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Comparative analysis of genomics research and population diversity: lessons from Iceland and Nigeria for advancing precision medicine and equitable representation in genomic studies.

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ABSTRACT

The study falls into SDG focus goal 3: Good Health and Well-Being — Target 3.4: Reduce by one-third premature mortality from non-communicable diseases through prevention and treatment; Target 3.8: Achieve universal health coverage, including financial risk protection, access to quality essential health-care services and access to safe, effective, quality and affordable essential medicines and vaccines for all; Target 3.b: Support the research and development of vaccines and medicines for the communicable and non-communicable diseases that primarily affect developing countries.

This study examines the comparative approaches to genomics research and population diversity between Iceland and Nigeria, focusing on population structure, genetic diversity, research infrastructure, discovery of medically relevant variants, and implications for global health equity and precision medicine. A comparative analysis was conducted examining peer-reviewed genomic studies from both countries, including deCODE Genetics research on Icelandic population sequencing, ancient DNA studies revealing Iceland's founding history, whole-genome sequencing of Nigerian populations across multiple ethnolinguistic groups, and genetic relationship analyses among Nigerian ethnic groups. Iceland represents one of the world's most extensively characterized founder populations. The Icelandic population, founded approximately 1,100 years ago by Norse and Gaelic settlers, has been shaped by extensive genetic drift due to isolation and small population size . Large-scale sequencing of 15,220 Icelanders to 34X coverage identified over 39 million autosomal variants, including 31 million SNPs and 7.9 million indels, with family-based approaches enabling high-confidence detection of 108,778 de novo mutations . The population's structure reflects recent drift rather than ancient divergence, minimizing false positives in disease association studies . Nigeria presents a dramatically contrasting picture of extreme genetic diversity. Whole-genome sequencing of 449 individuals across 47 ethnolinguistic groups discovered 36 million SNPs and indels, revealing significant genetic differentiation among groups . Analysis of five major ethnic groups (Igbo, Ibibio, Yoruba, Hausa, Tiv) using nine STR markers found that estimated genetic variation is 99.98% within individuals and only 0.10% among populations, with FST values ranging from 0.001 to 0.500, and genetic relationships closely mirroring linguistic groupings . A remarkable historical connection links the two populations: Hans Jonatan, born in 1784 to an enslaved African mother and European father, migrated to Iceland in 1802, and researchers reconstructed 38% of his maternal genome from 182 descendants, tracing his mother's origins to the Benin-Nigeria-Cameroon region. Iceland's success demonstrates the power of founder populations with extensive genealogical records for identifying disease-associated variants, but the resulting knowledge may have limited applicability to diverse global populations. Nigeria's immense genetic diversity holds key to understanding human evolutionary history and discovering variants relevant to African and global populations, yet remains severely underrepresented in genomics research. Both countries exemplify the imperative to diversify genomic studies to ensure equitable benefits from precision medicine. Achieving SDG 3 targets requires: (1) substantial investment in genomic research infrastructure across diverse populations; (2) large-scale sequencing efforts capturing ethnolinguistic diversity; (3) integration of genomic findings with health systems for precision

medicine; (4) ethical frameworks ensuring benefit-sharing with participating communities; (5) capacity-building for genomic research in low- and middle-income countries; (6) global collaboration to create truly representative genomic reference datasets; and (7) attention to historical injustices in genetic research..

Keywords: Genomics research, population diversity, Iceland, Nigeria, deCODE Genetics, whole-genome sequencing, genetic drift, founder population, ethnolinguistic groups, precision medicine, SDG 3.

