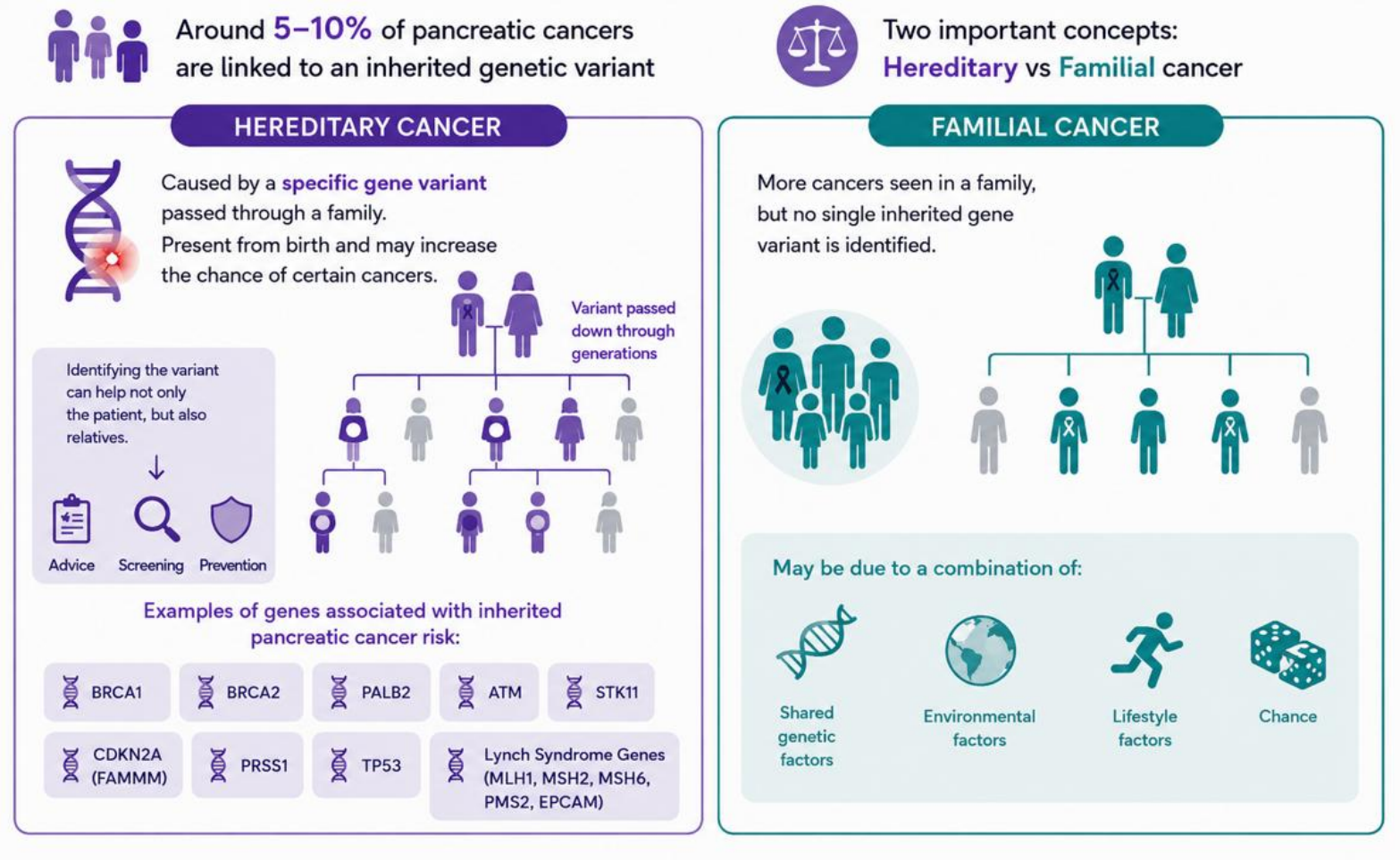
- *BRCA1 / BRCA2*
- *PALB2*
- *ATM*

May influence:

