https://doi.org/10.4038/sljma.v26i2.5587

The Sri Lanka Journal of Medical Administration

https://sljma.sljol.info/ Original Article Volume 26 Issue 02

From Capacity to Action: Charting National Genomic Sequencing Services Roadmap for Precision Health in Sri Lanka

Singhaprathapa SWMKK[1], Dolamulla SS[1], Dharmatilake NH[1], Hewamanna SSK[2]

[1]Ministry of Health and Mass Media, Colombo, Sri Lanka, [2]Regional Directorate of Health Services, Monaragala.

Correspondence to:
SWMKK Singhaprathapa

E-Mail
dr.swmkks@gmail.com

Key Words:

Genomic sequencing, roadmap, Sri Lanka, precision health, capacity building

Abstract

Introduction: Genomic sequencing (GS) is a powerful tool in modern medicine underpinning disease surveillance, precision diagnostics, antimicrobial resistance monitoring, and public health research. Sri Lanka has developed laboratory and research infrastructure, but lacks a unified national roadmap for GS services. This study aimed to assess GS capacity in Sri Lanka, identify key challenges, and prioritise strategies to guide roadmap development.

Methods: A cross-sectional descriptive study was performed in 2025 among professionals, including medical administrators (25%), consultants (30%), academics (20%) and laboratory technologists (25%). Data were collected via a validated self-administered questionnaire (Cronbach's $\alpha = 0.87$) and a resource mapping tool. Quantitative results were summarised using descriptive statistics; qualitative data were thematically synthesised. Proposed strategic actions were ranked using a Delphi-based scoring framework.

Results: Significant gaps emerged: infrastructure inadequacies (N=80; 68%), funding shortages (N=80; 72%), limited bioinformatics capacity (N=80; 25%), and weak inter-institutional coordination (N=80; 64%). Resource mapping revealed that while equipment was available in many institutions, 78% reported a critical shortage of trained personnel. The highest-priority strategies (mean scores out of 5) were: establishing national governance (4.8), workforce training (4.6), enhancing data sharing (4.5), integrating GS into health policy (4.3), strengthening ethical or legal frameworks (4.1) and developing sustainable funding (3.9).

Conclusions and Recommendations: Although Sri Lanka has foundational infrastructure for GS, critical gaps in human resources, governance, and financing constrain its utility. A coordinated roadmap addressing governance, capacity building, data integration, and sustainability is urgently needed to harness GS for health, research, and national health security.

Introduction

Background

Genomic sequencing is the process of determining the complete DNA or RNA sequence of an organism's genome. It reveals genetic information, including mutations, variations, and pathogen identity. This technology supports diagnostics, disease surveillance, research, and precision medicine by enabling targeted interventions and understanding of genetic and infectious disease patterns. GS is increasingly central to biomedical science and public health. It enables pathogen surveillance, genomic epidemiology, AMR tracking, precision oncology, and pharmacogenomics[5,14].

In global health emergencies such as the COVID-19 pandemic, countries with established GS systems gained a distinct advantage in real-time variant detection and evidence-based public health decision-making[3,16]. In Sri Lanka, molecular diagnostics and limited next-generation sequencing (NGS) platforms exist in select research institutions and tertiary hospitals[6,17]. However, these are isolated efforts, often dependent on external funding or academic projects, and lack integration into national health systems or public health policy[18]. Without a strategic framework, GS remains underutilised in disease surveillance, diagnostics, and health planning[5,18]. Many LMICs face similar challenges, such as limited workforce, weak data systems and fragmented governance[14]. Nonetheless, successful national genomic initiatives, such as the Africa CDC Pathogen Genomics Initiative[3,16] and Thailand's National Genomics Infrastructure[4,15], show that governance, workforce training, sustainable financing, and ethical data systems are the key pillars of progress. Given Sri Lanka's evolving health research ecosystem and increasing emphasis on genomic resilience, a situational assessment is timely. This study assessed GS capacity, identified gaps and opportunities. Furthermore, the study prioritised the strategies to guide the development of a national GS roadmap for Sri Lanka.

