MSI Journal of
Medicine and Medical Research (MSIJMMR)

Integrating Bioinformatics and Genomic Profiling to Improve Early Detection of Chronic Kidney Disease in Nigeria

Adebiyi A.O[1], Amoo A.H[1*], Adeosun M.O[1], Daud M.A[1], Salaudeen O.S[1], Abdulrauf A[1], Olowolayemo K.O[1], Hamzat F.O[1], Ibrahim S.O[1], Eze S.M[1], Atoyebi A.T[1], Lawal I.A[1], Dare B.J[1]

[1]Department of Human Anatomy, Faculty of Basic Medical Sciences, Al-Hikmah University, Ilorin, Nigeria.

*** Correspondence:** Amoo Ayomide Hammed

The authors declare that no funding was received for this work.

OPEN ACCESS

Received: 10-January-2026
Accepted: 25-February-2026
Published: 03-March-2026

This article is published in the **MSI Journal of Medicine and Medical Research (MSIJMMR)**
ISSN 3049-1401 (Online)

The journal is managed and published by MSI Publishers.

Volume 3, Issue 1 (Jan-Apr), 2026

ABSTRACT: Chronic kidney disease (CKD) remains a major global health burden, with sub-Saharan Africa, particularly Nigeria, experiencing disproportionately high prevalence and mortality rates. Conventional diagnostic markers such as serum creatinine, estimated glomerular filtration rate (eGFR), and urine albumin primarily detect functional decline and do not capture early molecular alterations. This limitation highlights the urgent need to integrate genomics and bioinformatics approaches to improve early detection and precision management of CKD in Nigeria. This review examines the genetic and molecular mechanisms underlying CKD, focusing on key susceptibility genes such as *APOL1, MYH9, UMOD,* and *COL4A5*, which influence disease onset and progression in African populations. Evidence from genome-wide association studies (GWAS) and next-generation sequencing (NGS) demonstrates the importance of identifying population-specific genetic variants to enhance predictive accuracy. Bioinformatics tools and public repositories, including the Gene Expression Omnibus (GEO), were also explored for their role in biomarker discovery, such as CCR7, and in the development of predictive

and personalized therapeutic models. The findings indicate that Nigeria's genomic research landscape is expanding through initiatives led by NIMR, LASU, Covenant University, and ACEGID, supported by continental collaborations like H3Africa. However, progress remains limited by inadequate funding, poor infrastructure, insufficient training, and weak translational frameworks. Emerging opportunities such as affordable sequencing technologies, regional genomic databases, and structured capacity-building initiatives offer promising pathways for advancement. In conclusion, integrating genomics and bioinformatics into Nigeria's CKD management framework requires stronger policy support, improved research infrastructure, and targeted human-capacity development. Strengthening these areas will promote the adoption of precision medicine, enable earlier disease detection, and enhance health equity across Nigerian populations.

Keywords: *Chronic kidney disease; Genomic profiling; Bioinformatics integration; Genetic susceptibility; Precision medicine; Nigeria.*

1. Introduction

1.1 Overview of Chronic Kidney Disease and Its Burden in Nigeria

Chronic kidney disease (CKD) is a major global health problem, affecting approximately 13.4% of the world's population, with an even higher prevalence of 15.8% in sub-Saharan Africa (Govender *et al.,* 2021). Globally, reduced kidney function accounts for about 1.2 million deaths and 19 million disability-adjusted life years each year, reflecting a 32% increase in kidney-related deaths since 2005 (Aderinto *et al.,* 2024).

In Nigeria, CKD presents a serious public-health and socioeconomic challenge, as most patients are diagnosed at advanced stages, when access to dialysis or transplantation is limited. Renal disease consumes over 3% of national health budgets in high-income countries, a cost level unsustainable for low-resource settings like Nigeria (Aderinto *et al.,* 2024). The predominant risk factors include hypertension and diabetes mellitus, while infectious diseases such as HIV, hepatitis B and C, malaria, and schistosomiasis further exacerbate kidney damage (Govender *et al.,* 2021). Furthermore, APOL1 and MYH9 gene variants, which are common among individuals of African ancestry, increase susceptibility to severe renal

complications (Aderinto *et al.,* 2024). Given these challenges, integrating bioinformatics and genomic profiling offers a potential solution for improving early CKD detection through population-specific risk assessment and precision-based prevention strategies (Alobaidi, 2025).

1.2 Gaps in Current Diagnostic and Management Approaches

Despite advances in clinical practice, current CKD diagnostic tools like serum creatinine, estimated glomerular filtration rate (eGFR), and urine albumin testing remain limited in identifying early disease. A low eGFR (<60 ml/min/1.73 m^2) only becomes evident after substantial nephron loss, making it a late marker of kidney damage (Govender *et al.,* 2021). Similarly, while albuminuria is often used as an early indicator, CKD progression can occur even in its absence, reducing its predictive reliability. The serum creatinine test, though widely used, is influenced by muscle mass, age, diet, and hydration, leading to inaccurate results, particularly in undernourished or elderly individuals (Aderinto *et al.,* 2024).

