NHS National Services Scotland

Major Review Report

Genetics & Molecular Pathology Laboratory Service

March 2022

Table of Contents

Executive Summary

The major review of the Scottish Genetics Laboratories took place between January 2021 and March 2022.

The review considered the current approach, the existing capability and capacity in the context of levels of activity and workload across all four Laboratories that comprise the consortia.

This included key processes, the volume and variety of tests provided, laboratory infrastructure, resources and demand and supply issues.

The major review has identified a number of opportunities to right size and future proof the genetic laboratory capability and capacity in Scotland.

A clearer strategy and approach would prioritise developments and opportunities for the laboratories to consolidate and improve services, to provide the basis for the significant increase in demand for existing and future testing services.

A more consistent, standardised approach to laboratory activity reporting would provide a better informed approach to how future demand could be fulfilled by the existing laboratories, absorb increasing demand and accommodate additional demand for new pathways. The current basis for calculating workload (Genetics Units or GenU's) is generally considered no longer fit for purpose and the use of different Laboratory Information Management System (LIMS) raises concerns at a number of levels about the quality and integrity of activity data that is being reported.

The laboratories effectively operate on a standalone basis within a consortia model. Many of the potential benefits of a consortia model including management processes, collaboration and driving improvements are not optimised at a consortia level. Technical excellence and performance at individual laboratories does not compensate for how the consortia model is currently organised and managed.

There is no clear basis to plan for future requirements nor how existing and new scientific and medical roles will be resourced in future in the context of changing testing technology and methods.

The development of the Laboratory Test Directory is a significant development. The Directory provides the basis to promote the service, understand the type and range of tests available and a baseline to understand the dynamics within the consortia and how services could be optimised and improved. The Directories are available in full under Appendix 2-3.

Progress has been made against previous 2017 review recommendations, but some key issues have not been addressed and are still relevant in what is a rapidly evolving and growing service area.

The Review recommends developing a clear strategy and new business model planning approach that delivers Process, Organisational and Technology improvements with more appropriate financial and resource planning to address known issues whilst ensuring the principles of realistic medicine are adhered too.

A User survey in May 2021 indicates that service users were able to access an appropriate range of tests to support clinical decision making and treatment selection with some frustration particularly form Clinical / Medical Oncologists and Pathologist who were least satisfied with the range of services available and feedback about slow reporting times out with the clinical guideline.

30% of staff responded to a Consortium Laboratory Staff Survey in November 2021. The results suggest that laboratory staff want more collaborative working, sharing of best practice between laboratories and to have greater involvement and engagement in making decisions.

Benchmarking Genetic Laboratories in other countries suggests different approaches and many of the aspects of the Scottish “network” model and collaboration are still aspirational features in other countries.

1. Background

1.1 Role of National Services Division

NSD annually receives top-sliced, ring-fenced funding from the Scottish Government Health and Social Care Directorates (SGHSCD) to commission and performance manage nationally designated specialist services and screening programmes, National Managed Clinical & Diagnostic Networks and National Network Management Services. NSD currently commissions more than 121 national designated specialist services including genetics and molecular pathology laboratory testing services on behalf of Scottish Government and NHS Scotland's boards.

National commissioning is reserved for those specialist services where local or regional commissioning is not appropriate and through designation aims to:

- ensure equity of access for all Scottish residents to specialist services
- ensure the best possible clinical outcomes
- provide a secure funded environment for the establishment and development of new national services
- provide a risk-sharing arrangement for NHS Boards where incidence is sporadic and treatment involves specialist skills or expensive equipment
- avoid unnecessary and inappropriate proliferation of duplicate services, thus promoting clinical quality and cost effectiveness

A nationally commissioned service is expected to deliver all aspects of the Quality Ambitions as set out in Scottish Government's Quality Strategy.

NSD works to maximise service delivery, ensuring that patients have access to high-quality service and standards of care that meet set standards.

Each designated service is subject to strict governance and performance reviews. NSD does this by:

- developing service agreements with provider boards, detailing the service specification, performance and quality standards, finance and activity expectations
- conducting regular meetings between service colleagues and health board managers to continually review auditing processes, measure clinical outcomes and identify service improvements
- performing an annual cycle of performance reviews to discuss audit, clinical outcomes and service improvements

1.2 Genetics & Molecular Pathology Services

Molecular genetics testing was nationally designated in 1985 and cytogenetics in 2009. Molecular pathology testing came on line as a nationally commissioned service delivered through the established consortium multi-site model from 1st April 2013

Table 1: Service Designation Timeline

#	Service	Designated
1	Molecular Genetics	1985
2	Cytogenetics	2009
3	Molecular Pathology	2013

Review – Genetics & Molecular Pathology Laboratory Service

NSD commission four regional genetics centres in Aberdeen, Dundee, Edinburgh and Glasgow. The centres work under and a formal consortium arrangement, with the aim of providing equitable, high quality genetic testing service for Scotland.

The funded service though NSD includes;

- Laboratory scientific and technical staff
- Reagents and consumable, including validation costs for new tests / technologies
- £300,000 funding for capital per annum
- 3x Consortia Leads
- Send away test requests for Germline (rare and inherited) conditions

The review of the centrally commissioned, four regional Scottish Genetics Laboratory Consortium (SGLC) which provides testing for rare disease and cancer for Scottish patients has been conducted by NSD who are the national commissioners of the service to ensure that the service is delivered safely & efficiently and has the ability to meet the future needs of this complex service.

An independent review group (IRG) which included experts and service commissioners from across the UK, representation from patient interest / third sector organisations, clinical user for cancer and rare disease and Scottish Government policy representatives, considered the following;

- Updates and Presentations from the Service
- The current Test Directories & Gap Analysis
- The last service review and progress against the recommendations
- Laboratory Activity Data Analysis from previous 5 years Laboratory Annual Reports
- Audit of Scottish Genomics Laboratories (examination of capacity and capability including space, equipment, staffing, IT, future proofing)
- Customer Engagement and User Survey Feedback
- Staff Engagement and Staff Survey Feedback
- PHG Report: International models of service delivery for laboratory genomics

The recommendations formulated from the findings of this review will be presented to the Scottish Genomics Leadership Group (SGLG), who have the responsibility for agreeing the recommendations and the future strategy for genomics for Scotland. The newly formed Scottish Genomic Strategic Network supported by a Transformation Team will be responsible for implementing the agreed recommendations and ensuring that the service if fit for the future.

This will be delivered in a phased approach identifying what can be delivered in the short, medium and long term.

The full list of review recommendations is set out in the Conclusions and Recommendations sections set out at the end of this report.

1.3 Aim and objectives of review

NSD is committed to reporting the outcome of the review to the National Specialist Services Committee (NSSC) which is the governance group for NSD and the Board Chief executives. NSD aims to provide assurance that the commissioned service continues to perform against the designation criteria which includes:

1. Current and predicted future need for the service
2. Equitable access to services for the Scottish population.
3. Continued performance of the service in achieving clinical quality standards / adherence to best practice and any regulatory requirements (i.e. UKAS)
4. Patient outcomes are comparable with other UK and International laboratories
5. The service achieved Financial Balance
6. Continually Horizon Scanning and developing to meet the needs of the Scottish health care system
7. Continually aims to meets customer and staff satisfaction
8. Service efficiency and effectiveness and cost effectiveness
9. The sustainability of the current service
10. Future developments within the services
11. Current issues faced by the service and how they are being addressed
12. How the service should develop over the next five years taking into account future developments
13. Whether the service continues to fit National Specialist Services Committee (NSSC) criteria

Findings from the 2021/22 major review of service against designation criteria are set out in section 13 of this report.

1.4 Approach to Task

This review was undertaken using NSD procedural guidelines for conducting a major review. An Independent Review Group (IRG) was formed (membership as listed in Appendix 1). The Group met three times to reflect on evidence and undertook the following:

Table 2: IRG Meetings Timeline

Milestone	Meeting Focus
June 2021	• Scene Setting, • Presentation from the service,
Sept 2021	• Review of collated activity data, • Capacity Audit, • PHG Foundation presentation: European models of service delivery for laboratory genomics, • Stakeholder / User survey feedback,
Dec 2021	• PHG foundation report: European model of service delivery for genomics, • Consortium Laboratory staff survey feedback, • Consider options for future delivery / conclusions & recommendations.

The review and its conclusions were presented to the NSD SMT on 20th Jan 2022, to the National Professional Patient and Public Reference Group (NPPPRG) on 17th Feb 2022 and the National NSSC on 17th March 2022 for endorsement of the review recommendations. The NSSC advises NHS Board Chief Executives and Scottish Government on the outcome of the reviews and future designation of national services.

The outcomes from the review have also been considered by the Scottish Genomics Leadership Group (SGLG) on the 24th February 2022 and subsequently SGLG secretariat took the approved report to the Cabinet Secretary for Health and Social Care.

1.5 Progress Against Recommendations from the 2017 Review of Service

The previous Review undertaken in 2017 made a number of recommendations. Progress has been made against some recommendations, but others are work in progress or have not been progressed. Actions not completed since 2017 have been considered in terms of their current relevance.