Objectives

General Objective

To assess the current status and available resources to develop a strategic framework for genomic sequencing services in Sri Lanka.

Specific Objectives

- To evaluate the current status of genomic sequencing services in Sri Lanka, including infrastructure, technical capacity, and policy environment.
- To identify available resources and gaps critical to developing a strategic framework for national genomic sequencing services.
- To analyse key factors influencing the implementation and scalability of genomic sequencing in the context of precision health.
- To provide recommendations for establishing a comprehensive roadmap to enhance genomic sequencing capabilities for precision health initiatives in Sri Lanka.

Literature Review

Genomic Sequencing has emerged as a transformative technology in public health, enabling precision medicine, surveillance of infectious diseases, and genomic epidemiology[1]. The World Health Organization has emphasised the integration of GS into national health systems to strengthen disease surveillance, outbreak response, and antimicrobial resistance monitoring[2]. WHO's Global Genomic Surveillance Strategy (2022–2032) outlines the need for country-level capacity building, governance mechanisms, and prioritisation frameworks for effective implementation[3].

Several countries, including the United Kingdom, Australia, and India, have successfully developed national genomic strategies focusing on infrastructure development, skilled workforce training, and data governance[4–6]. These frameworks provide valuable models for low- and middle-income countries aiming to establish sustainable genomic programs.

Prioritisation tools have been used in many settings to identify strategic interventions in public health genomics, allowing systematic assessment of feasibility, impact, and readiness[7]. Tools such as the WHO's health technology prioritisation frameworks and resource mapping instruments assist policymakers in aligning genomic investments with national priorities[8].

In Sri Lanka, genomic sequencing capacity remains limited, and systematic assessments of readiness and prioritisation have not been comprehensively undertaken. Thus, understanding global best practices and applying prioritisation approaches are essential for developing a national GS roadmap.

Methods

Study design

A descriptive cross-sectional mixed-method study design was conducted from January 2025 to July 2025. Reporting adheres to the STROBE guidelines for cross-sectional studies[1,2,7,8]. Checklist items related to design, participants, variables, bias, study size, and analysis were systematically applied to ensure transparency[11,12].

Study setting

The study was conducted at national, regional, university, and hospital institutions engaged in laboratory services, public health, or research[17,18] in Sri Lanka.

Study population

The study population comprised senior-level professionals in medical administration, clinical practice, academics, and laboratory services who were directly involved in GS-related governance, research, or diagnostic activities. Participants ($n = 80$) were purposively selected to represent four categories: Medical Administrators (25%), Consultants in laboratories (30%), Academics (20%), and Laboratory staff, including Medical Laboratory Technologists (25%).

Study instruments

The study employed three main instruments: an online Google Form, a Resource Mapping Tool, and a Prioritisation Tool.

A Self-Administered Questionnaire (SAQ)

The online Google Form was used as the primary data collection instrument to obtain information from key stakeholders across public, private, and academic institutions. It consisted of structured and semi-structured questions designed to capture data on institutional capacity, workforce expertise, laboratory facilities, and existing GS activities. SAQ was developed based on literature and expert input[5,14,15,20]. It covered infrastructure, funding, workforce, bioinformatics, governance, policy environment, and collaboration. The pretest among 10 professionals achieved Cronbach's $\alpha = 0.87$, indicating high internal consistency.

The form utilised a five-point Likert scale to capture respondents' perceptions and levels of agreement with various statements. The scale consisted of 1 – Strongly Agree, 2 – Agree, 3 – Neutral, 4 – Disagree, and 5 – Strongly Disagree. This format enabled the study to measure the degree of consensus or divergence among stakeholders regarding existing capacities, challenges, and opportunities in the GS landscape. Additionally, open-ended questions were included to allow respondents to provide detailed explanations, examples, and recommendations. The Google Form was distributed electronically to ensure broad participation across institutions and to facilitate convenient, time-efficient data collection while maintaining confidentiality and data integrity.