- Platinum-based chemotherapy
- Targeted treatments in some patients
- Clinical trials
- Family risk assessment



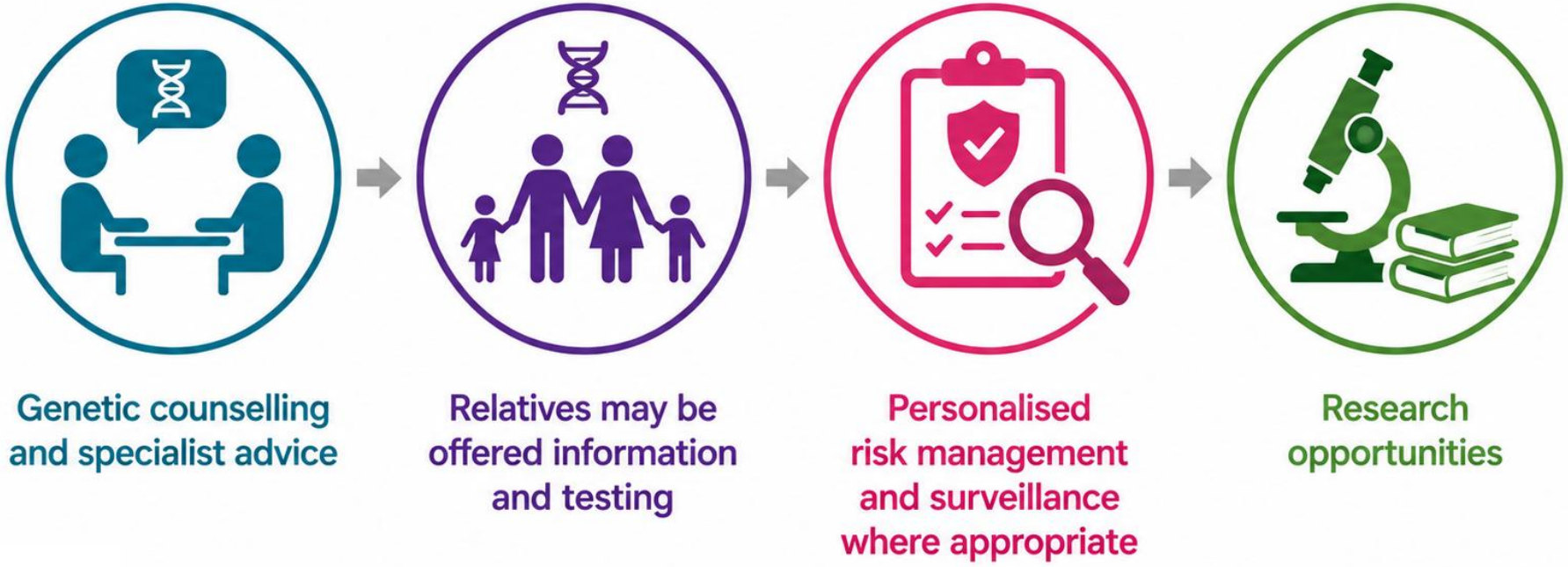
Inherited pancreatic cancer risk



Important!

- Family History
  - Cancer at a young age
  - Multiple Relatives affected
  - Linked cancers e.g. Pancreatic cancer, melanoma, breast, ovarian, bowel or endometrial cancers in the family
  - Known cancer genetic diagnosis

Inherited genomic findings: what happens next?



https://bsgm.org.uk/healthcare-professionals/list-of-genetic-clinics/

Why might genomic testing be done before giving some medicines?

