Healthier Scotland
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NHS SCOTLAND

Genomics Explained - How Our Future Scientists Will Lead Change

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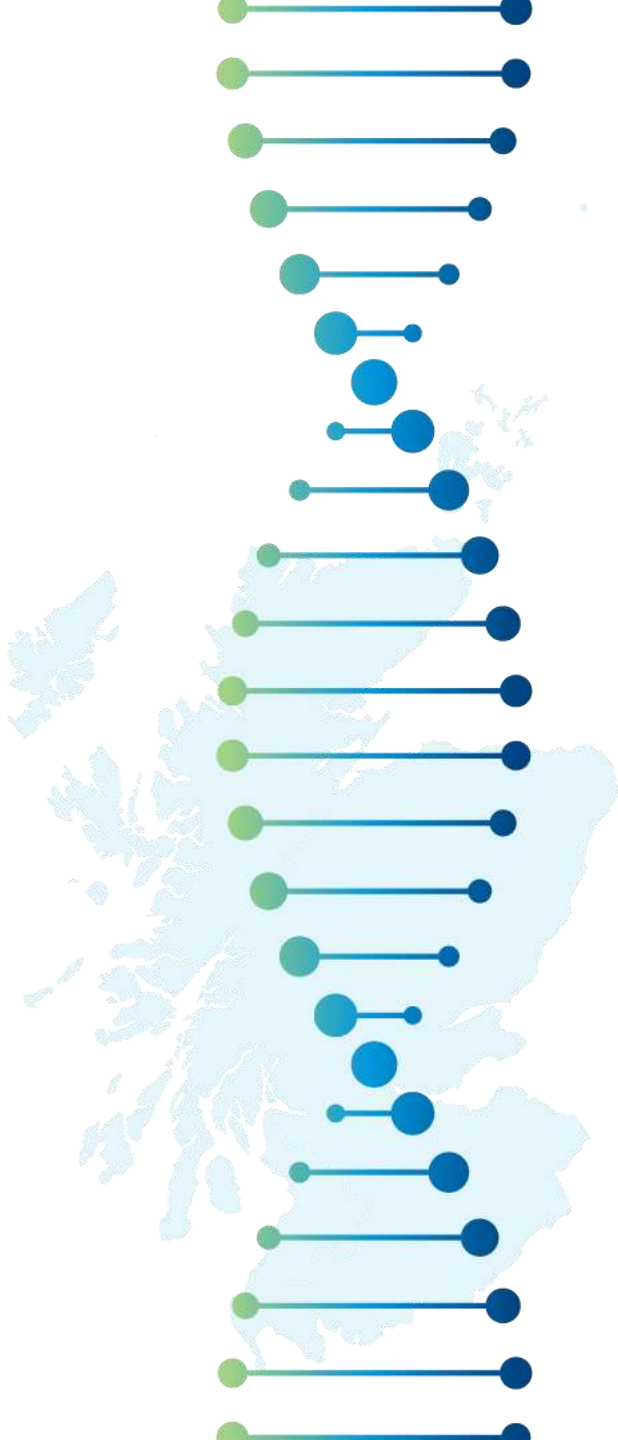


Evolution of NHS Genomic Services: timeline

- The Human Genome Project (HGP) was a research project to sequence the Human Genome.
- Began in 1990 and was ‘completed’ in 2003, 92% by 2003 and fully sequenced by 2022.
- International collaboration involving >20 universities and research centres, cost >$3 billion.
- Today you can sequence and analyse a human genome in <7 days, cost <$1000.
- I became a trainee clinical scientist in 1998 based in the Manchester genetics laboratory.
- New sequencing technology emerged 1995 – 2000, more genetic testing available in the NHS.
- In 1998 genetic laboratory service undertook small scale targeted testing for core/ common genetics inherited disorders.
- In 1998 clinical scientists undertook laboratory wet work and analysis reporting of result.
- By 2022 genetic laboratory services under large scale genomic test for inherited disorders (common and rare disease) and cancer.
- By 2022 clinical scientists office based delivering analysis and reporting of complex results.

Why are NHS Genomic Services Changing?