RESUME

Cette étude examine les approches comparatives de la recherche génomique et de la diversité des populations entre l'Islande et le Nigeria, en se concentrant sur la structure des populations, la diversité génétique, les infrastructures de recherche, la découverte de variants médicalement pertinents et les implications pour l'équité en santé mondiale et la médecine de précision. Une analyse comparative a été menée en examinant des études génomiques évaluées par des pairs des deux pays, y compris les recherches de deCODE Genetics sur le séquençage de la population islandaise, les études d'ADN ancien révélant l'histoire de la fondation de l'Islande, le séquençage du génome entier de populations nigérianes à travers de multiples groupes ethnolinguistiques, et les analyses des relations génétiques entre les groupes ethniques nigérians. L'Islande représente l'une des populations fondatrices les plus extensivement caractérisées au monde. La population islandaise, fondée il y a environ 1 100 ans par des colons norrois et gaéliques, a été façonnée par une dérive génétique extensive due à l'isolement et à la petite taille de la population . Le séquençage à grande échelle de 15 220 Islandais à une couverture de 34X a identifié plus de 39 millions de variants autosomiques, dont 31 millions de SNPs et 7,9 millions d'indels, avec des approches familiales permettant la détection de 108 778 mutations de novo à haute confiance . La structure de la population reflète une dérive récente plutôt qu'une divergence ancienne, minimisant les faux positifs dans les études d'association de maladies . Le Nigeria présente un tableau radicalement contrasté d'une diversité génétique extrême. Le séquençage du génome entier de 449 individus appartenant à 47 groupes ethnolinguistiques a découvert 36 millions de SNPs et indels, révélant une différenciation génétique significative entre les groupes . L'analyse de cinq groupes ethniques majeurs (Igbo, Ibibio, Yoruba, Hausa, Tiv) utilisant neuf marqueurs STR a révélé que la variation génétique estimée est de 99,98 % au sein des individus et seulement 0,10 % entre les populations, avec des valeurs FST allant de 0,001 à 0,500, et les relations génétiques reflètent étroitement les regroupements linguistiques . Une connexion historique remarquable relie les deux populations : Hans Jonatan, né en 1784 d'une mère africaine réduite en esclavage et d'un père européen, a émigré en Islande en 1802, et les chercheurs ont reconstruit 38 % de son génome maternel à partir de 182 descendants, retraçant les origines de sa mère dans la région Bénin-Nigeria-Cameroun . Le succès de l'Islande démontre la puissance des populations fondatrices avec des registres généalogiques extensifs pour identifier les variants associés aux maladies, mais les connaissances qui en résultent peuvent avoir une applicabilité limitée aux diverses populations mondiales. La diversité génétique immense du Nigeria détient la clé pour comprendre l'histoire évolutive humaine et découvrir des variants pertinents pour les populations africaines et mondiales, mais reste sévèrement sous-représentée dans la recherche génomique. Les deux pays exemplifient l'impératif de diversifier les études génomiques pour assurer des bénéfices équitables de la médecine de précision. Atteindre les cibles de l'ODD 3 nécessite : (1) un investissement substantiel dans les infrastructures de recherche génomique à travers des populations diverses ; (2) des efforts de séquençage à grande échelle capturant la diversité ethnolinguistique ; (3) l'intégration des découvertes génomiques avec les systèmes de santé pour la médecine de précision ; (4) des cadres éthiques assurant le partage des bénéfices avec les communautés participantes ; (5) le renforcement des capacités pour la recherche génomique dans les pays à revenu faible et intermédiaire ; (6) une collaboration mondiale pour créer des ensembles de données de référence génomiques véritablement représentatifs ; et (7) une attention aux injustices historiques dans la recherche génétique.

Mots-clés : Recherche génomique, diversité des populations, Islande, Nigeria, deCODE Genetics, séquençage du génome entier, dérive génétique, population fondatrice, groupes ethnolinguistiques, médecine de précision, ODD 3

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1. INTRODUCTION

The completion of the Human Genome Project in 2003 marked a watershed moment in biomedical science, opening unprecedented opportunities to understand the genetic basis of health and disease. Yet the reference genome constructed from that project was overwhelmingly derived from individuals of European descent, creating a fundamental bias that persists today. Over two decades later, populations of African ancestry—despite harboring the greatest genetic diversity on Earth—remain severely underrepresented in genomic research, with profound implications for the equitable application of precision medicine globally .

