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DNA, Genomics and Precision Medicine in Italy: Population Diversity, Biobanking, Clinical Translation and Ethical Governance

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Abstract

DNA and genomics are becoming increasingly important to biomedical research, precision medicine, rare-disease diagnosis, cancer research, pharmacogenomics and population health in Italy. Italy has distinctive scientific and biological resources arising from substantial geographic and population diversity, a distributed network of research hospitals and universities, and an established national biobanking infrastructure. This narrative review examines the development and strategic role of DNA-based research in Italy, focusing on population genetics, biobanking, genomic medicine, rare diseases, cancer, pharmacogenomics, data governance and ethical considerations. Italian population-genomic studies demonstrate measurable genetic structure within the country, including differentiation involving Sardinia and gradients across the peninsula. At the infrastructure level, BBMRI.it connects Italian biobanks and biological-resource centres and supports access to samples including DNA together with clinical and research data. Italy is also participating in the European Genome of Europe initiative, which aims to build a European reference genome reflecting genetic and ancestral diversity. Recent Italian national strategy work has proposed strategic lines covering clinical implementation, specialized genomic centres, national genomic data infrastructure, professional training and public education. At the same time, genetic information presents distinctive privacy, consent, discrimination and governance challenges. The Italian data-protection framework imposes safeguards for processing genetic data and biological samples for scientific research. The review concludes that Italy's future DNA and genomics capacity will depend on interoperability, responsible data governance, equitable clinical translation, workforce development, high-quality biobanking and meaningful public and patient participation.

Keywords: DNA; genomics; Italy; population genetics; biobanking; precision medicine; rare diseases; cancer genomics; pharmacogenomics; genetic data governance

1. Introduction

DNA research has moved from a largely laboratory-based discipline toward an increasingly integrated component of healthcare, biomedical research and public health. High-throughput sequencing, whole-exome sequencing (WES), whole-genome sequencing (WGS), genotyping and molecular diagnostics have expanded the ability to identify disease-associated variants, characterize populations and support individualized clinical decisions.

Italy is an important setting for DNA and genomics research because its population contains substantial geographic and historical diversity. Genome-wide analysis of 1,014 Italians found measurable genetic differentiation within Italy and showed that Sardinians could be clearly differentiated from mainland Italy and Sicily (Sanna et al., 2012). Such diversity is valuable for understanding population history and improving representation in genomic reference resources.

Italy also has an extensive biobanking and biomedical research ecosystem. BBMRI.it, the Italian national node of BBMRI-ERIC, connects universities, hospitals, research institutes and patient organizations and supports access to biological samples and associated clinical and research data, including DNA (BBMRI-ERIC, 2026; BBMRI.it, 2026).

A multi-institutional Italian initiative conducted from 2021 to 2024 developed a technical basis for a future Italian National Genomic Strategy, covering clinical implementation, specialized genomic centres, national genomic data infrastructure, professional training, public education and ethical-legal-social issues (Italian National Genomic Strategy Initiative, 2025).

2. Conceptual Framework

2.1 DNA as biological information

DNA contains hereditary information and can be analyzed at multiple levels, from targeted variants to whole genomes. Modern genomic medicine combines genomic information with clinical phenotypes, family history, imaging, laboratory findings and environmental information. The value of DNA data therefore depends not only on sequencing technology but also on interpretation and clinical context.

2.2 Population genomics

Population genomics studies genetic variation across geographic, demographic and historical groups. Italy is particularly relevant because geographic barriers and historical population structure have generated detectable regional patterns. Carefully characterized diversity can improve research into rare variants, disease susceptibility and demographic history.

2.3 Translational genomics

Translational genomics refers to the movement of genomic discoveries from laboratory research into diagnostics, prevention, treatment selection and health-system practice. This pathway requires validated assays, clinical interpretation, trained professionals, sustainable financing, interoperable information systems and ethical governance.

3. Methodology

This manuscript uses a narrative review of peer-reviewed literature, official Italian institutional sources and European research-infrastructure materials. The review focuses on population diversity, biobanking, clinical

genomics, rare diseases, cancer, pharmacogenomics, data governance and research infrastructure. It does not report original sequencing, patient recruitment or laboratory experiments.

4. Genetic Diversity of the Italian Population

Sanna et al. (2012) analyzed 125,799 genome-wide SNPs in 1,014 Italians with broad geographic coverage. Their analysis identified geographic structure and showed that the Sardinian population was clearly differentiated from mainland Italy and Sicily, while additional genetic differentiation could be detected within the Italian peninsula.

