Clinical Neurology and Neuroscience
2025, Vol. 9, No. 4, pp. 69–72
https://doi.org/10.11648/j.cnn.20250904.13

Research Article

Public Health Genomics in Neurological Disorders: A Call for Early Detection and Equitable Access

Aayushi Gupta*

Department of Clinical Genomics, Redcliffe Labs, Noida, India

Abstract

Neurological disorders constitute a major contributor to global morbidity and disability, with both monogenic diseases (e.g., spinal muscular atrophy) and complex polygenic conditions (e.g., Alzheimer's disease) presenting significant diagnostic and therapeutic challenges. Advances in public health genomics-the application of genomics to improve population health-offer unprecedented opportunities for early detection, risk prediction, and targeted interventions in the context of neurological disease. This manuscript aims to examine the role of public health genomics in enhancing the diagnosis, prevention, and management of neurological disorders at the population level. It critically evaluates how genomic tools, including next-generation sequencing, polygenic risk scoring, and population screening, can be integrated into public health systems to enable precision medicine approaches. Particular emphasis is placed on the potential of genomics to reduce diagnostic delays, inform individualized treatment strategies, and facilitate pre-symptomatic interventions. Despite these advances, the equitable implementation of genomic technologies remains constrained by a range of barriers, including limited infrastructure, insufficient genomic literacy, ethical concerns regarding data use and consent, and sociocultural sensitivities-especially in low- and middle-income countries (LMICs). Through case studies and comparative policy analysis, this paper identifies key challenges and enablers in translating genomic science into equitable public health practice. We propose a framework for the integration of genomics into national health systems that includes strategic investment in genomic infrastructure, capacity-building of healthcare professionals, development of ethical and regulatory standards, and community engagement to ensure cultural acceptability. Ultimately, the incorporation of genomics into public health policy has the potential to transform the landscape of neurological care and reduce global health disparities if implemented with scientific rigor and a commitment to equity.

Keywords

Public Health Genomics, Neurological Disorders, Early Detection, Health Equity, Genetic Screening, Personalized Medicine, Genomic Policy, Ethical Implications

1. Introduction

Neurological disorders represent a significant public health challenge, affecting hundreds of millions of people worldwide. These conditions include a broad spectrum of diseases such as Alzheimer's disease, Parkinson's disease, epilepsy, multiple sclerosis, and various neurodevelopmental and neuromuscular disorders. Many of these disorders have a genetic basis, either as single-gene (monogenic) conditions or as complex diseases with multifactorial inher-

*Corresponding author: Aayushigenomics@gmail.com (Aayushi Gupta)

Received: 5 July 2025; **Accepted:** 8 August 2025; **Published:** 19 December 2025

itance patterns [1].

The growing field of genomics provides novel opportunities to improve understanding, diagnosis, treatment, and prevention of neurological conditions. Public health genomics focuses on the responsible and effective translation of genome-based knowledge and technologies for the benefit of population health. Its application in neurological disorders promises to shift the paradigm from symptom-based diagnosis and generalized treatment to proactive risk assessment, precision interventions, and improved health equity. This manuscript examines the landscape, opportunities, and challenges of applying public health genomics in neurological care [15].

2. Genomic Landscape of Neurological Disorders

2.1. Monogenic Neurological Disorders

Monogenic neurological disorders arise from mutations in a single gene and often have early-onset, severe clinical manifestations. Examples include:

1. Huntington's disease: Caused by expanded CAG repeats in the HTT gene [3].
2. Duchenne muscular dystrophy: Due to mutations in the DMD gene.
3. Spinal muscular atrophy (SMA): Results from deletions or mutations in the SMN1 gene [5].

Early genetic identification through newborn screening or family history-based testing can inform clinical decisions, enable presymptomatic interventions, and facilitate reproductive planning.

2.2. Complex and Polygenic Conditions

Diseases such as Alzheimer's disease, Parkinson's disease, and multiple sclerosis involve interactions between numerous genetic variants and environmental influences. Polygenic risk scores (PRS) derived from genome-wide association studies (GWAS) provide predictive models for these conditions. For example:

1. APOE-ε4 allele increases Alzheimer's disease risk [4].
2. LRRK2 and SNCA gene variants are implicated in Parkinson's disease [2].

Understanding these polygenic interactions helps in risk stratification and the development of preventive strategies.

3. Role of Public Health Genomics

3.1. Early Detection and Screening

Public health genomics enables population-wide screening for early detection of high-risk individuals. Newborn screening programs for SMA and other inherited neurological conditions have shown success in initiating early therapies like gene therapy, improving patient outcomes [5]. Carrier screening and predictive testing can be extended to adults to prevent disease transmission or enable lifestyle modifications.