These tests largely measure functional decline rather than the molecular or structural changes that precede it, leading to delayed detection and missed opportunities for early intervention. As Govender et al. (2021) note, there is a pressing need for sensitive and specific biomarkers capable of identifying subclinical kidney injury before irreversible damage occurs. Integrating bioinformatics and genomic profiling could address these limitations by enabling detection of molecular alterations and genetic susceptibilities earlier in disease development, facilitating more effective and personalized management strategies (Alobaidi, 2025).

1.3 Rationale for Genomic and Bioinformatics Integration

The limitations of traditional CKD diagnostics highlight the need for innovative molecular tools that detect disease before significant renal impairment develops. Genomic and bioinformatics approaches meet this need by enabling large-scale analysis of genetic, proteomic, and metabolomic data, allowing researchers to identify biomarkers associated with early kidney injury (Alobaidi, 2025). Through genomic profiling, risk alleles and polygenic risk scores can be developed to stratify individuals by genetic susceptibility, supporting earlier and more targeted interventions (Aderinto *et al.,* 2024).

Furthermore, bioinformatics and machine learning enhance this process by integrating diverse datasets to build predictive models that improve disease forecasting and treatment optimization (Alobaidi, 2024). Together, these technologies form the foundation of precision medicine, offering a pathway toward earlier detection, personalized prevention, and improved CKD outcomes in Nigeria.

1.3 Search Strategy and Inclusion Criteria

This narrative review synthesized peer-reviewed literature on genomic profiling and bioinformatics applications in chronic kidney disease (CKD), with particular emphasis on Nigerian and African populations. Databases searched included PubMed, Google Scholar, and Web of Science for studies published between 2010 and 2025. Search terms included combinations of "chronic kidney disease," "CKD," "genomics," "bioinformatics," "GWAS," "NGS," "Nigeria," and "Africa."

Studies were included if they:

I. Reported genomic or bioinformatics findings relevant to CKD
II. Focused on African or Nigerian populations
III. Examined biomarker discovery or predictive modeling

2. Genomic and Molecular Insights into Chronic Kidney Disease

2.1 Genetic and Molecular Basis of Chronic Kidney Disease

Chronic Kidney Disease (CKD) involves complex molecular mechanisms driven by oxidative stress, inflammation, and fibrosis. As illustrated in Figure 1, these interconnected processes contribute to renal injury and progressive loss of kidney function.

1. Oxidative stress results from an excessive amount of reactive oxygen and nitrogen species, leading to disruption of redox signaling and oxidation of lipids, proteins, and DNA. The kidneys, being highly metabolic organs, are particularly vulnerable to oxidative damage caused by mitochondrial dysfunction and NADPH oxidase activity. Uremic toxins such as indoxyl sulphate and p-cresyl sulfate increase oxidative stress by stimulating superoxide production and activating profibrotic factors like transforming

growth factor-β1 (TGF-β1). This oxidative damage contributes to glomerular injury, albuminuria, interstitial fibrosis, and chronic inflammation.

2. Inflammatory pathways involving cytokines such as interleukins (IL-1, IL-6, and IL-20) play key roles in CKD progression by promoting immune cell infiltration, apoptosis, and fibrotic changes. Macrophage activation and the imbalance between M1 and M2 phenotypes further amplify renal inflammation and tissue remodeling. Moreover, nod-like receptor protein 3 (NLRP3) inflammasome activation triggers caspase-1–mediated cytokine release and pyroptosis, linking immune activation to kidney injury. Other molecular mediators, including neutrophil gelatinase-associated lipocalin (NGAL) and matrix metalloproteinases (MMP-2, MMP-7, and MMP-9), are implicated in tubular damage, extracellular matrix remodeling, and fibrosis, serving as potential biomarkers for kidney injury.
3. Matrix metalloproteinases (MMPs) are proteolytic enzymes involved in extracellular matrix deposition, cell differentiation, angiogenesis, inflammation, proliferation, vascular damage, and apoptosis. There are about 20 mammalian MMPs, divided into collagenases, gelatinases, stromelysins, matrilysins, membrane-type, and other MMPs. MMPs play a significant role in the pathogenesis of many diseases, including cardiovascular and metabolic disorders, and are crucial in the progression of CKD (Frąk *et al.,* 2022; Patera & Gatticchi, 2024).