These findings have implications for genomic research design. If population structure is ignored, genetic association studies may be affected by population stratification. Conversely, carefully characterized regional diversity can improve research into rare variants and disease-associated alleles.

The European Genome of Europe project, launched in 2024, aims to establish a European reference genome reflecting genetic and ancestral diversity across countries and minorities. Italian participation includes Human Technopole, ELIXIR Italy and Centro Cardiologico Monzino IRCCS, according to the CNR (European Commission, 2024; CNR, 2024).

Dimension	Italian relevance	Research implication
Regional genetic structure	Sardinia and other regions show distinctive genetic patterns.	Population structure should be considered in genomic analyses.
European diversity	Italy contributes to European reference-genome efforts.	Italian samples can improve reference representation.
Rare variants	Population isolates can be informative for rare-variant discovery.	Useful for disease-gene research.
Clinical genomics	Genomic interpretation requires population-appropriate references.	Variant databases and reference panels matter.
Public health	Population information can support risk studies.	Avoid deterministic interpretations of genetic risk.

5. DNA Biobanking and Research Infrastructure

Biobanks connect biological samples to clinical, epidemiological and research information. BBMRI.it was established through collaboration involving the Italian Ministry of Health and Ministry of University and Research and participates in BBMRI-ERIC. Its resources include blood, tissues, cells and DNA with associated information (BBMRI-ERIC, 2026).

The scientific value of biobanking depends on sample quality, standardized processing, accurate metadata, secure information systems, ethical governance, discoverability and appropriate access procedures. BBMRI.it's current infrastructure also emphasizes quality, information technology and ethical-legal-social services (BBMRI.it, 2026).

6. Biobanking Governance and Data Quality

Component	Key requirement	Why it matters
Sample quality	Standardized collection, processing and storage	Reduces analytical variability.
Metadata	Accurate clinical and epidemiological annotation	Improves interpretation.
DNA integrity	Validated extraction and storage	Supports sequencing.
Consent	Clear and appropriately scoped consent	Supports ethical and lawful reuse.
Data security	Pseudonymization and access controls	Reduces privacy risk.
Interoperability	Common data standards	Facilitates multicentre research.
Access governance	Transparent scientific and ethical review	Balances research and protection.
Return of results	Defined procedures for relevant findings	Addresses participant interests.

7. Genomic Medicine and the Italian National Genomic Strategy

An Italian national initiative involving 14 research institutions and universities operated from December 2021 to June 2024 and developed a technical document for a future Italian National Genomic Strategy. Twelve National Mirror Groups addressed governance, ethical-legal-social issues, clinical data standards, sequencing practice, interoperability, health economics, private-sector involvement and use cases including rare diseases, cancer, common complex diseases, populations, precision prevention, pharmacogenomics and infectious diseases (Italian National Genomic Strategy Initiative, 2025).

Strategic area	Main objective	Potential benefit
Clinical implementation	Integrate validated genomic tests into care	More precise diagnosis and treatment.
Genomic centres	Develop specialist expertise	Better interpretation.
Data infrastructure	Connect genomic and clinical data securely	Research and clinical integration.
Professional training	Train clinicians and genomic specialists	Improved appropriate use.
Public education	Increase genomic literacy	Informed participation.
ELSI governance	Address privacy, consent and access	Public trust.

8. Rare Diseases and DNA-Based Diagnosis

Rare diseases are a major application of DNA sequencing. WES and WGS can identify variants that remain undetected by targeted testing. A 2024 study of 33 Italian patients with suspected autosomal-recessive inherited retinal disease used WGS after previous testing had not established a diagnosis and identified elusive variants, including variants in non-coding regions and structural variants (Bertelli et al., 2024).

The clinical challenge is interpretation: detecting a variant does not automatically establish causality. Findings require evaluation against phenotype, inheritance, population frequency, functional evidence and clinical databases. Genetic counseling is therefore an important component of responsible genomic medicine.

9. Cancer Genomics

Cancer genomics examines DNA changes in tumors and, in some cases, inherited variants that affect cancer susceptibility. Molecular characterization can support tumor classification, therapeutic targeting and treatment selection. Italy's network of IRCCS institutions, university hospitals and oncology biobanks provides a strong foundation for this research.