Table 3: Progress Against 2017 Review Recommendations

#	2017 Recommendations	Status
1	A single service agreement / specification is now in place across all four centres.	Completed
2	The Genetics Evaluation Panel (GEP) was established, replacing the previous 'User Group' to aid clinical engagement, transparency of process and rigorous evaluation of new germline testing services to ensure clinically and cost effective delivery of the service.	Completed
3	The role, remit and membership of both Consortia was reviewed and yearly work plans with targeted objectives developed to ensure a coordinated approach to delivering the service for Scotland based on the priorities identified by the GEP, Molecular Pathology Evaluation Panel (MPEP), the Laboratory service, NSD and Scottish Government.	Completed
4	A National Genetics Laboratory Management Committee was in operation Oct 2017 – Dec 2020 with a remit to manage the combined genetics / molecular pathology service on a "Once for Scotland" basis. The committee was rescinded having achieved improved communication, engagement and transparency across the consortium.	Completed
5	Clinical exome sequencing (CES) has been implemented, providing testing that would have previously been sent out-with Scotland.	Completed
6	Whole exome sequencing (WES) trio analysis has been implemented for patients, predominantly children, with developmental delay disorders (DDD).	Completed
7	Non-Invasive Prenatal Testing (NIPT) using cell free foetal DNA was introduced in line with changes to the National Pregnancy Screening Programme from 28 Sept 2020.	Completed
8	Circulating tumour DNA testing has been implemented for Non-Small Cell Lung Cancer (NSCLC).	Completed
#	2017 Recommendations	Status

9	Any future rates of variation for genetic testing should be reviewed to understand demand for testing and the reasons for any variation.	The laboratories have monitored demands on testing and conducted horizon scanning to predict future need – little action taken as to how to address issues associated with increasing workload
10	All sites should continue to review skill mix and service design learning and apply the methods that have been used in each of the centres.	Limited progress
11	The consortia must develop their current working model to ensure ongoing shared learning between centres, undertake collaborative strategic planning and decision making to deliver the most cost effective delivery of the service for Scotland.	Skills mix review / service design discussions tend to be centre centric
		Some sharing as to advances in technology and validations
		Working model is unchanged
		Planning discussions take place but service tends to be responsive rather than strategic due to service / activity pressures
12	NSD and consortia review the responses to the stakeholder survey and the suggestions for change and develop a work plan to respond to comments and issues raised by users completing the survey.	Local lab / clinical engagement reported to be very good regarding the advancement of services and awareness raising events with specialties
		GEP implemented and some initiative undertaken to encourage wider clinical engagement but to limited effect
13	Future model of service should ensure adequate training opportunities and succession planning. A Consortium approach with collaborative working would be an appropriate vehicle to support future sustainability.	Postgraduate Scientist Training Programme now in place
		Workforce / succession planning tends to be centre centric
14	The recording / reporting of activity must be rigorously standardised.	No progress due to challenges with various local LIMS
15	The service should plan for the introduction of NIPD.	Some discussion at GEP as to clinical need / send away requests sent to NHSE labs
16	Continue participating in the UKGTN / Improve application / reporting of GenUs	UKGTN dissolved / recommendation no longer relevant

2. Policy

A Fairer, Greener Scotland: Programme for Government 2021-22 (Published: 7 Sep 2021), references a clear commitment to NHS Scotland to embed and invest in Genomics:

"Over the coming years, through advances in research, medicine and diagnostics, there will be increasing demand for the genetic capacity and capabilities within NHS Scotland. Many of the new medicines being accepted by the Scottish Medicines Consortium require genetic tests. We will invest in the genetic labs and frontline genetics services required to embed genomics into routine healthcare"

Prior to this Scottish Government signalled support for *Genome UK: the future of healthcare* (26 Sept 2020)[1] which sets out a strategic vision to extend the UK's capabilities in genomic healthcare:

"Over the next ten years our ambition is to create the most advanced genomic healthcare system in the world, underpinned by the latest scientific advances, to deliver better health outcomes at lower cost. We will do this by working together across our four nations and reducing boundaries between clinical care and research. We will support earlier detection and faster diagnoses, use genomics to target interventions to specific groups of patients"

Genome UK sets out 3 strategic pillars:

1. Diagnosis
2. Personalised medicine, prevention & early detection
3. Research

These are underpinned by 5 cross-cutting themes covering:

1. patient and workforce engagement,
2. workforce development,
3. data and analytics,
4. industrial growth, and
5. ethical and regulatory frameworks.

Scottish government policy colleagues are currently working in consideration of *Genome UK* and the creation of an Implementation Plan in relation to this, which will inform a broader Scottish strategy for the implementation of genomics into mainstream care.

It is anticipated that this work will be progressed by the Scottish Genomics Leadership Group (SGLG) who will decide the future strategy for genomics for Scotland. The SGLG provides leadership and expertise in the development of Scotland's long-term genomic healthcare agenda. The group will oversee the development and delivery of Scotland's longer-term genomics strategy, implementation and action plans and reports directly to the Cabinet Secretary for Health and Social Care.

Upon the recommendation of the SGLG the Cabinet Secretary for Health and Social Care has approved the establishment of a Scottish Genomic Strategic Network to support the planning, finance and service change for improvement to achieve better patient outcomes and more efficient service delivery models.

3. Impact of COVID-19 Pandemic

Throughout 2020 the provision of routine NHS services across the UK has been impacted by the response to COVID-19 and genomics services have been no exception. In order to

[1] The Genome UK: the future of healthcare, Executive Summary (pp 6), published 26 September 2020 https://assets.publishing.service.gov.uk/government/uploads/system/uploads/attachment_data/file/920378/Genome_UK_-_the_future_of_healthcare.pdf. Accessed; 27/10/2020.

safeguard delivery of vital testing services, laboratories across the UK prioritised testing for the following “Urgent” referrals to support patient care:

- Prenatal testing for rare disorders and the common trisomies.
- Urgent parental testing to inform pregnancy management.
- Solid tumours and haematological malignancies where genetic testing will inform treatment or management.
- Dihydropyrimidine dehydrogenase (DPYD) testing to inform chemotherapy dosage.
- Predictive testing in family members with a high risk of an inherited condition.

Genetics laboratories across Scotland and the rest of the UK mobilised to redeploy staff and equipment to support and add capacity for COVID-19 testing.

Testing reduced as a result on the pause of most non-urgent services across the NHS. The Consortium laboratories continued to accept Non-Urgent referrals and continued to deliver testing where staffing levels allowed caveated by a potential for delayed turnaround times.

Testing levels for all laboratory genetics services have recovered to pre-COVID levels. Measures put in place to aide service recovery included;

- Extended laboratory hours including weekends to enable shift working to reduce the number of laboratory staff on site at any one time.
- Installing software systems to enable test analysis, interpretation and report writing while working from home.

While germline rare disease referrals are resuming normal expected levels of service activity, urgent somatic solid tumour and haematological cancer requests have seen a sharp increase. This activity spike has been associated with resumption of services and patients presenting later than normal due to a delay in engaging with GP and specialist oncology services as a result of the pandemic.

Although the SGLC has been resilient in taking measures to ensure and expedite the recovery of all testing services, it has become apparent that there is an urgent need to start future proofing the service in terms of capacity building and innovation.

It is noted that due to the impact of COVID-19 the focus has been on remobilisation to business as usual. This will continue to be the case until such time as special measures allow resumption of normal activities.

It is noted that due to the adverse impact of the COVID-19 pandemic on the delivery of laboratory services it was agreed that, for the purposes of the review 2019/20 data would be used as the most recent representation of laboratory workload and activity levels.

4. Overview of Service

The Scottish Genetics Laboratory Consortia (SGLC) model comprises four laboratories based in Aberdeen, Dundee, Edinburgh and Glasgow.

Review – Genetics & Molecular Pathology Laboratory Service

Genetic Testing is categorised as;

1. Somatic (acquired diseases),
2. Germline (rare and inherited diseases)
3. Pharmacogenomic (PGx)

The Service is characterised by;

- Samples In,
- Workload,
- Number and types of test and associated scientific effort,
- Reports produced.

Some of these characteristics are straightforward and unambiguous service indicators relating to the quantity and logistics of samples with good high quality information available and reported.

However, due to difficulties encountered by the laboratories in providing data to the level of granularity required the Review was unable to establish the numbers of type of tests for individual testing pathways / clinical indications. Across the laboratory sites the interrogation of data is a manual, labour intensive process and highly dependent on the skill of the individual undertaking the task and could not be completed within the timescale of the Review.

A compromise was agreed to focus on Somatic data only to try and understand the dynamic between the number of tests by test type beyond the current generally discredited workload calculation based on GenUs.

As set out in the consortia test directories the Laboratories provide a large number of tests (404) for different disease types across all four Laboratories (367 Germline, 34 Somatic and 3 Pharmacogenomic tests). Not all tests are provided at all Laboratories and some tests are only provided at one Lab.