A. To predict every side effect

B. To understand how someone may process or respond to a medicine

C. To diagnose cancer

D. To replace clinical judgement

Genomics and Personalised Medicine – Treatment and Safety

Pharmacogenomics — the patient

DPYD
→ Fluoropyrimidine chemotherapy
→ Reducing risk of severe toxicity

Tumour biomarkers – Understanding the Cancer

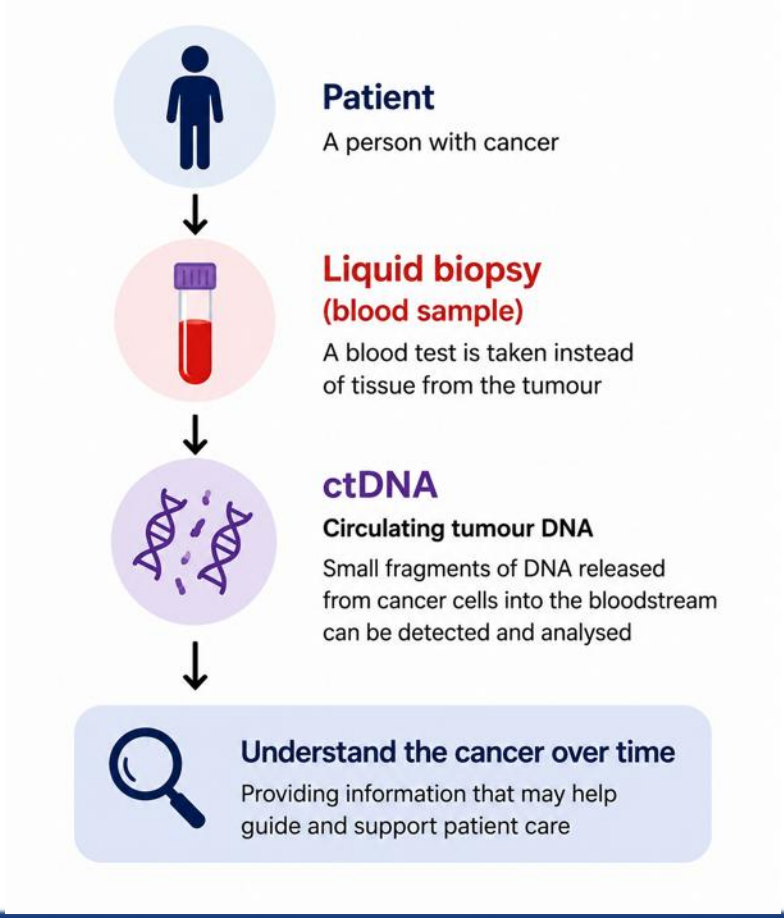
BRCA1/BRCA2
→ DNA repair pathway

MSI-high/MMR deficiency
→ Immunotherapy approaches

KRAS
→ Emerging targeted therapies / trials

NTRK/HER2
→ Targeted therapies

The future: Liquid Biopsy and Circulating Tumour DNA (ctDNA)



- Tumour Information
  - Helps understand what is driving the cancer
- Monitoring
  - Treatment response and recurrence research
- Precision Medicine
  - Supporting future personalised care and clinical trials

Benefits, Challenges and Limitations of Genomic Medicine

Opportunities
Understand the cancer
Personalised treatment
Safer medicines
Family prevention
Research & Innovation

Challenges & Limitations
Uncertain results
Equal access for all
Supporting conversations
Data Security & Trust
Science still evolving

When should we think about genomics?

- At diagnosis – could genomic testing guide care?
- When considering treatment options – Targeted Treatments/Clinical Trials
- When tumour results identify important biomarkers e.g. *BRCA,KRAS, MSI High,PALB2* - Understanding what is driving the cancer
- When family history suggests inherited risk
  - Younger age at diagnosis
  - Multiple relatives with pancreatic, breast, ovarian, prostate or bowel cancer
  - Known familial cancer gene variant
- When results may have implications for relatives