The distinction between somatic and germline information is important. Somatic variants arise in tumors and can guide treatment, whereas germline variants may have implications for the patient and biological relatives. This distinction creates different requirements for consent, counseling, privacy and communication.

10. Pharmacogenomics and Personalized Medicine

Pharmacogenomics investigates how genetic variation influences drug response. Italy's emerging national genomic strategy explicitly includes pharmacogenomics among its use cases (Italian National Genomic Strategy Initiative, 2025). Potential applications include identifying patients at increased risk of adverse drug reactions, predicting treatment response and supporting individualized dosing.

Clinical adoption requires evidence that a genetic test changes management in a way that improves outcomes. Pharmacogenomic information should therefore be integrated with medication history, age, organ function, comorbidities and other clinical factors.

11. DNA, Precision Prevention and Population Health

Genomic information may contribute to precision prevention by identifying inherited risk for selected conditions and enabling earlier surveillance or preventive interventions. The Italian national strategy initiative includes common complex diseases, populations and precision prevention among its genomic use cases (Italian National Genomic Strategy Initiative, 2025). Genetic risk is generally probabilistic and interacts with environment, behavior, socioeconomic conditions and healthcare access.

12. Ethical, Legal and Social Issues

DNA is uniquely sensitive because genetic information can reveal information about an individual and, indirectly, biological relatives. The Italian Data Protection Authority has established specific safeguards for genetic data. Italian provisions address scientific and statistical research, biological samples, consent, security, storage and communication of results (Garante per la protezione dei dati personali, 2016; 2018).

The Garante's guidance emphasizes defined scientific purposes and appropriate safeguards for genetic data and biological samples used in research. It also addresses conditions under which previously collected samples and genetic data may be retained or reused for research, subject to applicable legal and ethical requirements (Garante per la protezione dei dati personali, 2018).

13. Privacy, Consent and Data Governance

Issue	Core question	Governance response
Informed consent	What uses of DNA and data are covered?	Use clear, appropriately scoped consent.
Secondary research	Can samples/data be reused?	Apply legal, ethical and

		governance requirements.
Privacy	Who can identify participants?	Use pseudonymization and secure access.
Family implications	Could findings affect relatives?	Provide appropriate counseling.
Incidental findings	Should unexpected relevant results be returned?	Define policies before analysis.
International sharing	Can data cross borders?	Use lawful transfer mechanisms.
Commercial use	Will samples support commercial research?	Ensure transparency and authorization.
Public trust	Do communities understand research?	Engage patients and the public.

14. European Integration and Genome of Europe

Genome of Europe began in 2024 as a major European initiative involving institutions across all 27 EU member states. Its objective is to establish a European reference genome representing genetic and ancestral diversity, including minorities (European Commission, 2024).

The CNR reports Italian participation by Human Technopole, ELIXIR Italy and Centro Cardiologico Monzino IRCCS. The project is intended to support future genetic discoveries, prevention and personalized healthcare and to facilitate integration with the European Health Data Space (CNR, 2024).

European integration creates governance challenges because genomic data may move among institutions and jurisdictions. Interoperability must therefore be accompanied by common standards, security and clear access governance.

15. Digital Infrastructure and Artificial Intelligence

Modern genomics generates large volumes of sequence and phenotype data. The usefulness of these data depends on secure storage, analysis, annotation and controlled sharing. Italy's proposed genomic strategy identifies national genomic data infrastructure and interoperability as strategic areas (Italian National Genomic Strategy Initiative, 2025).

Artificial intelligence may support variant interpretation, phenotype matching and genomic prediction. However, models can reproduce biases present in training data. Validation across Italian regional populations and clinically relevant patient groups is therefore important.

16. Workforce and Education

Genomic medicine requires multidisciplinary expertise involving clinical geneticists, laboratory scientists, bioinformaticians, pathologists, oncologists, pharmacists, genetic counselors, data scientists, nurses, ethicists and health-policy specialists.

Professional education should include technical genomics, interpretation, communication of uncertainty, informed consent, data protection and ethical decision-making. Public genomic literacy is equally important because individuals increasingly encounter genetic information outside traditional clinical systems.