Notably Aberdeen provide the largest number of test types (206) and there are 31 test types provided by all four Laboratories. 279 test types are only done at one location. 30 (13 Germline, 17 Somatic and one Pharmacogenomic) tests are provided at all four locations.

This represents a mix of high volume / low variety and low volume / high variety testing.

Table 4: Number of Test by Laboratory Site according the Scottish Laboratory Consortium Test Directories

#	Lab	Test Types	Germline	Somatic	Pharmacogenomic
1	Aberdeen	206	179	26	1
2	Dundee	144	117	24	2
3	Edinburgh	103	73	29	1
4	Glasgow	96	67	27	2

4.1 Laboratory Accommodation / Infrastructure

Key drivers for change:

The review has identified that overall accommodation and current infrastructure arrangements are not fit for optimal service delivery/efficiency/growth and that there is a need for a strategy to deliver resilience, sustainability and to provide the ability to expand and adapt going forward.

The accommodation is in single laboratories at each location except Lothian with laboratories in three separate buildings. A description of current arrangements and challenges is provided in the table 5 below:

Table 5: Laboratory Accommodation

#	*Laboratory*	*Accommodation*	*Description*
1	GGC	Purpose built laboratory with good work flows possible.	Sufficient space available both in laboratory areas and offices with space for expansion at least in the short to medium term.
2	Grampian	Additional space has been made available for short term needs.	A new purpose built state of art laboratory building has been approved by NHSG and work is currently underway to initiate a project board to take this work forward.
3	Lothian	Molecular Pathology is based at RIE, but staff do also work in the Haematopathology laboratory at the WGH and travel between.	The Genomics laboratory is also based at the WGH but in a different building to Haematopathology with no sharing of services.
		The Genomics laboratory, comprising cytogenetic and molecular genetic, is based in two buildings with continual flow of personnel and samples between the two.	Despite the significant and ongoing challenges presented to pre-existing long term plans due to COVID-19 (the impact of which on the nature, extent, timing and resourcing remains uncertain), a number of priorities are being progressed by the service which includes plans to progress an Initial agreement (IA) for a purpose-built laboratory medicine building for histopathology, genetics/molecular diagnostics and Mortuary forensic services in the Edinburgh BioQuarter space.
4	Tayside	Require space for expansion particularly with respect to expansion of Molecular Pathology services.	Potential for additional space if agreement with the university can be reached. However, the workflow pattern falls below the ideal.

4.2 Activity / Reporting

Key Driver for Change:

The significant challenge encountered to capture and report data on the number and type of tests for 40% of the samples received relates to complexities of the germline service. The lack of data capture standardisation across the consortium is a significant problem in terms of planning for future demand, resource and future investment and financing the service.

Table 6: Total Consortium Activity (Germline & Somatic) 2019/20

#	Description (2019/2020)	Quantity	% Growth v 2015
1	Samples In	80,621	21%
2	Workload (GenUs)	521,185	35%
3	Reports	61,101	14%
4	Staff WTE (Lab Staff +Consultant Time)	212.59	~

The total number of samples received (Samples In / received) in 2019/20 was 80,000.

The samples received has grown by 21% since 2015.

By whatever measure the laboratories are delivering more activity and workload since 2015 in 2019/20 with effectively the same number of staff. By all accounts the laboratories have no capacity to absorb more activity and workload in the next five-year period to 2025.

The major challenge is to right size the consortium laboratories to cope with future increasing demand.

The Laboratories provided a further breakdown of activity for Cancer testing. This comprises both Somatic and Germline testing. This includes 30,000 Samples In / received, comprising 27,000 Somatic and 2,700 Germline cancer samples for testing.

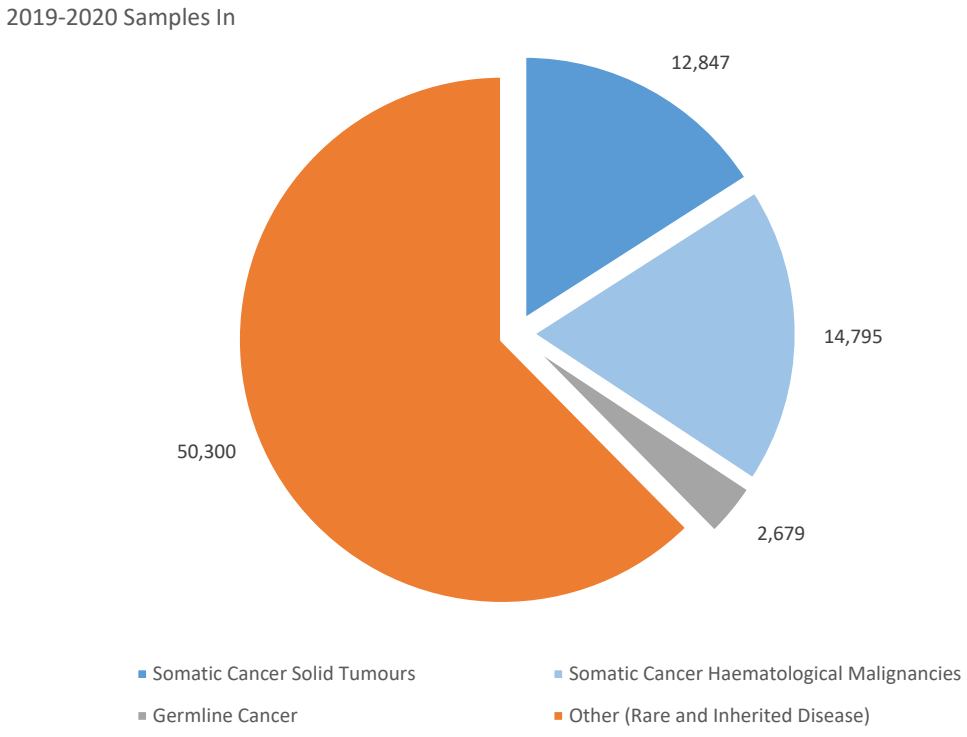


Table 7: 2019/20 Consortium Samples in (Test Requests Received) by Cancer Type

#	Cancer Testing Samples In	Totals
1	Somatic Cancer Solid Tumours	12,847
2	Somatic Cancer Haematological Malignancies	14,795
3	Germline Cancer	2,679
4	Other (Rare and Inherited Disease)	50,300
5	Total	80,621

This analysis highlighted the differences between how the laboratories recorded and presented information and some of the practical difficulties this causes as a result of individual site reporting.
With no consistency in how information is recorded and reported future planning based on multiple criteria becomes more complex and difficult, as does how the Laboratories will be able to absorb future demand and the bottlenecks in this process.

The most obvious basis for future planning would be workload (GenUs) but this is now generally discredited as a reliable method of determining workload associated with testing when it is applied inconsistently for between laboratories for germline and was not intended for use in the somatic setting.

The level of workload in the future will increasingly be determined by new technology and testing methodologies, it is therefore essential to ensure optimum skill mix of laboratory staff profiles to efficiently exploit advances in sequencing technology and maximise to use of automation.

Broadly speaking there are currently there are 12 test types for Somatic and 3 test types for Germline cancer testing in use across the consortium. The laboratories have different protocols about how many tests they apply to samples to achieve a diagnosis which is another complication if relative comparisons are considered e.g. more workload generated for each clinical indication when multiple test are applied.

The levels of Activity and Workload across the Laboratories breaks down as set out below.

Table8: Activity & Workload across the Consortium Laboratories

	Laboratory	Quantity	%
Samples In (Activity)	Aberdeen	12,972	16%
	Dundee	8,871	11%
	Edinburgh	24,749	31%
	Glasgow	34,029	42%
	Totals		80,621
GenUs (Workload)	Aberdeen	131,313	25%
	Dundee	75,710	15%
	Edinburgh	93,788	18%
	Glasgow	220,374	42%
	Totals		521,185
Tests	Aberdeen	3,732	13%
	Dundee	5,371	19%
	Edinburgh	7,274	25%
	Glasgow	12,285	43%
	Totals		28,662

Reports	Aberdeen	9,424	15%
	Dundee	8,292	14%
	Edinburgh	15,738	26%
	Glasgow	27,647	45%
	Totals		61,101
WTE (Lab Staff +Consultant Time)	Aberdeen	32.7	15%
	Dundee	33.64	16%
	Edinburgh	53.36	25%
	Glasgow	92.89	44%
	Totals		212.59

There are different Test Types applied for Somatic and Genetic cancer testing by Laboratories, which includes up to 16 Somatic Cancer Test Types and three Germline Cancer Test Types.

Table 9: Test Methodologies Utilised by the Consortium Laboratories for Somatic & Germline Cancer

#	Testing Type	Somatic	Germline Cancer
1	PCR-FLA	●	
2	FFPE FISH	●	
3	qRT-PCR	●	
4	Sanger	●	●
5	NGS	●	●
6	Cell Suspension FISH	●	
7	Karotype	●	
8	MRD ASO qPCR	●	
9	RT-PCR	●	
10	MRD Work Up	●	
11	IHC	●	
12	Methylation	●	
13	Allele Specific PCR (COBAS)	●	
14	High Resolution Melt	●	
15	SNP Array	●	
16	Pyrosequencing	●	
17	MLPA		●

The data provided by the laboratories included an analysis for Somatic Cancer Testing for Solid Tumours and Haematological Malignancies. This accounted for approximately 40% of Samples In (received) by the laboratories in 2019/20.