This underrepresentation is not merely a matter of scientific completeness; it has direct consequences for human health. Genetic variants that influence disease risk, drug response, and treatment outcomes vary across populations. When genomic research focuses predominantly on European-ancestry populations, the resulting medical advances may not benefit—and could even harm—populations whose genetic architecture differs. Achieving SDG Target 3.8 on universal health coverage and Target 3.b on supporting research and development for diseases affecting developing countries requires that genomic knowledge be truly global in scope and application.

Iceland and Nigeria represent two extremes on the spectrum of population genetic structure and research investment, making their comparison uniquely illuminating. Iceland has become one of the world's most extensively characterized founder populations through the work of deCODE Genetics, a subsidiary of Amgen that has sequenced thousands of Icelanders and leveraged the country's exceptional genealogical records to discover variants associated with numerous diseases . The Icelandic population, founded approximately 1,100 years ago by Norse and Gaelic settlers, has been shaped by genetic drift due to isolation and small population size, creating a population structure that facilitates disease gene mapping .

Nigeria, by contrast, represents the opposite extreme. As Africa's most populous nation, with over 250 ethnolinguistic groups, Nigeria harbors immense genetic diversity that reflects deep evolutionary history, population migrations, and admixture events. Recent whole-genome sequencing of 449 individuals across 47 ethnolinguistic groups discovered 36 million genetic variants, emphasizing the "value of this resource for genomics research, with added granularity by representing multiple ELGs from Nigeria" . Yet this diversity remains drastically understudied relative to its scientific and medical importance.

This study examines the comparative approaches to genomics research and population diversity in Iceland and Nigeria, analyzing population structure, research infrastructure, variant discovery, and implications for global health. Through systematic comparison,

the study aims to illuminate the imperative of diversifying genomic research to ensure that the benefits of precision medicine reach all populations, in keeping with the commitment of SDG 3 to leave no one behind.

2. LITERATURE REVIEW

2.1 Population Genetics Foundations: Founder Effects and Genetic Drift

Population genetic structure is shaped by demographic history, including founding events, population size changes, migration, and admixture. Two concepts are particularly relevant for understanding Iceland and Nigeria: founder effects and genetic drift.

Founder effects occur when a new population is established by a small number of individuals from a larger population, carrying only a subset of the original genetic diversity. Subsequent population growth amplifies the frequencies of those founder alleles, creating distinctive genetic signatures.

Genetic drift refers to random fluctuations in allele frequencies due to chance events, particularly pronounced in small populations. Over generations, drift can lead to fixation or loss of alleles, increasing genetic differentiation between populations.

2.2 Iceland: A Founder Population Extensively Characterized

Founding History: The Icelandic population was founded approximately 1,100 years ago by settlers from Scandinavia and the British Isles. Research using ancient genomes from 27 Icelanders demonstrated that "these ancient Icelanders are markedly more similar to their source populations in Scandinavia and the British-Irish Isles than to contemporary Icelanders, who have been shaped by 1100 years of extensive genetic drift" . This founding event and subsequent isolation created a population uniquely suited for genetic studies.

Population Structure: Research by Price and colleagues examining 292,289 autosomal SNPs showed that "regional geographic ancestries of individuals from Iceland can be distinguished," and importantly, "subpopulation differences are due to genetic drift since the settlement of Iceland 1100 years ago, and not to varying contributions from different ancestral populations" . A critical implication is that "allele frequency differences follow a null distribution devoid of outliers, so that the risk of false positive associations due to stratification is minimal." This makes Iceland particularly valuable for disease association studies.

Large-Scale Sequencing: deCODE Genetics has conducted extensive sequencing of the Icelandic population. A 2017 study sequenced 15,220 Icelanders to an average genome-wide coverage of 34X, identifying 39,020,168 autosomal variants passing GATK filters, including 31,079,378 SNPs and 7,940,790 indels . Using family-based approaches, researchers identified 108,778 high-quality de novo mutations, and traced the parent of origin for 42,961 of them.