A Resource Mapping Tool (RMT)

The resource mapping tool was improved and tailored to the Sri Lankan context. It helps systematically document and analyse available infrastructure, human resources, laboratory capabilities, and data management systems relevant to GS. This tool assists in identifying the geographic and institutional distribution of resources and highlights existing gaps and overlaps.

A Prioritization Tool

This tool was designed to guide stakeholders in ranking strategic actions for the development of the national GS roadmap. Using predefined criteria such as feasibility, impact, and sustainability, it enabled the systematic evaluation and prioritisation of proposed interventions through a participatory process. This instrument ensured comprehensive data collection, objective assessment, and evidence-based decision-making for roadmap formulation.

A Semi-Structured Questionnaire (SSQ)

A SSQ was developed to guide key informant interviews with experts and stakeholders engaged in genomic services in Sri Lanka. The tool was designed to obtain in-depth qualitative information on existing capacities, challenges, and opportunities related to genomic sequencing implementation. It allowed for flexibility in exploring emerging themes while ensuring consistency across interviews. Data were collected through direct interviews, either in person or online. They were analysed thematically to identify key insights and perspectives relevant to strengthening national genomic sequencing capacity.

Data collection and analysis

Data were collected through online Google Forms and Mentimeter software. Selected participants participated in key informant interviews to validate responses and enrich qualitative findings. Quantitative data were analysed using descriptive statistics; qualitative data were coded thematically. A Delphi-based scoring framework was utilised to prioritise proposed strategies[9,10,13].

Results

Participant profile

All 80 participants completed the survey. The response rate was 100%. The sample included medical administrators (20; 25%), consultants (24; 30%), academics (16; 20%), and laboratory technologists (20; 25%). Participants represented central, provincial, and institutional levels, ensuring comprehensive national coverage across diverse healthcare and academic settings.

Human resources availability (Table 1)

Human resource distribution across genomic sequencing institutions demonstrated variable capacity. The National Hospital of Galle (NHG) and National Hospital Kandy (NHK) had similar profiles with one consultant, one trained consultant, two medical officers, and several MLTs, indicating moderate operational capacity. The Medical Research Institute (MRI) had more consultants and a research officer, reflecting stronger research involvement. The National Cancer Institute (NCI)

Table 1. Availability of human resources for genomic sequencing services in Sri Lanka

Institution	Consultants using the Genomics	Trained Consultants	Medical Officers	Trained Medical Officers	Research Officers	Trained Research Officers	Bioinformatician	Trained Bioinformatician	MLTs	Trained MLTs
NHG	1	1	2	0	0	0	0	0	5	2
NHK	1	1	2	0	0	0	0	0	7	2
MRI	2	1	1	0	1	0	0	0	2	1
NCI	6	3	0	0	0	0	1	1	3	3
NSACP	2	1	1	0	0	0	0	0	3	3

had the highest consultant representation and the only trained bioinformatician, highlighting advanced analytical capability. The National STD/AIDS Control Programme (NSACP) showed balanced staffing with trained MLTs. Overall, trained personnel were limited, emphasising the need for workforce development
Most respondents (68%) rated infrastructure as inadequate. Only select universities and national laboratories had functional NGS platforms, with 61% having at least one sequencer. However, underutilisation was common due to reagent shortages, maintenance gaps, or downtime[17,18,20].

Availability of equipment and tests at institutes (Table 2)
Oxford Nanopore Technologies (ONT) and Illumina are currently the leading platforms for genomic sequencing, each employing distinct technologies with complementary strengths.
ONT devices, such as the Mk1C, utilise nanopore-based sequencing, enabling real-time, portable, and long-read analysis, which is particularly useful for field diagnostics and rapid outbreak investigations. Illumina platforms, including NextSeq and iSeq, use sequencing-by- synthesis technology to generate highly accurate short reads, making them ideal for large-scale genomic studies, clinical diagnostics, and transcriptomic analyses. In Sri Lanka, institutions such as the University of Colombo (UoC) and the University of Sri Jayewardenepura (USJP) have integrated these technologies into their genomic research