While the Scottish consortium laboratories are largely self-sufficient, a proportion of testing is sent out with Scotland, predominantly to NHS laboratories in the UK. Approximately 2700 were sent away to Laboratories outside the Consortia in 2019/20.

A full breakdown of laboratory activity used for the review is available in Appendix 4-9.

4.3 Service Model

Key Driver for Change:

There is a requirement to optimise service delivery to achieve equity of access to testing services and release efficiencies to reinvest resource in new services. Leadership with authoritative decision making and lines of accountability are required to take this forward to ensure delivery of the national strategy for genomics.

The demand for future services is currently based on the addition of 12 new Pathways. Combined with generalisations about the exponential growth in Workload based on historical and Public Health Scotland data for increased cancer incidences the 12 new Pathways provide a basis to quantify the level of growth required for Somatic cancer testing.

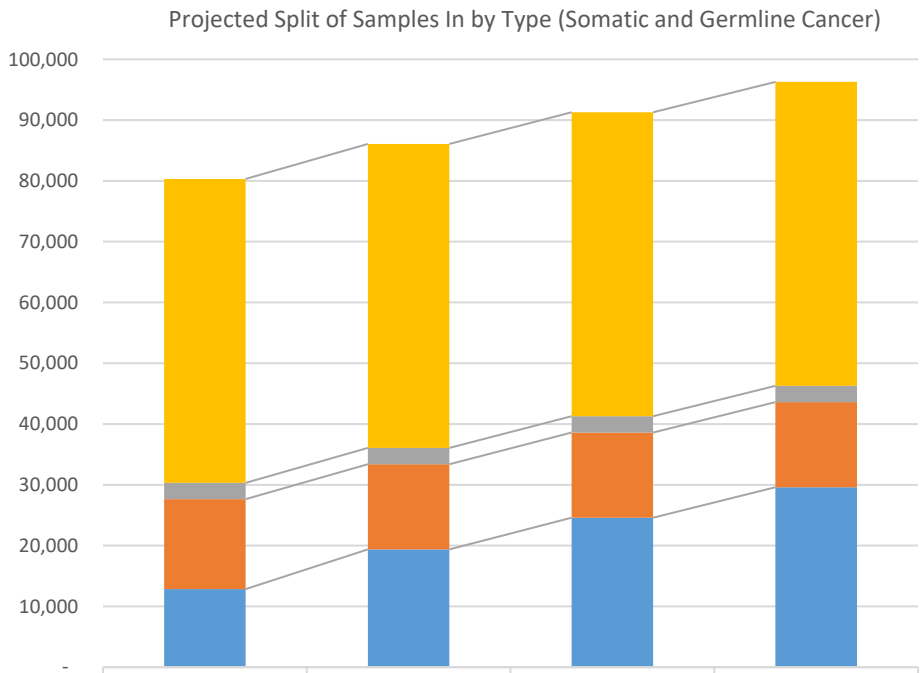
The Laboratories will have to provide tests for somewhere in the region of an additional 20,584 cancer tests to accommodate increases of Cancer incidences in the period 2023 to 2027 (versus 2018 to 2022) based on Public Health Scotland data[2].
The 12 prioritised Cancer Pathways provide more specific detail.

Table 10: Cancer Testing Pathways Prioritised for Implementation

#	Pathway	Yr1	Yr2	Yr3
1	Dihydropyrimidine Dehydrogenase (DPYD)	3,000	3,000	3,000
2	NTRK & core solid tumour fusion gene testing	1,300	5,000	10,000
3	Rare fusion gene testing pathway	115	115	115
4	Thyroid cancer molecular testing	355	355	355
5	Somatic BRCA1/BRCA2 gene testing for ovarian cancer	250	250	250
6	Endometrial cancer molecular testing	160	320	320
7	Molecular testing for patients with breast cancer	113	225	225
8	Neuropathology molecular testing	240	480	480
9	Renal cancer molecular testing	83	165	165
10	Acute Myeloid Leukaemia (AML) minimal residual disease (MRD) monitoring	143	285	285
11	Lymphoid malignancy molecular testing	280	560	560
12	Prostate Cancer (germline / somatic)	500	1,000	1,000
	Additional total Patients per annum	6,539	11,755	16,755

This would mean an increase by Year 3 of 17,000 additional Samples In for Somatic cancer testing. The 2019/20 level of Sample In for Solid Tumour Cancer Testing is 13,000. The new Pathways will add more testing requirements in this one category alone (everything else being equal) than the Laboratories do at the moment.

[2] https://www.isdscotland.org/Health-Topics/Cancer/Cancer-Statistics/Incidence-Projections/
Accessed Monday 8th November 2021



	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

Considering the 12 new Pathways in isolation does not consider growth in other categories of testing.

5. Finance

Key Driver for Change:

There is a requirement to embed Demand Optimisation strategies into testing models to ensure the identification of unwarranted variation across genetic testing facilitates, the identification and implementation of interventions that will drive more appropriate testing and use of resources and ultimately better patient outcomes. The need to ensure financial sustainability of the service through optimal service delivery is essential going forward.

High level analysis of consortia budget versus actual spend taken from laboratory annual reports set out in table 11 demonstrates an increasing negative variance.

Despite annual incremental increases or budget uplifts, to accommodate rising costs (e.g. NHS staff pay), the variance has continued to grow.

There was an 3% difference between actual spend versus budget in 2015/16. This has grown to 15% in 2019/20.

Table 11: Laboratory Annual Report Budgets versus Actual 2015/16 to 2019/20 and % Variance

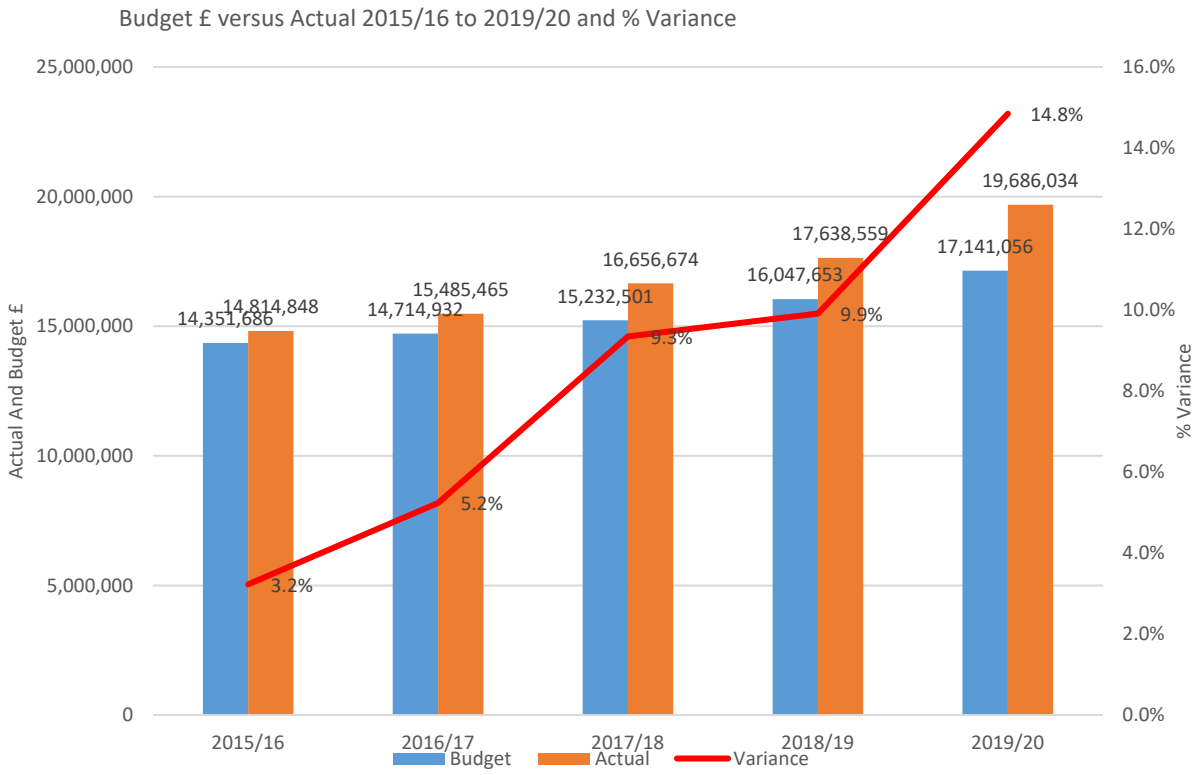


Table 12 provides a detailed breakdown of the consortia budget for 2019/20. The table shows the 2019/20 negative budget variance broken down including,

- an underspend of staff, largely attributable to vacant posts,
- the largest variance is laboratory supplies, reagent and consumable (R&C) costs which are directly linked to testing activity,
- income from Tests was 9% less than budgeted, due to greater self-sufficiency of other NHS laboratories across the UK there was a reduction of test requests from out-with Scotland.