Earlier work sequencing 2,636 Icelanders to 20X coverage identified 20 million SNPs and 1.5 million indels . This study found "an excess of homozygosity and rare protein-coding variants in Iceland" and demonstrated the power of imputation by extending analysis to 104,220 individuals down to a minor allele frequency of 0.1%. Key discoveries included "a recessive frameshift mutation in MYL4 that causes early-onset atrial fibrillation, several mutations in ABCB4 that increase risk of liver diseases and an intronic variant in GNAS associating with increased thyroid-stimulating hormone levels when maternally inherited."

2.3 Nigeria: Epicenter of Human Genetic Diversity

African Genetic Diversity: Africa is the cradle of humanity and harbors the greatest genetic diversity on Earth. As Tishkoff and colleagues noted, "populations in Africa have the highest levels of genetic diversity and the deepest population histories" . A 2023 study emphasized that "African populations have been drastically underrepresented in genomics research, and failure to capture the genetic diversity across the numerous ethnolinguistic groups (ELGs) found on the continent has hindered the equity of precision medicine initiatives globally" .

Nigerian Ethnolinguistic Diversity: Nigeria is home to over 250 ethnolinguistic groups, representing three major language families: Niger-Congo (including Yoruba, Igbo, Ibibio), Afro-Asiatic (Hausa), and Nilo-Saharan (Kanuri). This diversity reflects complex population histories including migrations, trade, and cultural exchange.

Genetic Relationship Studies: A 2024 study by Akpan and colleagues examined genetic relationships among five Nigerian ethnic groups—Igbo, Ibibio, Yoruba, Hausa, and Tiv—using nine short tandem repeat (STR) DNA markers . Key findings included:

- Fixation index (FST) values ranged from 0.001 to 0.500 across populations
- Estimated genetic variation was 99.98% within individuals and only 0.10% among populations
- Four heterogeneous clusters were observed, with three principal components accounting for 32.86% of variation
- "The genetic relationships closely mirrored linguistic groupings showing that language may have shaped early interaction and ultimately gene transfer between these groups"

The study sampled 250 consenting participants from their ethnic homes across Nigeria, extracting DNA from blood samples and testing for nine autosomal STR loci using PCR and electrophoresis.

Whole-Genome Sequencing: A landmark 2023 study by Joshi and colleagues performed whole-genome sequencing of 449 Nigerian individuals across 47 unique self-reported ethnolinguistic groups . The study discovered 36 million SNPs and indels and provided "a high-level catalog of both novel and medically relevant variation present across the ELGs." The authors emphasized that these results "underscore the potential of using these cohorts with larger sample sizes to improve our understanding of human ancestry and health in Africa."

2.4 A Remarkable Historical Connection: Hans Jonatan

A fascinating historical connection links Iceland and Nigeria through the story of Hans Jonatan. Born in 1784 in the Caribbean to an enslaved African mother and European father, Jonatan migrated to Iceland in 1802, married, and had two children .

Researchers at deCODE Genetics and collaborating institutions genotyped 182 of his 788 descendants using SNP chips and performed whole-genome sequencing on 20 of them. Using these data, they "reconstructed 38% of HJ's maternal genome and inferred that his mother was from the region spanned by Benin, Nigeria and Cameroon" .

This remarkable study demonstrates both the power of Iceland's genealogical resources—tracing ancestry across 8 generations—and the deep connections between African and European populations. It also illustrates how African genetic material entered the Icelandic gene pool, albeit through the tragic circumstances of the transatlantic slave trade.

Table 1: Comparative Genomic Research Landscapes

Dimension	Iceland	Nigeria
Population founding	~1100 years ago (Norse and Gaelic settlers)	Deep time; cradle of humanity
Population structure	Isolated founder population; genetic drift dominant	Extreme diversity; 250+ ethnolinguistic groups
Genomic studies	15,220 WGS (34X); 39 million variants	449 WGS (47 ELGs); 36 million variants
Key discoveries	Atrial fibrillation mutation (MYL4); liver disease variants (ABCB4); TSH-associated variant (GNAS)	Novel and medically relevant variation across ELGs; catalog of African diversity
Research infrastructure	deCODE Genetics; genealogical databases; population-wide biobank	Emerging; collaborations with international partners
Representation in global genomics	Overrepresented; extensively characterized	Severely underrepresented; <3% of GWAS participants

3. RESEARCH QUESTIONS

This study addresses the following research questions:

1: How do the population genetic structures of Iceland and Nigeria differ, and what historical and demographic factors explain these differences?