Gene
1kB data
1-2 variants

Gene panel
50kB data
~ 100 variants

Exome
60MB data
~ 55,000 variants

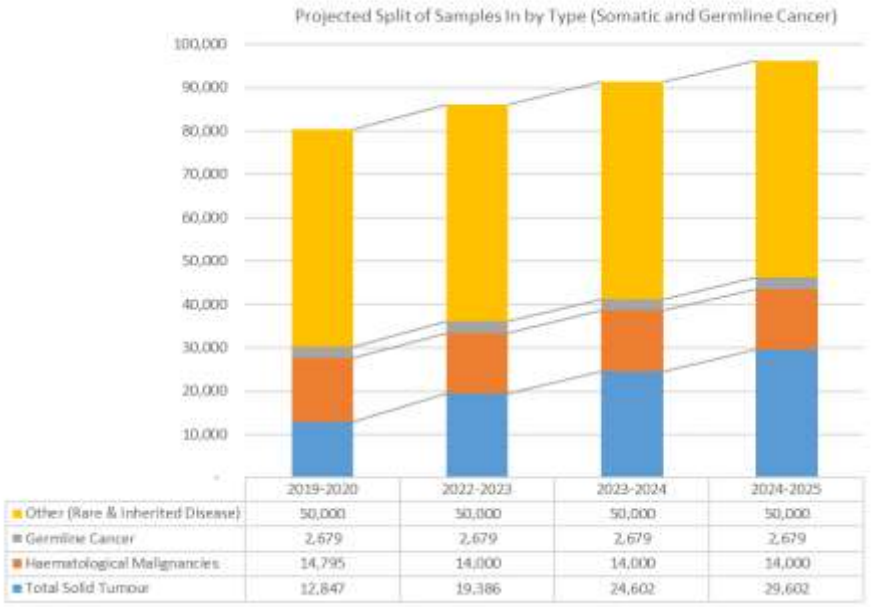
Genome
3.2GB data
~ 3 million variants

1998 → **2022**

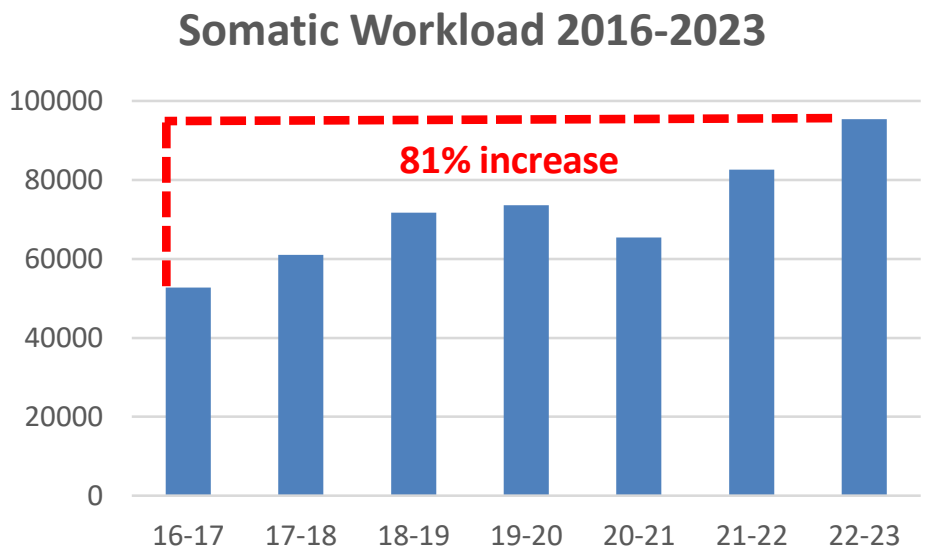
1998 → **2022**

Why are NHS Genomic Services Changing?