Table 12: Breakdown of the NSD Consortia Budget for 2019/20

Consortium Budget 2019-20			
Service Level Agreement (SLA)	NSD Budget	Actual Spend	Variance
SLA Salaries	£11,530,121	£10,657,648	£872,473
SLA Supplies (R&C) / Overheads	£6,241,681	£7,706,074	-£1,464,393
SLA Income from Tests	-£613,033	-£556,309	-£56,724
SLA Total	£17,158,769	£17,807,413	-£648,644
Additional Investment (not in SLA)			
Send-away Test Request (Out of Area Budget)	£750,000	£742,307	£7,693
Bridge Salaries	£544,435	£544,435	£0
Bridge Supplies (R&C) / Overheads	£591,879	£591,879	£0
Total	£19,045,083	£19,686,034	-£640,951

Furthermore, budgets as set out in laboratory Service Level Agreements (SLAs) do not reflect,

- Non-recurring additional investment, such as the Bridge for a Scottish Strategy for Genomics funding (form the Scottish Government, an additional £1.1 Million in 2019/20),
- Consortium spend on “send away” These tests are sent out with the consortium (usually to other NHS laboratories across the UK) for provision. Test requests which is covered by the NSD out of area (OOA) budget.

Bridge funding was utilised to implement exome sequencing services across all laboratory sites. Figures set out in table 12 reflect actual allocation from Scottish Government to cover 2019/20 salaries and activity.

Cost associated with send-away test requests fluctuates year on year. Actual send-away costs are covered by the NSD out of area (OOA) budget, from which there was a nominal allocation of £750k for 2019/20.

When additional non-recurring investment is factored in there remains an overspend of ~£650k for 2019/20.

The high level nature of financial reporting for staff, consumables and testing does not provide the level of granularity required to accurately identify costs of specific services and testing being undertaken to understand high volume / low variety and low volume / high variety testing pathways.

In the context of increasing demand and already known service constraints around staff, laboratory accommodation and technology and equipment and the significance of Genetic testing and implications for other services and significant future investment decisions understanding the costs of different methods of testing and for different disease types should be a priority for the service to quantify and report on a value for money transparent basis in order to plan for the future and secure funding on a sustainable basis.

6. Equipment & Technology

Key Driver for Change:

Underutilisation and outdated technology are key issues identified by the review process.

There is a large volume of aging equipment across the laboratories that needs to be replaced because it is either no longer supported or has reached the end of its expected life span. Accurate data is required to ensure a strategic approach to inform procurement and optimal use of new, large scale sequencing technologies.

In general, laboratories have capacity on current equipment for the existing services.

NSD allocates £300,000 funding for capital per annum to the commissioned service, this arrangement has been in place since the Calman Review (2006). The service has expanded exponentially since then, as has the cost of laboratory equipment.

The laboratories are at liberty to seek capital investment from alternative sources. The laboratories have noted that support from local health boards for purchase of new equipment is variable.

Prioritised bids for the consortium capital for 2021 totalled over £1.4 million. This includes equipment that needs to be replaced either because it no longer supported or which had reached the end of its expected life span. However, the laboratories have all also indicated a need to expand next generation sequencing services which requires the additional equipment accounting for close to £4 million worth of investment.

The laboratories engage with procurement at board level, although they can draw on national frameworks where available and invest in equipment to suit local workflows. Differences in equipment / platforms has proven beneficial in terms of resilience in the past. Where there have been problems with supply chain for reagents and consumables disabling testing services in one centre another consortium centre can take on the addition work to cover the service in the short term until the issue is resolved.

However, negotiations at a local level means that the consortium may be missing out on discounts for multiple pieces of the same equipment. Furthermore, an audit of the service revealed that there are significant differences in maintenance contacts for seemingly identical pieces of equipment across the sites.

7. Data Storage, Sharing & Laboratory Information Systems (LIMS)

Key Driver for Change:

Currently there is an inability to gather standardised, robust data timeously to inform service development, improvement and efficiencies.

As highlighted earlier in the report acquiring data for the purposes of the review of service proved challenging. Not only were there challenges due the different LIMS systems being used across the laboratory sites but the interrogation of data is a manual, labour intensive process and highly dependent on the skill of the individual undertaking the task and their interpretation of the nature of the request. Also relevant is that systems were set up when different technologies were in use and newer technologies are not always captured within LIMS.

A national LIMS system for Pathology has gone out to tender. This is a £216 million project, delivery of which is planned for by the end of 2025. Genomics has been included in this planned development. There is concern that functionality of this system could be limited for genomics as the requirements are very different to other pathology disciplines.

Currently the laboratories use different LIMS systems. This is not just between regions but within them. In Lothian, cytogenetics, molecular genetics and molecular pathology all have different systems which do not 'talk' to each other.

There are varying levels of IT / eHealth support across the consortium laboratories, on occasions the lack of IT support is a major barrier to developments. Examples given included delays to networking of new equipment for over 12 months and delays in installing software. In some instances, the laboratories themselves maintain and develop their systems. The cytogenetics LIMS in Lothian and the LIMS system in Tayside are maintained by a single member of staff which is not sustainable.

The laboratories also express a need for improved ordering of tests and reporting of results directly into the clinical systems. This point was reflected in both the clinical user survey and the survey of consortium laboratory staff.

All of the consortium laboratories have raised the major requirement for scalable data storage and the ability to share data between consortium laboratories. There is the potential to further advance a genomics instance within the National Digital Platform in collaboration with the NES digital services under a work-stream of the bridge to a Scottish strategy for genomics. This has been delayed due to COVID but it is hoped that this work will continue to be supported at a policy level going forward.

8. Workforce

Key Driver for Change:

- While high level comparison has been made with workforce infrastructure / skill mix in other UK genomic centres, a detailed examination should be undertaken to explore what lessons can be learned to inform the way forward for genomics in Scotland.
- There is no consortium wide workforce and succession planning for a significant number of staff retiring over the next few years.
- Staffing levels is having an adverse impact on capacity to increase the volume of testing and to facilitate service advancement.
- Currently there is no bioinformatic support within the NHS genomics laboratory workforce and no bioinformatic training in Scotland

Table 13 below displays NSD funded consortium staff numbers as set out in Service Level Agreements (SLA). These numbers do not include staff on fixed term contracts under temporary funding arrangements, staff time secured though research collaborations or trainees numbers.

NSD does fund some consultant time to support the laboratories advance testing services, the development of testing panels and interpretation / reporting of patient results.

*Table 13: NSD Consortia SLA Staff Profile 2019/20**

		Glasgow	Grampian	Lothian	Tayside	Total
Admin	**Band 2**	1	2	0	1	4
Admin	**Band 3**	1	0	1	0	2
Lab	**Band 3**	7	1	2	2	12
Admin	**Band 4**	3.16	0	2.2	1	6.36
Lab	**Band 4**	4	3.15	5	5.4	17.55
Lab	**Band 5**	15.57	3	11	1.8	31.37
Lab	**Band 6**	12.1	8	4.8	4.4	29.3
Lab	**Band 7**	37.85	5.65	23.7	11.84	79.04
Lab	**Band 8a**	7.8	1.5	5.94	3.8	19.04
Lab	**Band 8b**	4	1	2	0	7
Lab	**Band 8c**	1	4	0	1	6
Lab	**Band 8d**	1	1	2	1	5
Lab	**Band 9**	1	2	0	1	4
	Total Lab staff	**96.48**	**32.3**	**59.64**	**34.24**	**222.66**
	Lab staff	**£4,579,730**	**£1,780,415**	**£2,957,126**	**£1,729,549**	**£11,046,820**
	Consultant Time	**£197,123**	**£57,192**	**£209,367**	**£19,619**	**£483,301**
	Total staff costs	**£4,776,853**	**£1,837,607**	**£3,166,493**	**£1,749,168**	**£11,530,121**

**The table does not include additional staff appointed with temporary 'bridge' funding, see section 5*

The laboratories have highlighted that there needs to be a review of the workforce including skill mix, given the changes in technologies and service requirements.

Currently there is no bioinformatic training in Scotland. There is a need to assess the requirement for bioinformatician posts in the diagnostic service in Scotland to advance pipelines to process and analyse genomic and molecular data.

8.1 Scientist Training

NHS Education Scotland (NES) support the training and development of postgraduate scientist staff and other key groups in the healthcare science workforce. For Healthcare Science staff, NES commission around 20 supernumerary trainees annually, training involves undertaking a 3-year Scientist Training Post (STP). It is noted that funded trainee posts are distributed across a number of specialties, the consortium have been fortunate to secure between 4 and 7 trainees consecutively in recent years (table14).