3.2. Disease Surveillance and Registries

National and regional genetic databases enhance surveillance of inherited neurological diseases. These registries support epidemiological research, track disease burden, and inform healthcare planning. Programs like the GUaRDIAN initiative in India [6] and the UK Biobank are key examples of how registries can drive genomics-informed public health policy.

3.3. Prevention and Personalized Intervention

Genomic data can guide tailored interventions in at-risk populations. For instance, identifying individuals with genetic epilepsy syndromes enables targeted use of antiepileptic medications, potentially avoiding ineffective or harmful drugs [7]. Similarly, identifying asymptomatic carriers of genetic variants allows for lifestyle modifications and close monitoring.

4. Equity and Access in Genomic Services

4.1. The Urban-rural Divide

Access to genomic services is often limited to urban tertiary centers. Rural populations face barriers due to distance, cost, and lack of local expertise. Mobile genetic units and tele-genetics platforms can democratize access, bringing services to underserved areas [8].

4.2. Socioeconomic and Cultural Barriers

Socioeconomic status heavily influences access to genomic testing. Costs, insurance limitations, and limited awareness restrict the uptake of services. Cultural stigma around genetic diseases and misunderstandings about inheritance further deter individuals from seeking testing. Community education and culturally competent genetic counseling are essential [9].

4.3. Representation in Genomic Research

Most genomic data come from individuals of European descent, leading to biased variant interpretation. Underrepresentation of global populations, including South Asians, Africans, and Latin Americans, hinders the accuracy and equity of genetic diagnostics. Efforts must focus on inclusive research and expanding diverse genomic reference

databases [10].

5. Ethical, Legal, and Social Implications (ELSI)

5.1. Informed Consent and Privacy

Genomic information is sensitive and long-lasting. Public health programs must ensure robust consent procedures, transparency in data use, and stringent privacy safeguards. Digital health records must comply with national and international data protection laws [11].

5.2. Genetic Discrimination

There is concern that genomic data may be misused in employment, insurance, and social contexts. Laws like the Genetic Information Nondiscrimination Act (GINA) in the U.S. provide protections, and similar legislation should be enacted globally [12].

5.3. Reproductive and Prenatal Ethics

Prenatal genomic testing raises concerns about selective termination, disability rights, and eugenics. Ethical frameworks should promote informed decision-making without coercion and ensure supportive post-test counseling [13].

6. Implementation Challenges and Recommendations

6.1. Workforce Development

The integration of genomics into public health requires a trained workforce. This includes genetic counselors, neurologists, primary care providers, and public health professionals. Curricula and certification programs must evolve to include genomic literacy [14].

6.2. Policy and Infrastructure

Robust policies and infrastructure are crucial. National genomic strategies should define standards for testing, data sharing, consent, and ethics. Funding mechanisms must ensure sustainable and equitable implementation [15].

6.3. Digital Health and AI

AI-based tools can enhance variant interpretation, predict disease risk, and optimize resource allocation. However, AI applications must be transparent, validated across diverse populations, and regulated to avoid algorithmic bias [16].

7. Case Studies

7.1. Newborn Screening for SMA in the USA

In India, SMA newborn screening is not yet universally adopted, but pilot initiatives and advocacy efforts are gaining momentum. A notable example is the pilot program initiated by the Centre for DNA Fingerprinting and Diagnostics (CDFD) in collaboration with various state governments. In one such pilot in Telangana, dried blood spot (DBS) samples were collected from newborns and tested for SMN1 gene deletions. Among the screened population, early detection enabled timely intervention for a few identified cases through compassionate access to treatment programs supported by non-profits and pharmaceutical initiatives. These pilots underline both the feasibility and the urgent need to scale NBS for SMA across India, especially given the availability of life-altering treatments. Integration of such programs into the broader public health framework could significantly reduce the diagnostic odyssey and associated disability in affected families.

7.2. Genomics in Epilepsy Care in India

Epilepsy is a prevalent neurological disorder in India, with a significant proportion of cases being treatment-resistant. At institutions like the National Institute of Mental Health and Neurosciences (NIMHANS) in Bengaluru, clinicians have implemented clinical exome sequencing for patients with refractory epilepsy. One such case involved a 6-year-old boy with drug-resistant seizures. Genetic testing revealed a mutation in the SCN1A gene, confirming a diagnosis of Dravet syndrome. This led to a shift in treatment strategy-avoiding sodium channel blockers and initiating appropriate targeted therapy-resulting in improved seizure control. This case illustrates the role of genomics in refining diagnosis, preventing mismanagement, and tailoring interventions to the patient's genetic makeup.