Your role

- Recognise: When genomics might be relevant

- Explain : Basic concepts in plain language

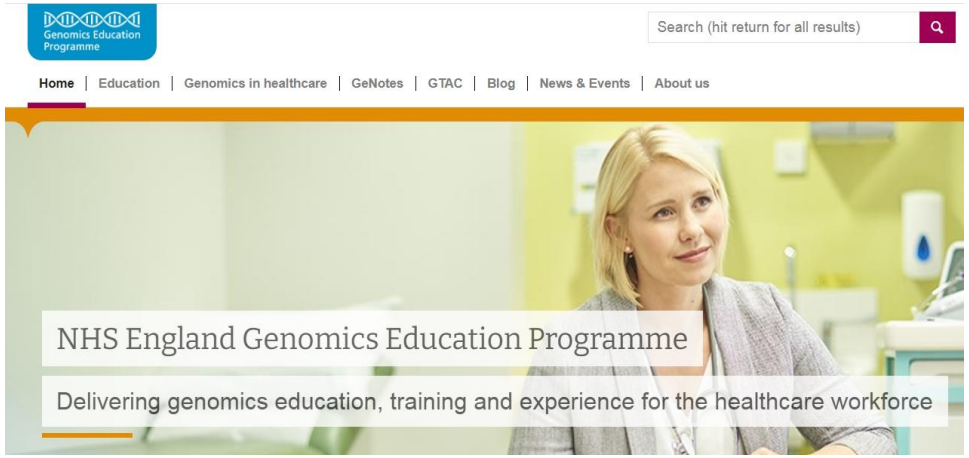
- Support: Patients and Families

- Signpost: Specialist services and Resources

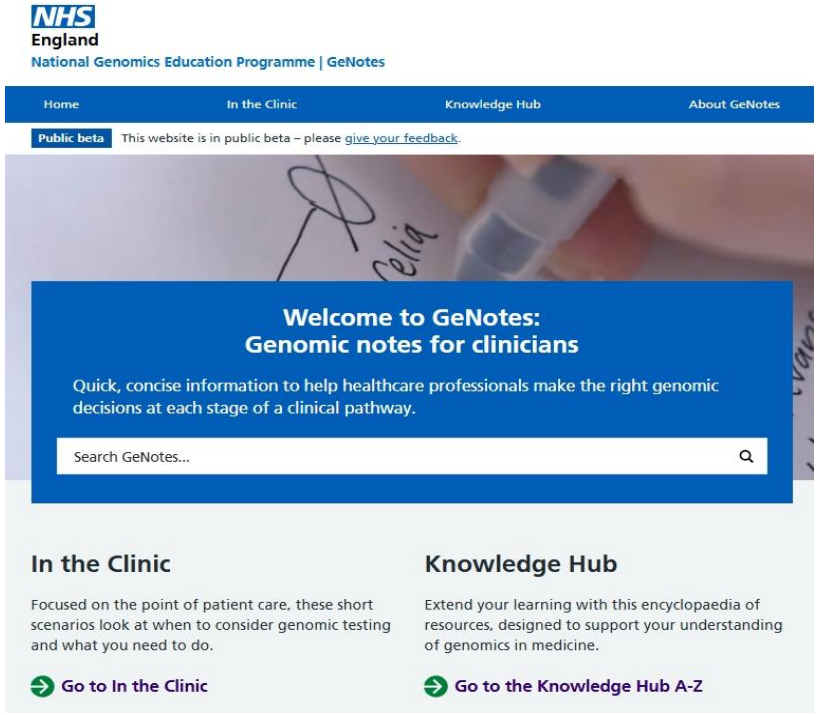
Final take-home messages

- All cancer is genomic, but not all cancer is inherited
- Genomics can influence treatment and make care safer
- Family History Matters: take it and act on it
- Pharmacogenomics supports safer prescribing
- You do not need to be the expert – Recognise, Support and Signpost