Table 2. Availability of equipment and tests for genomic sequencing services in Sri Lanka

Institution	Brand	Model	Funding	Responsibility of Reagent Procurement	Year of Installation	Service Agreement
NHG	ONT	Mk1C	Donation	No	2021	No
NHK	ONT	Mk1C	Donation	No	2021	No
MRI	ONT	Mk1C	Donation	No	2022	No
NCI	Illumina	NextSeq 1000	Donation	Yes	2022	Yes
NCI	Illumina	iSeq 100	Donation	Yes	2022	Yes
NSACP	Illumina	NextSeq 1000	Donation	Yes	2022	Yes
UoC	ONT, Illumina	-	-	-	-	-
USJP	ONT, Illumina	-	-	-	-	-

infrastructure. UoC has leveraged Illumina systems for high-throughput genomic projects, while USJP has piloted ONT devices for rapid pathogen surveillance and real-time outbreak monitoring. By combining the flexibility, portability, and rapid turnaround of ONT with the high accuracy and throughput of Illumina, these universities are enhancing national capacity in genomic surveillance, precision medicine, and translational research. This dual-platform approach provides a robust framework for strengthening both research and public health responses in Sri Lanka. A large majority (72%) identified funding constraints and dependence on short-term donor projects. Only a few institutions integrated GS into recurrent budgets, reflecting limited sustainability planning[5,18,19].

Availability of Storage and Backups (Table 3)

Assessment of data management systems revealed limited capacity for secure genomic data storage across institutions. NHG, NHK, and MRI relied mainly on computer hard disks without dedicated backup systems or LIMS databases, posing risks for data loss. In contrast, the National Cancer Institute (NCI) maintained more advanced infrastructure, using file servers, cloud storage, and a dedicated backup system. The National STD/AIDS Control Programme (NSACP) also had basic backup capacity through external hard drives. However, none of the institutions had an operational Laboratory Information Management System (LIMS) to link genomic sequences with metadata, indicating a critical need for digital integration.

Table 3. Availability of storage and backups for genomic sequencing services

Institution	Long-term data storage	Data backup storage	Sequence data backup location	Availability of LIMS database
NHG	Computer hard disk	No	None	No
NHK	File server, Computer hard disk	No	None	No
MRI	Computer hard disk	No	None	No
NCI	File server, Computer hard disk, Cloud storage	Yes	File server	No
NSACP	External hard drive, Computer hard disk	Yes	External hard drive	No

Strategic Priority Ranking (Table 4)

The prioritisation exercise identified key strategic areas for strengthening genomic sequencing (GS) in Sri Lanka. Establishing a national GS governance framework ranked highest (mean score = 4.8), emphasising the need for coordination and policy direction. Workforce training and capacity building (4.6) and an integrated data-sharing and bioinformatics hub (4.5) were also highly prioritised. Integration of GS into public health surveillance (4.3) highlighted the importance of linking sequencing to disease monitoring. Developing ethical and legal frameworks (4.1) for genomic data governance and ensuring sustainable funding and partnerships (3.9) were recognised as essential supporting elements for long-term implementation and system resilience.

Weak coordination (64%) and absence of a national governance or ethical framework (81%) were significant gaps. Only 42% reported having institutional data protection policies, and 46% lacked standard procedures for data sharing[18,19].

Table 4. Priority strategies of genomic sequencing services in Sri Lanka

#	Strategy	Mean Score / 5 (n=80)
1	National GS governance framework	4.8
2	Workforce training or capacity building	4.6
3	Integrated data-sharing and bioinformatics hub	4.5
4	Integration into public health surveillance	4.3
5	Ethical or legal frameworks for genomic data	4.1
6	Sustainable funding and partnerships	3.9

Discussion

Summary of findings

This study presents the first structured national assessment of GS readiness in Sri Lanka, providing a comprehensive overview of the current landscape across health, academic, and research institutions. It identified critical gaps in governance, workforce capacity, technical expertise, data management, and sustainable financing, despite the presence of isolated equipment and limited operational initiatives[6,17]. The findings underscore the urgent need for a coordinated national roadmap that aligns policy, infrastructure, and human resource development while fostering collaboration between health, academic, and research sectors to ensure effective and equitable integration of GS into Sri Lanka's public health system[5,18].