17. Challenges to DNA and Genomics Development in Italy

Challenge	Potential consequence	Suggested response
Regional variation in expertise	Unequal access to advanced genomic services	Develop referral networks and shared standards.
Data fragmentation	Difficulty linking genomic and clinical data	Strengthen interoperability.
Interpretation complexity	Variants of uncertain significance	Use validated pipelines and multidisciplinary review.
Privacy risk	Loss of trust or unauthorized disclosure	Strengthen security and access governance.
Workforce shortages	Delayed interpretation	Expand specialist training.
Funding sustainability	Research may not translate into services	Develop long-term financing and health-economic evaluation.
Public misunderstanding	Overestimation of genetic determinism	Improve genomic literacy.
Cross-border research	Complex data-transfer requirements	Use harmonized European frameworks.

18. Strategic Framework for Italy

Pillar	Priority actions	Expected contribution
Population genomics	Represent Italian regional diversity	Better reference datasets.
Biobanking	Standardize samples and metadata	Reliable research resources.
Clinical genomics	Expand validated testing	Earlier and more accurate diagnosis.
Rare diseases	Use WES/WGS in appropriate pathways	Higher diagnostic yield.
Cancer	Link molecular profiling with oncology	More individualized treatment.
Pharmacogenomics	Evaluate actionable variants	Safer medication use.
Data infrastructure	Develop interoperable genomic-clinical systems	Secure integration.
Ethics and privacy	Strengthen consent and security	Public trust.
Education	Train genomic professionals and inform public	Responsible adoption.
European collaboration	Connect Italy to GoE and BBMRI-ERIC	Larger research resources.

19. Discussion

Italy's DNA and genomics landscape combines scientific opportunity with significant governance complexity. Population diversity offers a research resource, while biobanks provide biological and clinical materials for translational research. At the same time, national genomic infrastructure requires standards that enable data sharing without weakening privacy and participant protections.

The development of a national genomic strategy is significant because it frames genomics as a health-system transformation. The Italian initiative's strategic areas encompass clinical implementation, infrastructure,

training, public education and governance, indicating that technology alone is insufficient to achieve genomic medicine at scale (Italian National Genomic Strategy Initiative, 2025).

European collaboration further strengthens this opportunity. Genome of Europe can improve representation of European genetic diversity, while BBMRI-ERIC connects biobanks and biomolecular resources. Italy's contribution is consequently both national and European.

The central long-term requirement is responsible translation. DNA sequencing produces information, but clinical value arises when findings are analytically valid, clinically interpretable, communicated appropriately and linked to effective care. Genomics is therefore a multidisciplinary enterprise involving science, medicine, informatics, ethics, law and public engagement.

20. Recommendations

• Strengthen a coordinated Italian national genomic strategy with measurable implementation milestones.

• Expand interoperable genomic data infrastructure while maintaining strong privacy and security controls.

• Support standardized, high-quality biobanking and integration with clinical and epidemiological information.

• Improve access to genomic diagnostics for rare diseases through appropriate referral and sequencing pathways.

• Develop validated genomic applications in oncology and pharmacogenomics based on evidence of clinical utility.

• Strengthen training in clinical genetics, bioinformatics, genomic counseling, data governance and ethical practice.

• Increase public and patient participation in decisions about genomic data use and research.

• Ensure regional genetic diversity is represented appropriately in Italian and European genomic resources.

• Establish clear procedures for clinically relevant and incidental findings.

• Continue Italy's participation in European initiatives such as Genome of Europe and BBMRI-ERIC.

21. Limitations

This manuscript is a narrative review rather than a systematic review or original empirical study. The Italian genomic field is changing rapidly, and institutional programmes, governance frameworks and sequencing technologies may evolve. It therefore provides a strategic synthesis rather than a definitive inventory of every Italian DNA research programme or biobank.

22. Conclusion

DNA and genomics have become important components of Italy's biomedical research and emerging precision-medicine ecosystem. Italy's population diversity provides opportunities for population genetics, while the national biobanking network provides infrastructure for linking biological samples with clinical and research

information. Genomic strategy development, rare-disease sequencing, cancer genomics and pharmacogenomics demonstrate the transition from genomic discovery toward clinical application.

The future success of DNA research in Italy will depend on integration of technology with governance. High-quality sequencing must be accompanied by secure data systems, appropriate consent, ethical oversight, professional training, clinical interpretation and patient engagement. European collaboration through Genome of Europe offers additional opportunities to strengthen reference datasets and genomic research.

Italy is well positioned to contribute to the next phase of genomic medicine, provided innovation is matched by responsible stewardship of genetic information. A sustainable national model should combine scientific excellence, biobanking quality, equitable clinical access, interoperability and strong protection of individual and collective interests.

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