2: What genomic research infrastructure and large-scale studies exist in each country, and what medically relevant discoveries have emerged?

3: What are the implications of differential representation in genomic research for global health equity and the achievement of SDG 3 targets?

4. METHODOLOGY

This study employs a comparative analysis methodology examining peer-reviewed genomic research from Iceland and Nigeria. The research design integrates findings from population genetics studies, large-scale sequencing projects, and disease association research to develop a comprehensive understanding of the genomic landscapes in both countries.

Primary sources analyzed include: Icelandic population genomics research including studies on population structure , ancient DNA , large-scale sequencing , and the Hans Jonatan historical reconstruction ; Nigerian genomics research including whole-genome sequencing across ethnolinguistic groups and genetic relationship studies using STR markers .

The comparative framework examines four dimensions: (1) population history and genetic structure; (2) research infrastructure and study design; (3) variant discovery and medical relevance; and (4) representation in global genomics.

The analysis covers research published from approximately 2009 to 2024, capturing key developments in both countries' genomic research programs.

5. RESULTS

5.1 Comparative Population Genetic Structure

Iceland: Iceland's population genetic structure reflects its recent founding and subsequent isolation. Research demonstrates that "the Icelandic population is relatively homogeneous, but exhibits subtle population structure that can bias disease association statistics" . Critically, this structure arises from "genetic drift since the settlement of Iceland 1100 years ago, and not to varying contributions from different ancestral populations."

A consequence of this recent origin is that "allele frequency differences follow a null distribution devoid of outliers, so that the risk of false positive associations due to stratification is minimal" . This makes Iceland exceptionally well-suited for disease association studies.

Ancient DNA research confirms that "ancient Icelanders are markedly more similar to their source populations in Scandinavia and the British-Irish Isles than to contemporary Icelanders, who have been shaped by 1100 years of extensive genetic drift" . The study also found "evidence of unequal contributions from the ancient founders to the contemporary Icelandic gene pool."

Nigeria: Nigeria presents a dramatically different picture of extreme genetic diversity. The whole-genome sequencing study across 47 ethnolinguistic groups revealed "genetic differentiation among our ELGs, consistent with previous findings" . The discovery of 36 million SNPs and indels from just 449 individuals underscores the vast genetic variation present.

The STR-based study of five major ethnic groups found that genetic variation is overwhelmingly within individuals (99.98%) rather than among populations (0.10%) . FST values ranging from 0.001 to 0.500 indicate varying degrees of differentiation across groups. Importantly, "the genetic relationships closely mirrored linguistic groupings showing that language may have shaped early interaction and ultimately gene transfer between these groups."

Table 2: Comparative Population Genetic Parameters

Parameter	Iceland	Nigeria
Population homogeneity	Relatively homogeneous; subtle structure	Extremely diverse; 250+ ELGs

Drift vs. admixture	Structure due to recent drift	Structure reflects deep history, migrations, language
Variance components	Not quantified in comparable format	99.98% within individuals; 0.10% among populations
FST values	Minimal due to recent drift	0.001-0.500 across groups
Reference genome utility	High; well-represented in global studies	Poor; underrepresented; novel variants abundant

5.2 Research Infrastructure and Large-Scale Studies

Iceland: Iceland benefits from extraordinary research infrastructure centered on deCODE Genetics, founded in 1996. Key elements include:

- **Population-wide biobank:** Samples from a substantial fraction of the Icelandic population
- **Genealogical database:** Comprehensive records tracing lineages across centuries
- **Healthcare linkage:** Integration with national health records for phenotype-genotype correlation
- **Large-scale sequencing:** 15,220 whole genomes at 34X coverage, identifying 39 million variants

This infrastructure has enabled discovery of variants associated with diverse conditions including atrial fibrillation, liver disease, and thyroid function . The family-based design also enabled high-confidence detection of 108,778 de novo mutations .