Comparison with other LMICs

Similar capacity constraints have been reported in other LMICs[14], highlighting common challenges in infrastructure, technical expertise, and sustained funding. The Africa CDC Pathogen Genomics Initiative exemplifies how regional hubs can effectively pool resources, build workforce capacity, and harmonise operational standards across countries[3,16]. Likewise, Thailand's national genomic centre model demonstrates the successful integration of sequencing technologies into disease surveillance and public health response networks[4,15].

Governance and policy implications

Effective governance is foundational to developing a sustainable genomic ecosystem. Establishing a National Genomic Steering Committee under the Ministry of Health could provide strategic oversight for regulation, ethical compliance, data sharing, and intersectoral coordination[18,19]. Such a mechanism would promote accountability and policy coherence. Furthermore, adherence to the STROBE framework ensures scientific rigour, transparency, and robust translation of observational data into evidence-informed policies and national genomic strategies[1,2,7,8,11].

Workforce capacity and retention

The severe shortage of trained genomic experts highlights the urgent need for structured postgraduate training programs, continuous professional development, and attractive retention incentives to sustain national capacity. Establishing regional mentorship networks and implementing twinning arrangements with established international genomic hubs, as recommended in global genomics implementation models, can accelerate skill transfer, strengthen institutional capacity, and foster a sustainable workforce pipeline for genomic medicine and public health applications[14,16,17].

1*Data infrastructure and ethics*

Establishing a national bioinformatics hub would enable centralised management of sequencing analysis, data storage, and controlled access, thereby improving efficiency, data security, and institutional governance[18,19,20]. Such a hub would also facilitate standardisation of analytical pipelines and promote equitable data sharing across sectors. Ethical oversight mechanisms should be embedded within this framework, aligned with OECD and WHO genomic data protection and privacy standards to ensure responsible data use[19,20].

Funding and sustainability

Reliance on donor funding is inherently unsustainable for long-term genomic system development. Integrating GS activities into national health budgets, fostering strategic private sector partnerships, and establishing a dedicated national genomics innovation fund would ensure financial continuity and resilience[5,15,18]. Moreover, implementing demonstration projects that showcase the cost-effectiveness and public health impact of GS applications such as AMR surveillance can build confidence and attract sustained stakeholder investment[3,14].

Phased implementation strategy

1. Establish governance and pilot coordination hub.
2. Build human resource capacity and regional sequencing units.
3. IntegrateGS into surveillance, diagnostics, and research networks.
4. Scale nationally with evaluation and sustainability measures[17,18,20].

Limitations

As a purposive sample, the study's representativeness is inherently limited, and the reliance on self-reported data may have introduced response or recall bias. However, the methodological rigour maintained throughout the research process, particularly adherence to the STROBE framework, systematic data verification and triangulation of information sources enhances the credibility, reliability, and overall validity of the findings, providing a robust foundation for policy and programmatic interpretation[1,2,7].

Conclusions

Sri Lanka currently possesses partial infrastructure and research interest in genomic sequencing, but critical gaps in workforce, governance, and funding prevent its integration into health systems. A national GS roadmap founded on governance architecture, capacity building, data integration, and sustainable financing is urgently required. The prioritised strategies generated in this study provide a practical, stakeholder-informed foundation. Implementing these will strengthen diagnostic, surveillance, and research capabilities in Sri Lanka.

Recommendations

Based on the conclusions, the following recommendations are proposed

1. Establish a national governance and regulatory framework for genomic sequencing services in Sri Lanka.
2. Develop and implement a phased national genomic sequencing roadmap aligned with health system priorities in Sri Lanka.
3. Strengthen human resource capacity, with emphasis on bioinformatics and clinical genomics in Sri Lanka.
4. Improve bioinformatics infrastructure and data integration within national health information systems in Sri Lanka.
5. Ensure sustainable financing mechanisms to support long-term genomic sequencing services in Sri Lanka.