Table 14: Allocation of NES funded STPs 2016-2021

The annual allocation of NES funded STPs were as follows:						
Year	2016	2017	2018	2019	2020	2021
Trainee Posts	4	5	5	7	6	7

9. User & Staff Surveys

9.1 User Survey

A User Survey to seek feedback from clinical users of the service was undertaken in May 2021. The survey attempted to reach a broad range of clinical users including members of the governance structure and regional cancer networks. The survey attracted 233 responses from 11 of the 14 regional Health Boards inclusive of 38 clinical specialties and general practice.

- 77% "were of the opinion that they are currently able to access an appropriate range of tests to support clinical decision making and treatment selection",
- 5% of the 233 respondents felt "that they did not have access to a sufficient range of tests to support their clinical practice",
    - the most dissatisfied group of clinicians were Clinical / Medical Oncologists and Pathologists,
- 69% were "satisfied with the responsiveness of the service and reporting times",
- 9% noted that "that results were slow or that reporting times were beyond those set out in clinical guidelines".

Eight references (3%) were made to incompatibility of electronic requesting / reporting software and inconvenience associated with this in accessing test results. Also, where results are emailed to individual requesting clinician there is no resilience during periods of staff absence, causing further delay in chasing patient results.

In response to being asked if the laboratory reports were clear and understandable, 5% of those surveyed found laboratory reports difficult to understand noting the use of complex, technical language and 'jargon'. However, the majority went on to note that the labs were very responsive and helpful when assistance in interpretation was requested. The User Survey is included in full as Appendix 10.

Key / recurring themes raised by clinical colleagues surveyed include;

- Electronic requesting / reporting and integration with local software systems / patient care record. Some highlighted concerns where results are emailed to a single clinician and delay caused in obtaining results associated with staff absence

- Calls for sustained investment in service improvement in particular NGS for personalised / precision care for cancer patients
- Inequity of services in Scotland to the standard testing available across the rest of the UK and the need to bridge the gap
- Greater integration with other laboratory services (e.g. histopathology, IHC, cellular pathology) and more collaborative working on testing pathways.
- Greater interaction with the clinical specialties including education events / information sessions user groups

9.2 Staff Survey

A Staff Survey was undertaken in November 2021, 74 Staff (30%) responded across all four laboratory Host Boards.

- 34% felt that "sharing experience / best practice across the laboratories" was a key feature of the consortia model,
- 23% thought that "open / transparent communication" was considered in service developments,
- 38% thought "workload was distributed" across laboratory sites,
- 51% thought that "the service delivers equitable access to testing for clinical user / patients",
- 62% felt the consortia "provides contingency / resilience in the event of any incidents which would impact on delivery of the service",
- 41% felt "new services were implemented in accordance with clinical need / best practice guidance",
- 45% felt the service is "sustainable / fit for future developments",
- 12% "feel that they are involved in decision making and able to influence changes within the consortium that may affect the range of available tests or way in which the service is provided".

The Staff Survey is included in full as Appendix 11.

The responses suggest that Staff feel that many of the benefits of consortia working including collaboration and working closely together were not strong features or characteristics of how the laboratories work. Notably only 12% feel they are involved in decision making or are able to influence change.

Generally, consortium laboratory staff who responded wanted;

- More consortium working
- The ability to share data, improved LIMS and web portal for test ordering / reporting
- More standardisation across the laboratories
- Rationalisation of services / service model
- Bioinformatics strategy / infrastructure
- A strategy / development planning for genomics

10. PHG Foundation: International models of service delivery for laboratory genomics

One aspect of the review is to understand models of service delivery for laboratory genetics and genomics utilised elsewhere in Europe, particularly in countries with similar demographic characteristics to Scotland. The evidence set out in the PHG Foundation Report will contribute to the wider review and help inform decision making on the future service configuration for genomics in Scotland[3].

The report provides an update on recent developments in the service delivery of laboratory genomics in the other UK nations – England, Wales and Northern Ireland – and three European countries with similar demographic characteristics to Scotland. A scoping exercise was carried out to identify suitable countries to include and the following three were chosen:

- Norway (population 5.3 million). Norway has similar demographic considerations to Scotland with an active and varied genomics landscape and clear links between clinical and research services
- Finland (population 5.5 million). Finland has similar demographic considerations to Scotland with an active and varied genomics landscape, with population level genomics initiatives underway and in the planning stages
- Belgium (population 11.5 million). The Belgian population is larger and more concentrated than in Scotland, however there are both clinical and research national genomics initiatives, and a wide range of services available across the population from different laboratories

For each European country, a literature review was carried out on the delivery of genomics services and key contacts were identified and invited for interview. Information in the public domain not written in English was also accessed.

The report identifies 6 cross-cutting themes;

1. Oversight of genomics and genomics policy – Each of the three countries have seen little central oversight in relation to genomics to date. National level genomics schemes are under development in the three countries, the nature of the oversight could vary, from government policy to collaborative working groups that help guide service development.
2. Genomic data – A more national / coordinated approach is being taken to genomic data, there is an interest in ensuring synergy and cooperation between clinical services and academia, and biobanks will have an important role to play in the wider data landscape
3. Laboratory development – Mostly local with little/no top down strategy. There is a lack of standardisation in terms of processes, procedures, techniques and interpretation but good innovation.
4. Test development and provision – Local developments can result in different trajectories for testing, creating challenges in terms of aligning services and optimising delivery.
5. WGS and WES – There is variation in terms of developments in WGS and WES, and for which indications, trends in each of the countries are moving towards wider use of both.
6. Pharmacogenomics (PGx) – Approaches to and availability of PGx testing varies, with DPYD being the most common test carried out. Planning is for the integration of PGx test information into electronic health record systems to support further future implementation

UK nations have followed their own approaches to developing laboratory genomics infrastructure and service delivery, on the whole these have benefited from national coordination and the commissioning of services. This is a major difference between the UK nations and the European countries that have been investigated in the report.

[3] The full report is available upon request from nss.specialistservices@nhs.scot

11. Research, Development & Innovation

All laboratories participate in translational research and collaborate with university departments and commercial partners on R&D projects and trials which is an important part of an expanding service. Examples of current projects include;

- Scottish Genomes project (all consortium centres),
- Pharmacogenomics – collaboration with university (Tayside),
- Development of a RET fusion panel for non-small cell lung cancer (Grampian),
- Bionano for detection of copy number variation (Lothian Cytogenetics),
- Precision Panc – NGS for pancreatic patients (GGC).

In terms of collaborative working and equipment, GGC, Lothian and Tayside currently access NovaSeqs shared with the University. In Glasgow the equipment is located within the diagnostic laboratory and managed by them. In Lothian the equipment is based within the university department. As services develop, particularly with respect to methods such as WGS, increased capacity will be required and sharing resources with universities may no longer be practical. In addition, sharing of equipment has implications for accreditation of tests, particularly if not under the control of the service laboratory and without careful planning and co-ordination accreditation may not be granted

It is very important that laboratories participate in R&D but in cases where projects are translational and can impact on the diagnostic service, there needs to be a forum for discussion at an early stage. This can prevent duplication of effort if others are evaluating similar developments, assist with validation by sharing of samples and to facilitate evaluation for the service moving forward.

12. Demand Optimisation & Realistic Medicine

Key Driver for Change:

To work with stakeholders to develop improvement plans to reduce unnecessary or inappropriate testing. This in turn will free up capacity to address rising demand and deliver testing that positively affects the patient pathway.

12.1 Demand Optimisation

Demand Optimisation is defined as the process by which diagnostic test use is optimised to maximise appropriate testing, which in turn optimises clinical care and drives more efficient use of a scarce resource[4].

The key areas to consider are:

- Minimising over / under-requesting, which can be damaging to patient care
- Reducing unnecessary repeat requests (e.g. through introduction of gateway permissions)
- Ensuring appropriate and useful tests are equitably accessible

[4] National Demand Optimisation Group; https://www.demandoptimisation.scot.nhs.uk/

- Standardisation of nomenclature/test coding to reduce unnecessary variation and allow automated data monitoring systems to extract laboratory test usage information in an efficient, consistent and timely manner
- Internal standardisation of laboratory practice – to ensure the optimal processes, procedures and testing protocols are in place and adhered to.

Once activity across all genomics work-streams is clearly defined and understood a technology assessment can be undertaken to drive efficient delivery of testing services. This will in turn improve turnaround times for timely issue of reports to inform patient care, reduce unwarranted duplication of effort across testing services and ensure efficient use of laboratory resource including staff time

Current deficiencies / areas for improvement;

- Inconsistent reporting / consistent method of reporting is required across the laboratories
- Data management and standards / the laboratories need to develop improved and consistent data management standards
- Current workload calculation method (GENU's) is no longer fit for purpose / a consistent way to demonstrate activity and workload is required

Consortium wide demand optimisation has not been undertaken and needs done urgently in order to inform a new model to improve the delivery of Genomics for Scotland.

12.2 Realistic Medicine

It is the NHS' vision that by 2025[5] anyone providing healthcare in Scotland will take a realistic medicine approach. This approach will ensure there is a focus on shared decision making, reducing harm, reducing waste and tackling unwarranted variation. Consequently, it is essential that realistic medicine is central to the strategy development and implementation.