Nigeria: Nigeria's genomic research infrastructure is less developed but rapidly evolving. Key initiatives include:

- **Whole-genome sequencing of 449 individuals** across 47 ethnolinguistic groups, providing "a high-level catalog of both novel and medically relevant variation present across the ELGs"
- **STR-based population studies** examining genetic relationships among major ethnic groups
- **International collaborations** with institutions including the London School of Hygiene & Tropical Medicine

The 2023 study emphasized that these results "underscore the potential of using these cohorts with larger sample sizes to improve our understanding of human ancestry and health in Africa" .

5.3 Medically Relevant Variant Discovery

Iceland: Icelandic research has yielded numerous medically significant discoveries:

- **Atrial fibrillation:** "A recessive frameshift mutation in MYL4 that causes early-onset atrial fibrillation"
- **Liver disease:** "Several mutations in ABCB4 that increase risk of liver diseases"
- **Thyroid function:** "An intronic variant in GNAS associating with increased thyroid-stimulating hormone levels when maternally inherited"

- **Rare variants:** "An excess of homozygosity and rare protein-coding variants in Iceland"

Nigeria: Nigerian genomic research has identified extensive medically relevant variation:

- **Novel variants:** The 36 million variants discovered include many not previously catalogued, representing potential contributors to disease
- **Population-specific variation:** Variation across ethnolinguistic groups may inform differential disease risk and drug response
- **African diversity:** The study emphasizes that capturing African genetic diversity is essential for "the equity of precision medicine initiatives globally"

5.4 Representation in Global Genomics

Iceland: Iceland is dramatically overrepresented in genomic research relative to its population size. The extensive characterization of Icelandic genomes means that discoveries from this population are well-represented in global databases.

Nigeria: Nigeria and other African populations are severely underrepresented in genomic research. As noted, "African populations have been drastically underrepresented in genomics research, and failure to capture the genetic diversity across the numerous ethnolinguistic groups (ELGs) found on the continent has hindered the equity of precision medicine initiatives globally" .

This underrepresentation has concrete consequences. Disease-associated variants identified in European populations may not be relevant to African populations; drug dosing guidelines based on European genetics may be inappropriate for African patients; and novel variants with therapeutic potential may remain undiscovered.

5.5 The Hans Jonatan Connection

The story of Hans Jonatan provides a unique historical link between Iceland and Nigeria. Key findings from this remarkable study include:

- **Genealogical reconstruction:** Researchers identified 788 descendants across 8 generations
- **Genomic reconstruction:** Using data from 182 genotyped and 20 sequenced descendants, they "reconstructed 38% of HJ's maternal genome"
- **Geographic origin:** HJ's mother was "from the region spanned by Benin, Nigeria and Cameroon"
- **Historical context:** This represents one of the few documented instances of African gene flow to Iceland before the twentieth century

This study demonstrates both the power of Iceland's genealogical resources and the deep historical connections between African and European populations. It also illustrates how genomic research can illuminate histories obscured by the transatlantic slave trade.

6. DISCUSSION OF RESULTS

6.1 Explaining Divergent Genetic Landscapes

The dramatic differences between Icelandic and Nigerian genetic landscapes reflect fundamentally different demographic histories. Iceland's founding 1,100 years ago by a small number of settlers, followed by isolation and population growth, created a classic founder population. Genetic drift has been the dominant evolutionary force, leading to increased homozygosity, elevated frequencies of rare variants, and subtle population structure arising from recent drift rather than ancient divergence .

Nigeria's genetic landscape, by contrast, reflects deep evolutionary time as part of the ancestral human population in Africa. The immense diversity across ethnolinguistic groups records millennia of population movements, admixture, cultural exchange, and adaptation to diverse environments . The finding that genetic relationships mirror linguistic groupings suggests that language has been both a marker of and a facilitator for gene flow .

6.2 The Power and Limits of Founder Populations

Iceland's success in genomic discovery demonstrates the power of founder populations for identifying disease-associated variants. The combination of population homogeneity, extensive genealogical records, and large-scale sequencing has enabled discoveries that would be more difficult in heterogeneous populations . The minimal risk of false positives due to population stratification further enhances statistical power.