References

1. Von Elm E, Altman DG, Egger M, Pocock SJ, Gøtzsche PC, Vandenbroucke JP; STROBE Initiative. The Strengthening the Reporting of Observational Studies in Epidemiology (STROBE) statement: guidelines for reporting observational studies. J Clin Epidemiol. 2008 Apr;61(4):344-9.
2. Vandenbroucke JP, von Elm E, Altman DG, Gøtzsche PC, Mulrow CD, Pocock SJ, Poole C, Schlesselman JJ, Egger M. Strengthening the Reporting of Observational Studies in Epidemiology (STROBE): Explanation and Elaboration. PLoS Med. 2007 Oct;4(10):e297.
3. Africa Centres for Disease Control and Prevention. Pathogen Genomics Initiative: Strengthening Public Health Genomics Across Africa. Addis Ababa; 2022.
4. Thailand Centre of Excellence for Life Sciences. National Genomics Infrastructure: Strategies and Implementation. Bangkok, 2023.
5. WHO. Global strategy for genomics and health: Harnessing genomic technologies to improve public health worldwide. Geneva: World Health Organization; 2023.
6. Jayaweera K, Ranasinghe C, Jayasooriya S, et al. The Sri Lankan Twin Registry biobank: South Asia’s first extensively phenotyped and genotyped cohort. BMC Genet. 2020;21:142.
7. Cuschieri S. The STROBE guidelines. Saudi J Anaesth. 2019;13(Suppl 1):S31-S34.
8. Abeysena C. Strengthening the reporting of observational studies in epidemiology (STROBE) statement: New guidelines for reporting observational studies. J Coll Community Physicians Sri Lanka. 2008;13(2):26–28.
9. Chan AW, Tetzlaff JM, Altman DG, et al. SPIRIT 2013 Statement: defining standard protocol items for clinical trials. Ann Intern Med. 2013;158(3):200-207.
10. Moher D, Liberati A, Tetzlaff J, Altman DG; PRISMA Group. Preferred Reporting Items for Systematic Reviews and Meta-Analyses: the PRISMA statement. PLoS Med. 2009;6(7): e1000097.
11. Benchimol EI, Smeeth L, Guttmann A, et al. The RECORD Statement: Extension of the STROBE Statement for studies using routinely collected health data. PLoS Med. 2015;12(10):e1001885.
12. Bossuyt PM, Reitsma JB, Bruns DE, et al. STARD 2015: an updated list of essential items for reporting diagnostic accuracy studies. BMJ. 2015;351:h5527.
13. Collins R, Bowman L, Landray M, Peto R. The magic of randomization versus the myth of real-world evidence. N Engl J Med. 2020;382(7):674-678.
14. Choudhary A, Srivastava S. Implementing genomics in developing countries: challenges and pathways. Front Public Health. 2023;11:1154329.
15. Tan DS, Koh WP. Precision medicine in Asia: Opportunities and policy considerations.Lancet Region Health West Pac. 2020;4:100043.
16. Githinji G, Mburu S, Miring’u G, et al. Building capacity for genomic surveillance in Africa. Nat Commun. 2022;13:484.
17. Singhaprathapa SWMKK, Dolamulla SS, Dharmatilake NH, Hewamanna SSK. Situational Analysis of Genomic Sequencing Capacity in Sri Lanka (Unpublished internal MoH report) 2025.
18. National Science Foundation, Sri Lanka. Strategic Directions for Biotechnology and Genomics Research in Sri Lanka 2023–2030. Colombo, 2023.
19. OECD. Guidelines on the Protection and Use of Human Genomic Data. Paris, 2023.
20. World Health Organization. Laboratory Quality Management System: Handbook. 3rd ed. Geneva; 2011.