Furthermore, it is imperative that outcome measures are identified and used to measure performance and delivery of safe, equitable, person centred and optimised service going forward. Using an outcomes based benefits realisation approach will ensure we can review and adapt as circumstances dictate in an agile and proactive way. This will be essential for resilience, and sustainability going forward.

By instilling the realistic medicine approach into our strategy and developing, measuring and reviewing outcomes data we can:

- Plan for the future
- Improve the health and wellbeing of the people of Scotland, whilst reducing health inequalities
- Deliver best value using our resources

[5] Chief Medical Officer's Annual Report 2015-16: REALISING REALISTIC MEDICINE, First published by The Scottish Government, February 2017 ISBN: 978-1-78652-673-1, https://www.gov.scot/publications/chief-medical-officer-scotland-annual-report-2015-16-realising-realistic-9781786526731/ accessed 02-02-2022

13. Review Findings

13.1. Findings Against NSSC Designation Criteria

This review considered the Genetics & Molecular Pathology Laboratory Service against the NSSC criteria for national designation.

Table 15: Review Findings Against NSSC Designation Criteria

#	NSSC Criteria	Comment
1	The clinical need for national commissioning of the service is significant and is within a clearly defined clinical area.	The majority of respondents to the user survey (180/233 or 77%) considered they are currently able to access a range of tests to support clinical decision making. Also see comments under criteria 2, 3 and 8 below.
2	There is a clear target patient group or subset distinct for clinical reasons.	The Consortium Test Directories specify which genomic tests are commissioned and the patients who will be eligible to access to a test / eligibility criteria.
3	The service is for a condition requiring diagnosis and/or treatment that is rare and/or unpredictable and has a low incidence. (Usually no more than 500 patients in one-year period).	While the service does not strictly meet low incidence criteria, it is highly skilled requiring specialist equipment and facilities. The laboratories are also co-located with Clinical genetics centers and other areas of clinical expertise. The service has benefited from national coordination and would best advance with greater strategic oversight.
4	The service has a proven evidence base and will have a greater clinical benefit than alternative forms of care.	The applications of genomics to deliver personalised medicine are numerous and are growing, the evidence base and impact of each intervention would require analysis. The service could improve though the delivery of health economic evaluation of testing pathways to assess impact on care.
5	The service is person centred demonstrating a clear clinical pathway which will include criteria for referral, discharge and follow up care.	Genomic medicine has the capacity to revolutionise the healthcare of an individual with a rare disease or cancer by offering prompt and accurate diagnosis, risk stratification based upon genotype and the capacity for personalised treatments.
6	The service can demonstrate/has an explicit plan to provide the service equitably to all patients who are eligible for NHS treatment in Scotland.	Where services are fully funded via NSD or Scottish Government investment access to testing is equitable available for all eligible patients. However, where the service has not been granted funding (e.g. thought the progression of business cases) services are not equitably provided.

		The testing service is not comparable to that available elsewhere in the UK e.g. availability / access to WGS as part of standard care, cancer tests prioritised for implementation, NIPD.
7	Provision requires at least one of the following: - a highly skilled multidisciplinary team - scarce clinical skills - specialist equipment and facilities	Service delivery is dependent of highly skilled healthcare and / or clinical scientists to analyse and report genomic data to delivery actionable patient reports. Laboratory staff are also frequently involved in MDTs to discuss individual cases or to advance the provision of service.
8	There will be significant benefits from national commissioning: demonstrating improved clinical quality, focused clinical expertise, more efficient use of NHS resources.	The laboratory genomics service delivery, on the whole these have benefited from national coordination and the commissioning of services but would benefit from more integrated consortia working and a clear service wide strategy to optimize delivery
9	There is evidence to support the cost of the service to determine that it will be cost effective that can only be provided clinically and cost effectively in one or two locations.	The service is provided across four locations and provides testing on a regional or specialist basis High level analysis of consortia budget to actual spend taken from laboratory annual reports demonstrates an increasing adverse variance. In spite of annual incremental increases or uplift on budgets, to accommodate rising costs (e.g. for NHS staff pay), the variance has continued to grow. Non-recurring funding has been provided to cover most of the variance, however longer term solutions are required.
10	There are statements of support for the service.	The majority of respondents to the user survey (180/233 or 77%) considered they are currently able to access a range of tests to support clinical decision making. The laboratory staff surveyed showed over half of the 74 respondents just over half thought that the consortium delivers equitable access to testing for clinical users / patients. However, both surveys identified a numbers of areas for improvement.

13.2. Findings Against Laboratory Service Review Criteria

In relation to the review terms of reference and defined aims and objective consideration was also given to additional criteria of specific relevance to the future advancement of genomic laboratory services and model/s of delivery;

Table 16: Review Findings Against Laboratory Service Review Criteria

#	Review Criteria	Review Conclusions
1	Current and predicted future need for the service	Samples being referred to the consortia have been steadily increasing for a number of years.

		It is predicted that demand for cancer testing to facilitate the delivery of precision medicine will increase considerably in the coming years The consortium has identified 12 cancer testing pathways that are prioritised for implementation, in line with current guidelines and are as standard care elsewhere in the UK.
2	Clinical quality standards / adherence to best practice and any regulatory requirements (i.e. UKAS)	Accreditation successfully maintained across all sites.
3	Patient outcomes are comparable with other UK and International laboratories	The testing service is not comparable to that available elsewhere in the UK e.g.; availability / access to WGS as part of standard care, cancer tests prioritised for implementation, NIPD.
4	The service achieved Financial Balance	High level analysis of consortia budget to actual spend taken from laboratory annual reports demonstrates an increasing adverse variance. In spite of annual incremental increases or uplift on budgets, to accommodate rising costs (e.g. for NHS staff pay), the variance has continued to grow. Non-recurring funding has been provided to cover most of the variance, however longer term solutions are required.
5	Service continually Horizon Scans and develops to meet the needs of the Scottish health care system	It is very important that laboratories participate in research and development but in cases where projects are translational and can impact on the diagnostic service, there needs to be a forum for discussion at an early stage. This can prevent duplication of effort if others are evaluating similar developments, assist with validation by sharing of samples and to facilitate evaluation for the service moving forward.
6	Continually aims to meets customer and staff satisfaction	The majority of respondents to the user survey (180/233 or 77%) considered they are currently able to access a range of tests to support clinical decision making. The laboratory staff surveyed showed over half of the 74 respondents just over half thought that the consortium delivers equitable access to testing for clinical users / patients. However, both surveys identified a numbers of areas for improvement.
7	Service efficiency and effectiveness and cost effectiveness	Lack of standardisation of data collection and reporting and inability to share data between the Consortium laboratories gives rise to inefficiencies. There is also a need for improved ordering of tests and reporting of results directly into the clinical systems to improve service efficiency. There are varying levels of IT / eHealth support across the consortium laboratories, on occasions the lack of IT support is a major barrier to efficiency developments. A new optimised service delivery model needs to be developed and implemented to ensure resilience, long term suitability/viability and adaptability to meet current and future demands

		Unable to measure the amount of tests done for constitutional (hereditary/germline) genetic disease as laboratories currently unable to measure this workload. Given the size of the service there is a requirement to maintain a regional model however further efficiencies and optimisation of services will be delivered by realigning testing within the regional model. Rationalisation and streamlining of germline testing and molecular pathology services is required.
8	Sustainability of the current service	The current model is not sustainable
9	Future developments within the services	The laboratories have all indicated a need to expand next generation sequencing services which requires substantial investment. There is no consortium wide strategic planning for equipment procurement. The laboratories engage with procurement at board level, although they can all draw on national frameworks where available and invest in equipment to suit local workflows. Negotiations at a local level means that the consortium may be missing out on discounts for multiple pieces of the same equipment. Furthermore, an audit of the service revealed that there are significant differences in maintenance contracts for seemingly identical pieces of equipment across the sites. Samples being referred to the consortia have been steadily increasing for a number of years. While currently more than half of the samples received by the service are for rare and inherited disease testing (excluding inherited cancer), it is predicted that demand for cancer testing service to facilitate the delivery of precision medicine will increase considerable in the coming years.
10	Current issues faced by the service and how they are being addressed	Consortium laboratories have highlighted workforce as one of the biggest challenges to sustainability and resilience of the service. The ability to undertake training and development, staff retention and service development were also flagged as challenging.
12	How the service should develop over the next five years taking into account future developments	An implementation plan to take forward review recommendation / next steps as detailed will facilitate service advancement over the coming years

After evaluation of the evidence examined as part of the review, it can be concluded that the delivery of the Scottish genomic laboratory services has benefited from some national coordination through the consortia model and being centrally commissioned.

However, going forward there needs the be greater integration and standardization to achieve an equitable service that is sustainable and can accommodate future growth. This will require a clear strategy to inform the implementation of a new service model that ensures optimal delivery of genomic testing.