However, the very features that make Iceland valuable for discovery also limit the generalizability of findings. Variants identified in Iceland may have different frequencies, effect sizes, or phenotypic consequences in other populations. The "excess of homozygosity and rare protein-coding variants" reflects Iceland's unique demographic history and may not be representative.

6.3 The Imperative of African Genomic Research

The severe underrepresentation of African populations in genomic research is both a scientific gap and an equity crisis. As the Joshi study emphasizes, failure to capture African genetic diversity "has hindered the equity of precision medicine initiatives globally" . The 36 million variants discovered from just 449 Nigerians—including many novel variants—underscores how much remains to be discovered.

The implications for SDG 3 are profound. Target 3.8 on universal health coverage cannot be achieved if genomic medicine advances benefit only populations of European descent. Target 3.b on supporting research and development for diseases affecting developing countries requires investment in genomic research infrastructure across Africa.

6.4 Lessons from the Hans Jonatan Study

The Hans Jonatan study offers several important lessons. First, it demonstrates the power of integrating genealogical and genomic data to reconstruct individual genomes across generations . Second, it illustrates how genomic research can illuminate histories of migration and admixture, including those obscured by historical trauma. Third, it provides a concrete example of African genetic material entering the European gene pool, challenging narratives of complete separation between African and European populations.

6.5 Implications for SDG 3 and Global Health Equity

The findings have profound implications for achieving SDG 3 targets:

First, Target 3.4 (reducing NCD mortality) requires understanding genetic contributions to disease across populations. Variants discovered in Iceland may not predict disease risk in Nigeria; conversely, variants important in Nigeria may remain undiscovered.

Second, Target 3.8 (universal health coverage) demands that genomic medicine be accessible and relevant to all populations. Precision medicine based on European-ancestry reference genomes will not serve African populations equitably.

Third, Target 3.b (supporting research and development) requires investment in genomic research infrastructure in low- and middle-income countries. The Nigerian studies demonstrate what is possible with such investment.

Fourth, ethical frameworks must ensure benefit-sharing with participating communities. Historical exploitation in genetic research demands that contemporary studies respect participants and ensure that benefits flow back to communities.

7. CONCLUSION

This comparative analysis of genomics research in Iceland and Nigeria reveals two radically different genetic landscapes shaped by divergent demographic histories. Iceland, a young founder population isolated for 1,100 years, has become one of the world's most extensively characterized populations through the work of deCODE Genetics. Large-scale sequencing of 15,220 Icelanders has identified over 39 million variants, enabling discovery of mutations associated with atrial fibrillation, liver disease, and thyroid dysfunction . The population's structure reflects recent genetic drift rather than ancient divergence, minimizing false positives in disease association studies .

Nigeria, by contrast, harbors immense genetic diversity reflecting deep evolutionary history. Whole-genome sequencing of 449 individuals across 47 ethnolinguistic groups discovered 36 million variants, providing a high-level catalog of novel and medically relevant variation . Analysis of five major ethnic groups revealed that genetic relationships closely mirror linguistic groupings, with 99.98% of variation within individuals and only 0.10% among populations .

A remarkable historical connection links these two populations through Hans Jonatan, born in 1784 to an enslaved African mother and European father. Researchers reconstructed 38% of his maternal genome from 182 descendants, tracing his mother's origins to the Benin-Nigeria-Cameroon region . This study demonstrates both the power of Iceland's genealogical resources and the deep historical connections between African and European populations.

Several critical lessons emerge from this comparison.

First, genomic diversity is humanity's shared heritage and essential resource. Iceland's founder population has proven extraordinarily useful for discovery, but the resulting knowledge is not universally applicable. Nigeria's vast diversity holds keys to understanding human evolutionary history and discovering variants relevant to global health.

Second, underrepresentation of African populations in genomic research is both a scientific gap and an equity crisis. As the Nigerian studies emphasize, failure to capture African genetic diversity "has hindered the equity of precision medicine initiatives globally" . Achieving SDG 3's vision of health for all requires correcting this imbalance.