Resources



https://www.genomicseducation.hee.nhs.uk/

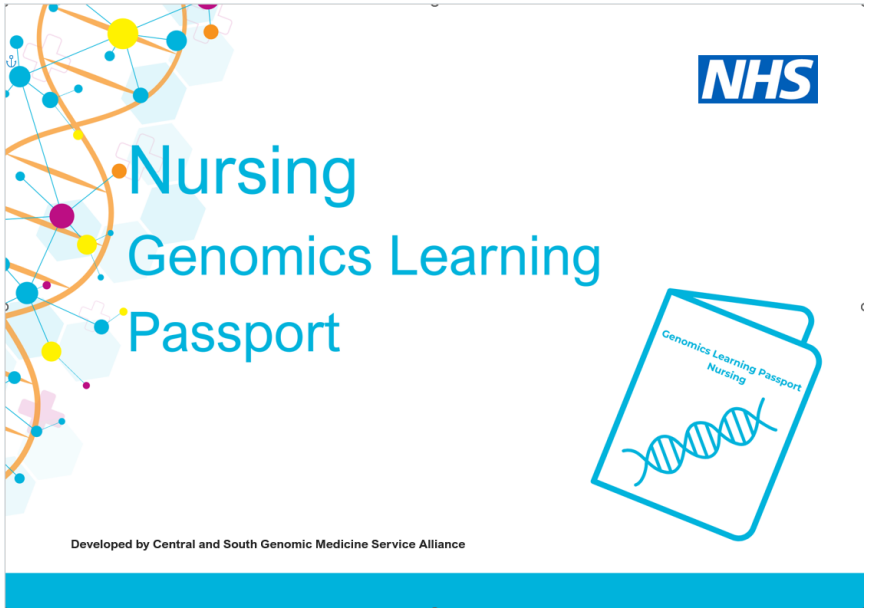


https://www.genomicseducation.hee.nhs.uk/genotes/

Genomic Medicine Service

Genomics Learning Passport for Nurses

- Self-directed tool
- Three levels: baseline essentials, intermediate and specialist.
- Learn at your own pace,
- Directs to trusted resources.

