

# Developing Patient Genomic Data Principles: Enabling Patients to Share and Receive Data Confidently

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## Abstract

Genomic data have become a valuable resource for accelerating scientific discoveries, improving personalized medicine, and addressing global health challenges. As the volume and complexity of genomic data grow exponentially, the need for robust data sharing policies becomes increasingly paramount. Developing well-crafted genomic data sharing policies is essential to strike a delicate balance between fostering scientific progress and protecting individual privacy, ethical considerations, and the responsible use of sensitive information. However, with the vast potential of genomic data sharing comes a host of ethical and privacy issues. Genetic information contains unique and identifiable characteristics, raising concerns about the inadvertent re-identification of individuals and the potential misuse of data. Ensuring participants' informed consent, safeguarding data privacy, and preventing unauthorized access to sensitive information are critical aspects that demand careful consideration when formulating data sharing policies. Our goal is to provide a framework for ethical genomic data sharing principles, highlight any gap in current practices, and assist Illumina in navigating the current policy landscape.

## Background

Illumina is an American biotechnology company and the world's leading manufacturer in integrated systems and products for large-scale analysis of genetic variation and function. With collaborations in over 140 countries, the company aims to improve global health equity by advancing personalized medicine and genomics research.

## Methods

- Scientific Literature Databases**
  - Duke Libraries
  - PubMed
  - Google Scholar
- Policy Briefs**
  - International genomic data sharing policies
  - State data sharing initiatives
  - Genomic repositories standards
- Business Documentation**
  - Illumina reports
  - Relevant news releases
  - Peer companies' privacy policies

## Results

### Glossary of Key Genomic Data Sharing Terms

A glossary of key genomic terms to ensure shared understanding of genomic concepts among different stakeholders and facilitate true informed consent is provided here.



### Recommendations on Safeguarding Patient Genomic Data Security and Privacy

Completely “de-identifying” genomic data might not be possible because genomes contain unique markers of identity. Discussion of gaps in data handling and opportunities for improvement is provided here.



### Landscape Analysis of Patient Genomic Data Sharing Policies and Initiatives

A catalog of policies and initiatives and a landscape analysis outlining trends and differences are provided here.

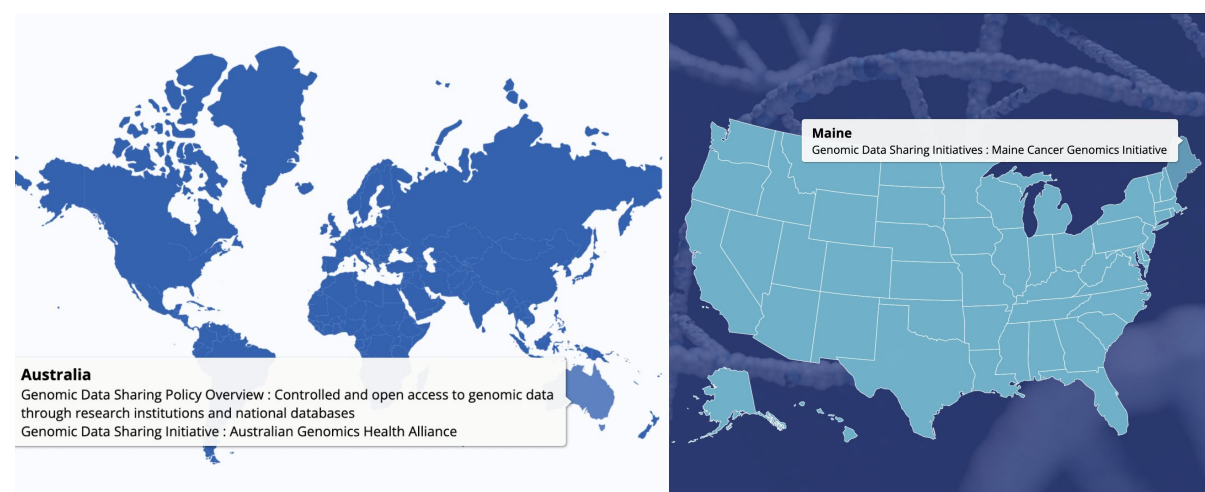


Figure 1. United States State-to-State Genomic Data Initiative Map

Figure 2. International Country-to-Country Genomic Data Initiative Map



Trend Analysis



United States Map



International Map

### Community Engagement Guidelines in Effective Genomic Data Sharing

Guidelines, case studies, and actionable plans for implementing community genomic healthcare programs among underrepresented communities are provided here.



## Results, cont.

### Design of a Creative Informed Consent Approach using Digital Application

Proposal of a multimedia app/website for patients to complete the informed consent form, communicate with healthcare providers, and learn about genomics is provided here.



Figure 3. Design of Informed Consent Proposal

## Conclusion

Given that genomic sequencing and related technologies are constantly changing and increasing in their reach on an international scale, it is important to consider the limitations within our own work and acknowledge that these observations and recommendations may need to be updated as new advancements occur. We found a lack of diversity in both genomic databases and policymaking, both personal and sociopolitical friction surrounding genomic data-sharing policies, and the lack of true informed consent as major issues throughout the field of genomics.

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