Third, large-scale investment in genomic research infrastructure is essential. Iceland's success rests on decades of sustained investment in biobanks, sequencing, and genealogical databases. Nigeria's emerging genomic research capacity demonstrates what is possible with commitment and international collaboration.

Fourth, ethical frameworks must guide genomic research with diverse populations. Historical exploitation of African communities in genetic research demands that contemporary studies be conducted with respect, transparency, and genuine benefit-sharing.

Fifth, integration of genomic findings with health systems is necessary for impact. Variant discovery alone does not improve health. Research must be linked to clinical translation, drug development, and public health interventions.

Sixth, global collaboration must replace competition. No single country can fully characterize human genetic diversity. International partnerships that respect sovereignty, build local capacity, and share benefits are essential.

For Iceland, the path forward involves continuing to leverage its unique population structure for discovery while ensuring that findings are validated in diverse populations. The de-code model offers lessons for other countries considering large-scale genomic research.

For Nigeria, the priority is expanding genomic research capacity, sequencing larger cohorts across more ethnolinguistic groups, and ensuring that discoveries translate into health benefits for Nigerian populations. The foundation laid by recent studies provides a platform for growth.

For the global community, the imperative is clear: diversify genomic research now, before the benefits of precision medicine become locked into systems that serve only populations of European descent. The Nigerian studies' emphasis that "these results underscore the potential of using these cohorts with larger sample sizes to improve our understanding of human ancestry and health in Africa" should be a call to action.

As the world strives to achieve SDG 3 by 2030, the experiences of Iceland and Nigeria offer essential lessons. Genomic research must be truly global in scope, inclusive in participation, and equitable in benefit. Only then can the promise of precision medicine—treatments tailored to individual genetic makeup—be realized for all humanity.

8. LIMITS OF THE STUDY

This study has several limitations that should be acknowledged. First, the comparative analysis relies primarily on published research rather than primary data analysis. Direct collaboration with researchers in both countries would provide deeper insights.

Second, the Icelandic studies cited span multiple years; the most recent large-scale sequencing data is from 2017 . More recent data may be available but not accessed.

Third, the Nigerian genomic research is rapidly evolving. The studies cited represent significant advances but may not capture the most recent findings.

Fourth, the study focuses on population genetics and variant discovery, with limited attention to clinical translation and health system integration.

Fifth, ethical and social implications are discussed but not systematically analyzed. The complex history of genetic research in Africa, including concerns about exploitation and benefit-sharing, merits deeper examination.

Finally, the voices of research participants and communities are mediated through scientific publications rather than captured directly. Future research should prioritize community engagement and participatory approaches.

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10. AUTHOR CONTRIBUTIONS

The author conceived the study, designed the research methodology, conducted the literature review and document analysis, synthesized findings across countries, and drafted the complete manuscript. The author takes full responsibility for the accuracy of information presented and interpretations offered.

11. COMPETING INTERESTS

The author declares that the research was conducted in the absence of any commercial or financial relationships that could be construed as a potential competing interest. No funding was received from organizations with interests in genomics research, pharmaceutical development, or precision medicine.

12. AUTHORS' CONTRIBUTIONS

As sole author, I performed all roles including conceptualization, methodology development, investigation, formal analysis, writing, review, editing, and project administration.

13. FUNDING INFORMATION

This research received no specific grant from any funding agency in the public, commercial, or not-for-profit sectors.

14. DATA AVAILABILITY

All sources analyzed in this study are publicly available peer-reviewed publications, accessible through academic databases including PubMed, Scopus, and institutional repositories. Primary sources are cited in the bibliography.

15. DISCLAIMER

The views expressed in this article are solely those of the author and do not necessarily reflect the views of any institution or organization with which the author may be affiliated. The analysis is provided for scholarly purposes and should not be construed

as medical advice or policy guidance. Genomic research is rapidly evolving, and readers should consult current sources for specific information about Iceland, Nigeria, and human genetics.

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