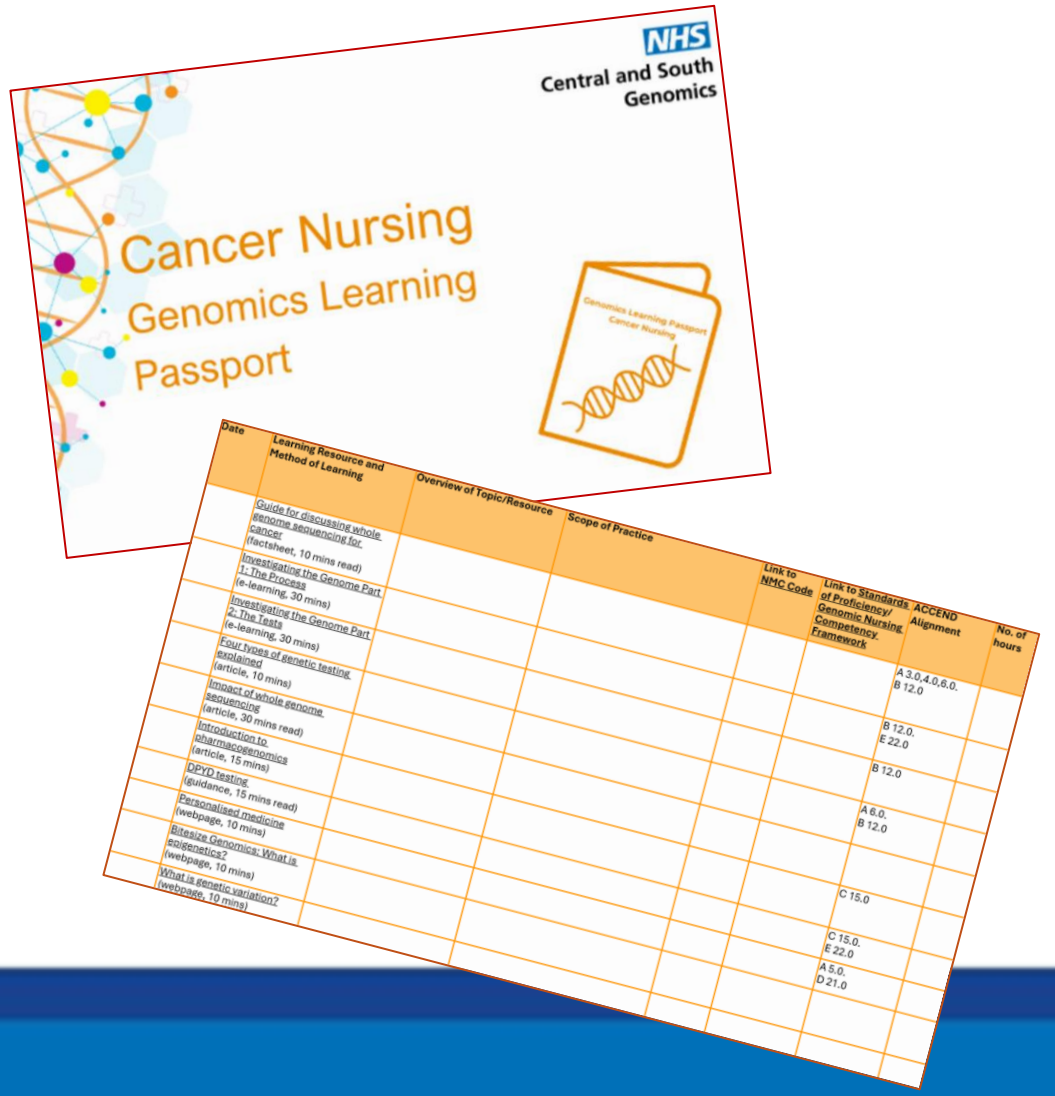


https://www.genomicseducation.hee.nhs.uk/documents/genomics-learning-passport-for-nurses/

Genomics for Cancer Nurses Passport

Available Soon!

- Mapped against specific NMC Code/Standards of Proficiency and ACCEND Framework
- Supports re-validation



References

NHS England.
NHS Genomic Medicine Service: Accelerating genomic medicine in the NHS.
NHS England.
https://www.england.nhs.uk/genomics/nhs-genomic-med-service/

Department of Health and Social Care.
Fit for the Future: 10 Year Health Plan for England.
Department of Health and Social Care / NHS England; 2025.
10 Year Health Plan for England: fit for the future - GOV.UK

NHS England.
National Genomic Test Directory.
NHS England.
https://www.england.nhs.uk/publication/national-genomic-test-directories/

NHS Genomic Medicine Service.
GeNotes: Genomics Notes for Clinicians.
NHS Genomics Education Programme.
https://www.genomicseducation.hee.nhs.uk/genotes/

Genomics Education Programme.
Genomics Learning Passport for Nurses.
https://www.genomicseducation.hee.nhs.uk/

Macmillan Cancer Support.
Genomics Toolkit.
The Macmillan Genomics Toolkit | Macmillan Cancer Support

British Society for Genetic Medicine
Consent, Confidentiality, Clinical Genetics Services
https://bsgm.org.uk/

Golan T, Hammel P, Reni M, et al.
Maintenance Olaparib for Germline BRCA-Mutated Metastatic Pancreatic Cancer.
New England Journal of Medicine. 2019;381:317–327.
Maintenance Olaparib for Germline BRCA-Mutated Metastatic Pancreatic Cancer | Ne England Journal of Medicine

Hu C, Hart SN, Polley EC, et al.
Association Between Inherited Germline Mutations in Cancer Predisposition Genes and Risk of Pancreatic Cancer.
JAMA. 2018;319(23):2401–2409.
Overall survival in patients with pancreatic cancer receiving matched therapies following molecular profiling: a retrospective analysis of the Know Your Tumor registry trial - The Lancet Oncology

British Pharmacological Society & Royal College of Physicians.
Personalised Prescribing: Using pharmacogenomics to improve patient outcomes.
2022.
Personalised prescribing: using pharmacogenomics to improve patient outcomes | RCP

CPIC Guideline.
DPYD genotype and fluoropyrimidine dosing recommendations.
Guideline for DPYD and Fluoropyrimidines

NICE.
Pancreatic cancer in adults: diagnosis and management (NG85).
National Institute for Health and Care Excellence; 2018 (updated).
Overview | Pancreatic cancer in adults: diagnosis and management | Guidance | NICE

Thank you!

Phil

philandra.costello1@nhs.net