14. Conclusions

The review has considered the current service model, the existing capability/capacity, levels of activity and workload across all four Laboratories that comprise the consortia. Furthermore, the review has considered performance against the designation criteria and delivery of the recommendations from the last review in 2017. Key processes, data sharing, storage and reporting, the volume and variety of different tests, infrastructure, workforce and financial sustainability were reviewed as part of the process.

Recommendations to optimise the service and build a sustainable, resilient service underpinned by the principles of realistic medicine are detailed in the next section.

Key finding include;

1. The review has identified that optimising service delivery is key for the ongoing sustainability and resilience of the service and to facilitate effective absorption of emerging testing and technologies. Whilst streamlining of germline testing has been attempted further work is required to realise greater efficiency. Similarly streamlining of molecular pathology services for a variety of testing pathways is now essential. The review recognises that to reduce duplication these require a focus going forward.

2. The current basis for calculating workload is generally considered no longer fit for purpose and the use of separate Laboratory Information Management System (LIMS) raises concerns at a number of levels about the quality and integrity of data that is being reported.

3. Prioritisation should be given to achieving standardisation of procedures, test requesting, data sharing, collection and reporting to improve benchmarking and ensure optimal service delivery. The laboratories would benefit from a common data environment / software to automate data interrogation and reporting.

4. Improved test ordering / reporting of results directly into the clinical systems and advancing a centralised genomics data repository that supports data sharing across sites would enable the laboratories to distribute work, improve genomics analytical capabilities and help absorb increasing demand to accommodate new testing / pathways.

5. The majority of the laboratories are not appropriately accommodated within physical infrastructures that can support service advancement nor increase. With the exception of GGC, space has significant capacity implications for the future development of services to meet predicted need over the next 5-10 years. The laboratories will require support from host boards to modernise to assure equipment, facility & interface requirements for optimal delivery. Integration of services, especially in Lothian, would increase efficient use of resource and economies of scale and provide resilience.

6. Greater integration and consortium working were highlighted by user and staff surveys as desirable. The Laboratories effectively operate on a standalone basis within a consortia model. Many of the potential benefits of a consortia model including management processes, collaboration and driving improvements are not optimised at a consortia level. Technical excellence and performance at individual Laboratories will not compensate for how the consortia model is currently organised and managed.

7. The publication of the Laboratory Test Directory is a significant development. The Directory provides the basis to promote the service, understand the type and range of tests and a baseline to understand the dynamic within the consortia and how services could be optimised and improved. The laboratories should now prioritise identifying tests/methodologies that are no longer required and where testing services can be consolidated to allow phased removal as new testing practices are implemented.

8. The capacity of larger NGS platforms is beyond the needs of each laboratory site and lends itself to reconfiguration of services. Networking of work, particularly “wet work”, would alleviate the need to duplicate equipment and also make better use of technical staff by operating a centralised facility. Horizon scanning mechanisms need to be initiated to identify and assess the viability of new testing/sequencing technologies.

9. Staffing levels impact on capacity to increase the volume of testing and to facilitate service advancement and require consideration as part of a strategic approach. A workforce planning exercise should be undertaken across the laboratories to ensure optimum skill mix and deployment / use of staff resource.

10. Resilience planning should include training and development opportunities for the laboratory workforce and, where appropriate, attrition. The required level for bioinformatics support needs to be identified and implemented to deliver safe & timely clinically actionable solutions for patients.

11. In spite of annual incremental uplift on budgets to accommodate rising costs the actual vs funding variance has continued to grow. The adverse variance is currently being covered by bridge funding from Scottish Government which is non-recurrent, therefore action to optimise service delivery and realise efficiencies (demand optimisation) is essential to ensure ongoing financial sustainability of the service.

12. The service should seek to future proof to accommodate service expansion, diversification and innovations. Currently there is no clear basis to plan for future requirements nor how existing and new scientific and medical roles will be resourced in future in the context of changing testing technology and the significant forecast increase in demand based on different measures and characteristics of the service.

13. The transition of whole genome sequencing (WGS) and the scale implementation of Pharmacogenomics from research to routine clinical service within NHS Scotland now needs to be planned and prioritisation given to implementing the 12 priority cancer testing pathways. To provide the focus required to deliver the cancer testing pathways the laboratories would benefit from a dedicated resource to enable delivery at pace.

14. There needs to be a robust framework for improved transparency of research collaborations & findings to facilitate translation of proven research into service delivery for NHSS standards of care. There should be greater engagement / education around the benefits of appropriate genomics to deliver precision care.

The Review concludes that there needs to be a clear strategy and new service model that delivers Process, Organisational and Technology improvements with more appropriate financial and resource planning to address known issues whilst ensuring the principles of realistic medicine are adhered too.

The strategy should prioritise emerging testing requirements/technology and highlight opportunities to optimise, consolidate and improve services and provide a roadmap to address the challenges around the significant increase in demand for existing and future services.

15. Recommendations

The review recommendations are presented in terms of short, medium and long term deliverables that are recommended to be progressed for optimal service development and delivery going forwards.

Recommended deliverables have been divided into categories around *process*, *organisation* and *technology* required to facilitate service change;

- ✓ Processes to manage change and measure achievement / improvement
- ✓ Organisation required to realise outcomes
- ✓ Technology required to support innovation

It should be noted that the recommended deliverables and desired outcomes will need to be incorporated / developed into an action plan to facilitate implementation.

Table 17: Review Recommendation Deliverables in terms of Process, Organisation & Technology

Key:	
Short	Short term – 6 months
Med	Medium term – 6-12 months
Long	Long term – 12 months +
	Commence & Complete
	Initiate / Plan
Note: Timeframe is reference to point of review report sign-off, anticipated March 2022	

Process

Priority	Deliverable	Short	Med	Long	Desired Outcomes
1	Data definitions, standardisation, collation & reporting				• Nationalise SOP's, equipment, technical manual
					• Nationalise referral process, documentation, reporting templates
					• Standardisation & consistency of approach irrespective of lab location for same/similar tests
					• Better communication & marketing of the service
					• Appropriate benchmarking & outcome measures to ensure benefits realisation
1	Undertake Demand Optimisation and introduce Forecasting and Planning process				• Optimise service and delivery • Increase capacity through efficient use of resource • Ability to scale-up & absorb new pathways
1	Cancer pathways options appraisal and SWOT				• Informed & evidence based decisions on service delivery • Stakeholder buy-in

	(Strengths, weaknesses, opportunities & threats) analysis				• Cost-benefit analysis to help inform strategy
1	Develop strategy				• Establish National Strategic Network
					• Implementation of priority cancer testing pathways at pace (appoint transformation team to support)
					• Approach that can manage both high and low volumes of activity
					• Better management & performance of the Consortia
					• Streamline testing services & efficient use of resources
2	Develop & Implement a new Service Delivery Model				• Agree National Model for service delivery across Scotland
					• Consolidation of tests to fewer laboratories
					• Appropriate test requesting and gateway controls supported by e-forms/software
					• Centralised service desk for processing referrals/reporting to optimise service delivery
					• Centralisation of ‘wet work’ and logistics to reduce turnaround times
					• Identify methodologies no longer required to allow phased removal as new testing practices implemented (stop non-value add practices)
1	Genomics mapping				• Identify methodologies no longer required to allow phased removal as new testing practices implemented (stop non-value add practices)
					• Assess/identify viability of new testing/sequencing technologies;
					• Agreed testing strategy in relation to projected sample numbers for optimal delivery
					• Identify equipment, facility & interface requirements for optimal delivery
					• Identify Genomics capabilities Scotland wide including diagnostic services across NHSS and academic institutions
3	Plan for WGS				• Understand clinical need and projected demand • Conduct options appraisal of possible delivery models

					• Transition from research to routine clinical service
3	Plan for scale implementation - Pharmacogenomics				Understand clinical need and projected demand
2	Horizon Scanning				• Improved transparency with research collaborations & findings
					• Improved mechanisms for engagement with the Scottish Medicines Consortium (SMC) around companion testing required to enable patients to access SMC approved treatments
					• Formalised mechanisms for engagement with industry and academic colleagues
					• Future proofing to accommodate service expansion / diversification / innovations

Organisation

Priority	Deliverable	Short	Med	Long	Desired Outcomes
1	Workforce Planning & development				• Optimum skill mix to ensure best deployment/use of resource
					• Attract, manage & retain resource
					• Data driven decisions
					• Resilience plan that includes for training, development and attrition
1	Bioinformatics Support				• Identify and implement level of support delivery of safe & timely clinically actionable solutions for patients
1	Integration of Genomics into Routine Care				• Informed clinical workforce that understand how and where genomic testing fits into a clinical pathway and how to use
					• Publication of Scottish test directories to raise clinical community awareness
					• Engagement & education around the benefits of appropriate genomics to deliver precision care

Technology

Priority	Deliverable	Short	Med	Long	Desired Outcomes
1					• Ability to store & share data securely

	Data Repository (Software agnostic)				• Interaction with local Laboratory Information Systems (LIMS)
3	Common data environment / software				• Automated processes for the interrogation and reporting of data sets by single sites & consortia