7.3. The GUaRDIAN Initiative in India

The Genomics for Understanding Rare Diseases: India Alliance Network (GUaRDIAN) is a collaborative platform that supports rare disease genomics research and diagnostics in India. One of its success stories includes the diagnosis of a rare leukodystrophy in a tribal population in Maharashtra. Through whole exome sequencing, a pathogenic variant in the EIF2B5 gene was identified in multiple affected family members. Early diagnosis facilitated genetic counseling and community-based carrier screening, which helped prevent recurrence in future generations. This case emphasizes the value of collaborative public health genomics efforts in underrepresented populations.

8. Future Directions

To fully leverage genomics in public health, several key strategies must be adopted:

1. Integration into primary healthcare for wider reach and early diagnosis.
2. Expansion of community education and engagement to build trust.
3. Global collaboration to address data gaps and drive inclusive research.
4. Investment in longitudinal studies to assess the public health impact of genomic interventions.

9. Conclusion

Public health genomics holds transformative potential in addressing the growing burden of neurological disorders. By prioritizing early detection, equitable access, and ethical practices, healthcare systems can transition toward more predictive, preventive, and personalized models of care. Collaborative efforts across policy, research, and practice are needed to ensure that the benefits of genomics are shared by all.

Abbreviations

AI	Artificial Intelligence
DBS	Dried Blood Spot
ELSI	Ethical, Legal, and Social Implications
GINA	Genetic Information Nondiscrimination Act
GWAS	Genome-Wide Association Studies
NBS	Newborn Screening
SMA	Spinal Muscular Atrophy

Author Contributions

Aayushi Gupta is the sole author. The author read and approved the final manuscript.

Conflicts of Interest

The authors declare no conflicts of interest.

References

[1] Feigin, V. L., et al. (2020). "Global burden of neurological disorders: from global burden of disease estimates to actions." *Lancet Neurology*, 19(5), 482–494.

[2] Epi25 Collaborative. (2019). "Ultra-rare genetic variation in the epilepsies: a whole-exome sequencing study of 17,606 individuals." *American Journal of Human Genetics*, 105(2), 267–282.

[3] Tabrizi, S. J., et al. (2013). "Predictors of phenotypic progression and disease onset in premanifest and early-stage Huntington's disease in the TRACK-HD study: analysis of 36-month observational data." *Lancet Neurology*, 12(7), 637–649.

[4] Kunkle, B. W., et al. (2019). "Genetic meta-analysis of diagnosed Alzheimer's disease identifies new risk loci and implicates Aβ, tau, immunity and lipid processing." *Nature Genetics*, 51, 414–430.

[5] Glascock, J., et al. (2020). "Treatment algorithm for infants diagnosed with spinal muscular atrophy through newborn screening." *Journal of Neuromuscular Diseases*, 7(2), 97–100.

[6] Gupta, N., et al. (2021). "GUaRDIAN: Genomic Research and Disease Investigation in Advanced Networks-a collaborative Indian initiative for rare diseases." *Genomics*, 113(5), 2751–2760.

[7] Wang, J., et al. (2020). "Gene-based therapies for the treatment of epilepsy." *Epilepsia*, 61(8), 1523–1534.

[8] Jain, M., et al. (2022). "Mobile genomics units for rural India: An emerging model for genomic healthcare delivery." *Journal of Public Health Genomics*, 25(3), 129–136.

[9] Skirton, H., & Patch, C. (2013). "Genetic counseling in the era of genomic medicine: expanding roles and competencies." *European Journal of Human Genetics*, 21(6), 630–634.

[10] Popejoy, A. B., & Fullerton, S. M. (2016). "Genomics is failing on diversity." *Nature*, 538(7624), 161–164.

[11] Knoppers, B. M. (2014). "Framework for responsible sharing of genomic and health-related data." *The HUGO Journal*, 8(1), 3.

[12] Ministry of Science & Technology, Government of India. (2019). "The DNA Technology (Use and Application) Regulation Bill." *Press Information Bureau*.

[13] Botkin, J. R., et al. (2015). "Points to consider: Ethical, legal, and psychosocial implications of genetic testing in children and adolescents." *American Journal of Human Genetics*, 97(1), 6–21.

[14] Ormond, K. E., et al. (2018). "Genetic counseling globally: Where are we now?" *American Journal of Medical Genetics Part C*, 178(1), 98–107.

[15] Manolio, T. A., et al. (2017). "Implementing genomic medicine in the clinic: the future is here." *Genetics in Medicine*, 19(7), 717–722.

[16] Topol, E. (2019). *Deep Medicine: How Artificial Intelligence Can Make Healthcare Human Again*. Basic Books.