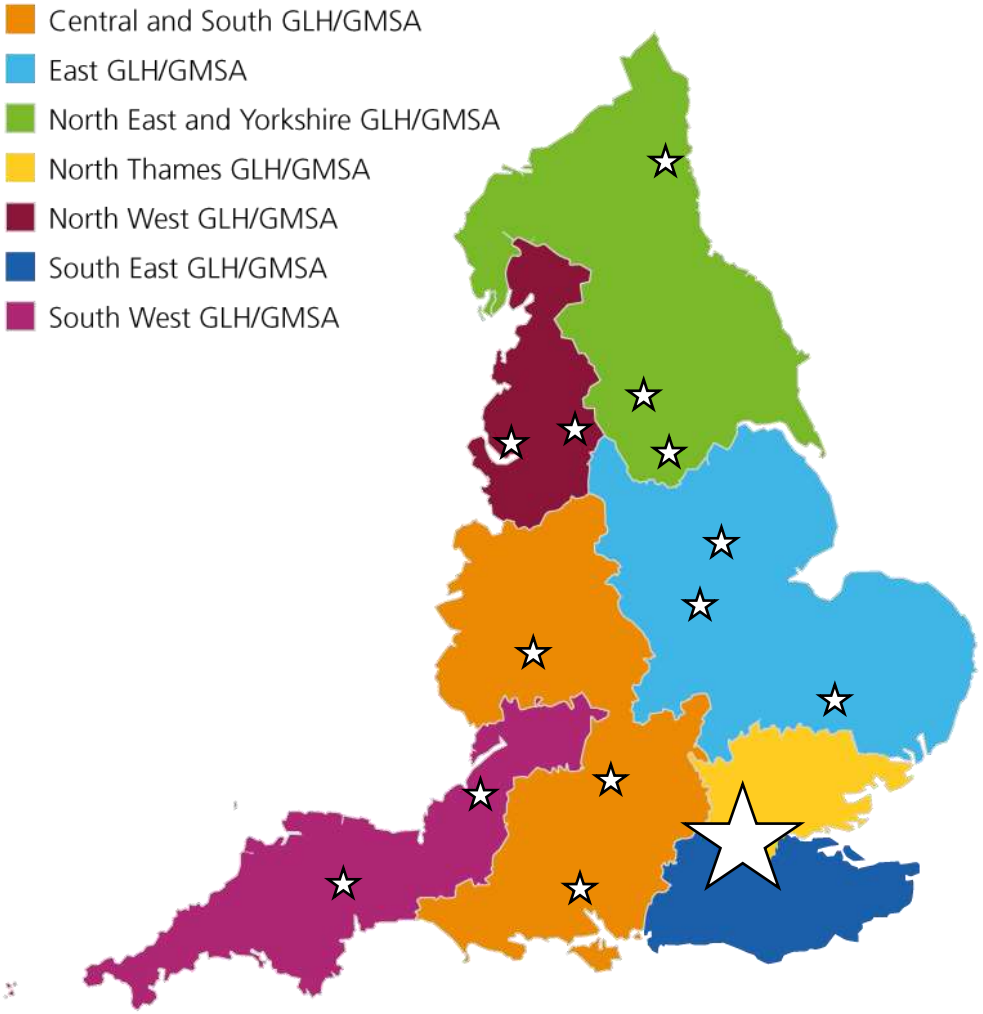
- Large increase in workload, particularly for cancer as genomics is used to determine diagnosis, prognosis and treatment of patients.
- ISD predicts a significant increase in cancer incidence from 2023-2027 when compared to 2008-2012, majority of which will require a genomic test.
- Cost of genomic service in Scotland is also increasing.



	2019-2020	2022-2023	2023-2024	2024-2025
Other (Rare & Inherited Disease)	50,000	50,000	50,000	50,000
Germline Cancer	2,679	2,679	2,679	2,679
Haematological Malignancies	14,795	14,000	14,000	14,000
Total Solid Tumour	12,847	19,386	24,602	29,602



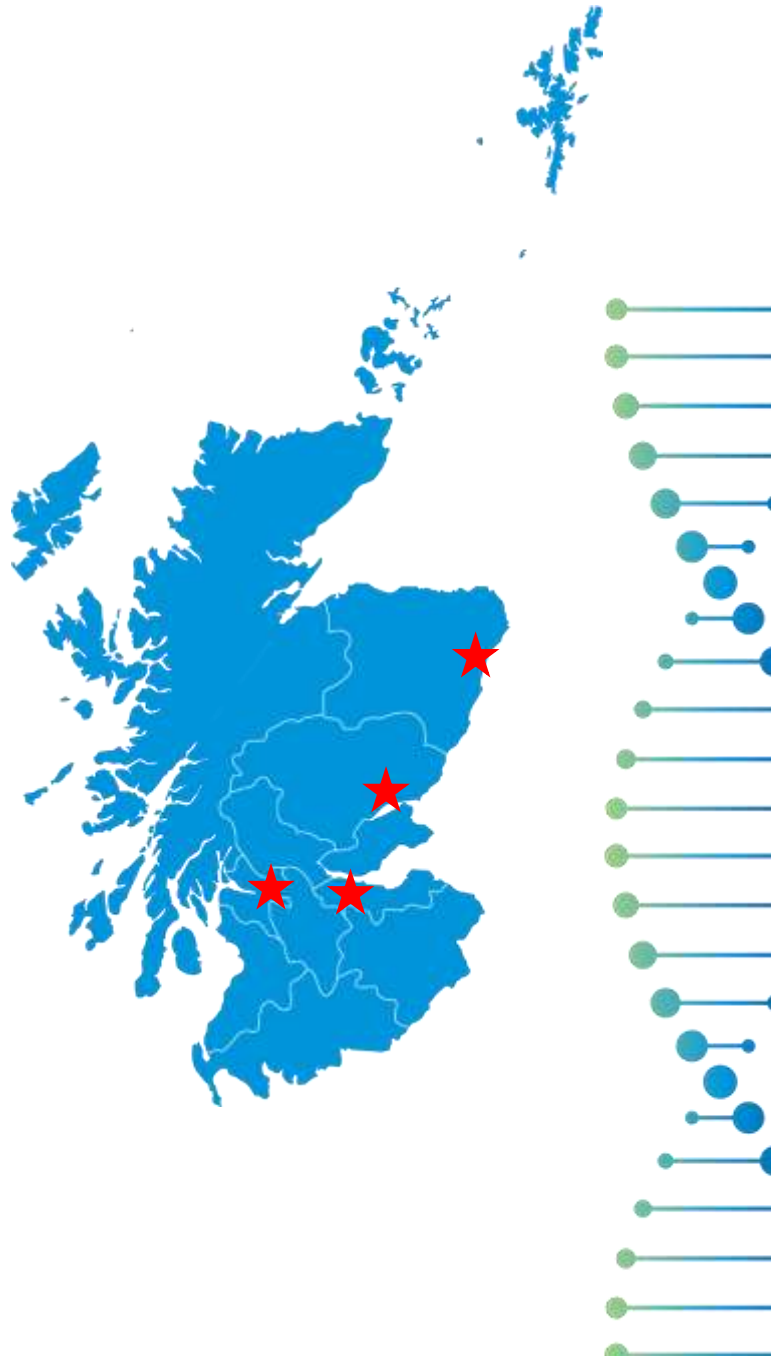
Genomics Landscape in England: 7 Genomic Laboratory Hubs (GLH)



- Population 56.5 million.
- Consolidated 20+ genomic laboratories down to 7 regional Genomic Laboratory Hubs.
- Each GLH serves a population between 7.5 9 million.
- Majority of ‘wet lab’ work has been centralised.
- Analysis and report of results harmonised across a GLH.
- As you scale up sequencing capacity to larger instruments, cost becomes cheaper.
- All GHLs have been tasked with delivering the same Genomic Test Directory = equity NHSE.

Genomics Landscape across in Scotland

- Population 5.5 million (10th size of England).
- 4 regional genomics laboratories located in Aberdeen, Dundee, Edinburgh and Glasgow.
- Aberdeen 17%, Dundee 8%, Edinburgh 26%, Glasgow 49%.
- Inherited disorders delivered as a distributed service delivery model.
- Cancer is largely delivered out of all 4 labs with some specialisation for rare solid tumour and leukaemia.
- Currently no standardisation across Scotland for laboratory wet work or analysis and reporting of results.
- NSD undertook a Major Review and the Genomics Services across Scotland which was published in March 2022.
- 17 recommendation including the formation of a new Genomics Strategic Network and a Genomics Transformation Team.



Transformation of Genomic Services in Scotland

- I’m the scientific lead for the new Scottish Strategic Network for Genomic Medicine (SSNGM) and the lead of the Genomics Transformation Team (GTT).
- 4 people working in the GTT, three of whom are scientists.
- Based within National Service Division (NSD) of National Services Scotland (NSS) and work closely with the Scottish Genomics Policy Team and the NSD Commissioning Team.
- New way of working taking a full system ‘Once for Scotland’ approach by linking transformation, future planning and service delivery to Scottish Government Policy.
- Genomics Transformation Team remit is to deliver the review recommendations and,
  - ✓ Standardisation of data and laboratory processes (including QM).
  - ✓ Financial savings for reinvestment of future services.
  - ✓ Demand optimisation.
  - ✓ Strategic planning and horizon scanning.
  - ✓ Delivery of new services.

Healthcare Scientist Leading Change in Genomics

- Fast moving scientific discipline, move at pace.
- Embracing change and getting involved.
- Moving into new leadership positions (GTT).
- Joining working groups and driving change forward.
- Get on board with the vision.
- Make friends and influence by building your leadership network.
- Small talk matters.
- Find other ways of doing things.
- Ask a friend.
- Be willing to compromise and adjust the plan.

Future Delivery Models: thinking outside the current box

- Current delivery model has four labs in Scotland all working independently.
- This model is not sustainable for the future due to rising cost.
- The infrastructure requirements for whole genome sequencing is expensive and we can’t all do it!
- What are the options?
- Less laboratories or connect the existing laboratories up?
- Moving to a ‘GLH’ like structure for Scotland?
- Digitally connecting to include Scotland wide LIMS?
- Centralised data and IT infrastructure?
- Centralisation of laboratory wet work?
- Outsourcing if cheaper?
- All have pros and cons but ultimately a new delivery model is required to fully transform the genomic services in Scotland.