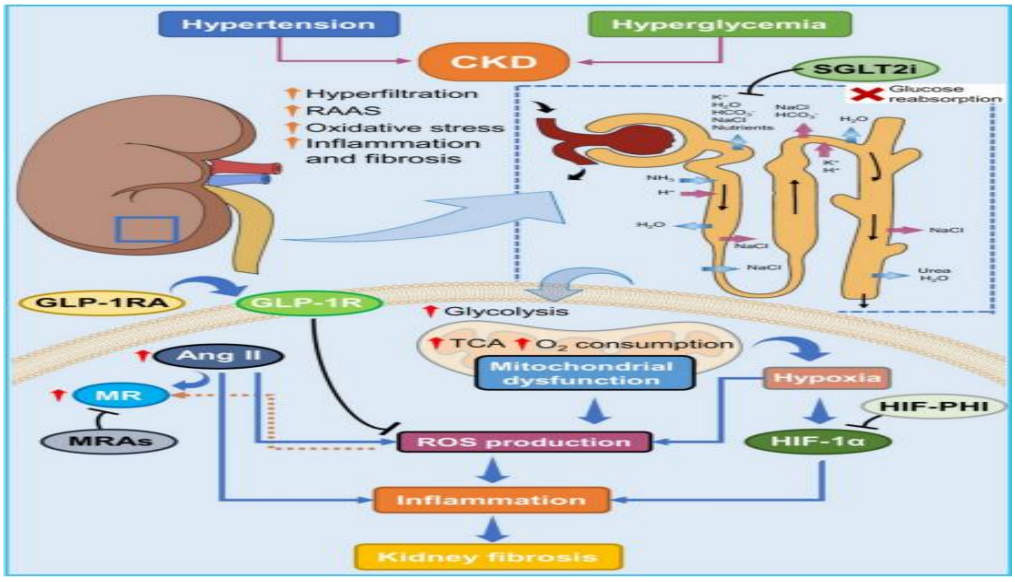

Figure 1: Molecular Pathway Driving Chronic Kidney Disease Progression (Patera *et al.,* 2024)

The genetic and molecular basis of chronic kidney disease (CKD) is complex, involving multiple forms of genetic alterations including copy number variations (CNVs), single nucleotide polymorphisms (SNPs), and mitochondrial mutations. CNVs, large genomic deletions or duplications represent a significant source of genetic diversity linked to renal disorders. Studies using chromosomal microarrays and sequencing technologies have demonstrated that CNVs contribute to both congenital and acquired forms of CKD, such as congenital anomalies of the kidney and urinary tract (CAKUT), posterior urethral valves, and nephronophthisis. Pathogenic CNVs involving genes like HNF1B, EYA1, and NPHP1 have been implicated in pediatric CKD, often requiring individualized clinical approaches. These structural variants typically arise from unequal recombination, replication stress, or chromothripsis, disrupting essential genes in kidney development and function (Lew *et al.,* 2018).

In addition, genome-wide association studies (GWAS) have identified several SNPs associated with CKD susceptibility. Variants in UMOD, APOL1, MYH9, and CUBN have been consistently linked with altered estimated glomerular filtration rate (eGFR) and urinary albumin-to-creatinine ratio (UACR). Among these, UMOD variants are strong genetic determinants of CKD risk, influencing uromodulin expression and sodium handling. Although over 50 genetic loci related to kidney traits have been identified, each contributes only modestly to disease heritability, underlining the polygenic and multifactorial nature of CKD. Recent next-generation sequencing (NGS) and whole-exome sequencing (WES) efforts have uncovered rare pathogenic mutations, such as those in PARN, a gene linked to telomere maintenance and renal dysfunction (Haas *et al.,* 2018; Cañadas-Garre *et al.,* 2019).

Mitochondrial stress or damage is also a central molecular mechanism in CKD progression (Figure 2). Under stress, mitochondrial biogenesis and oxidative phosphorylation (OXPHOS) activity decline, primarily through dysregulation of genes including PPARGC1A, NRF1, TFAM, UQCRH, COX6C, COX7C, MCAD, and AIFM1. This reduction impairs energy metabolism and triggers abnormal autophagy and mitophagy mediated by ATG5, CTSD, and BECN1, resulting in accumulation of dysfunctional mitochondria. Consequently, intrinsic apoptosis

pathways are activated via BCL2L1 and AIFM1, leading to renal cell death. Mitochondrial dysfunction also promotes inflammation and fibrosis through genes such as NLRP3, CASP1, PPARG, and TGFB1, while reduced expression of NRP2, KLF6, and CTSD disrupts podocyte function, contributing to glomerular injury and CKD progression. Conversely, upregulation of protective genes like BNIP3, BAX, BAK1, HIF1, and ITCH may represent compensatory responses attempting to mitigate mitochondrial stress. Overall, the dysregulation of mitochondrial biogenesis, autophagy, apoptosis, and inflammatory signaling provides a strong molecular link between genetic variation and CKD pathogenesis (Cañadas-Garre *et al.,* 2019).

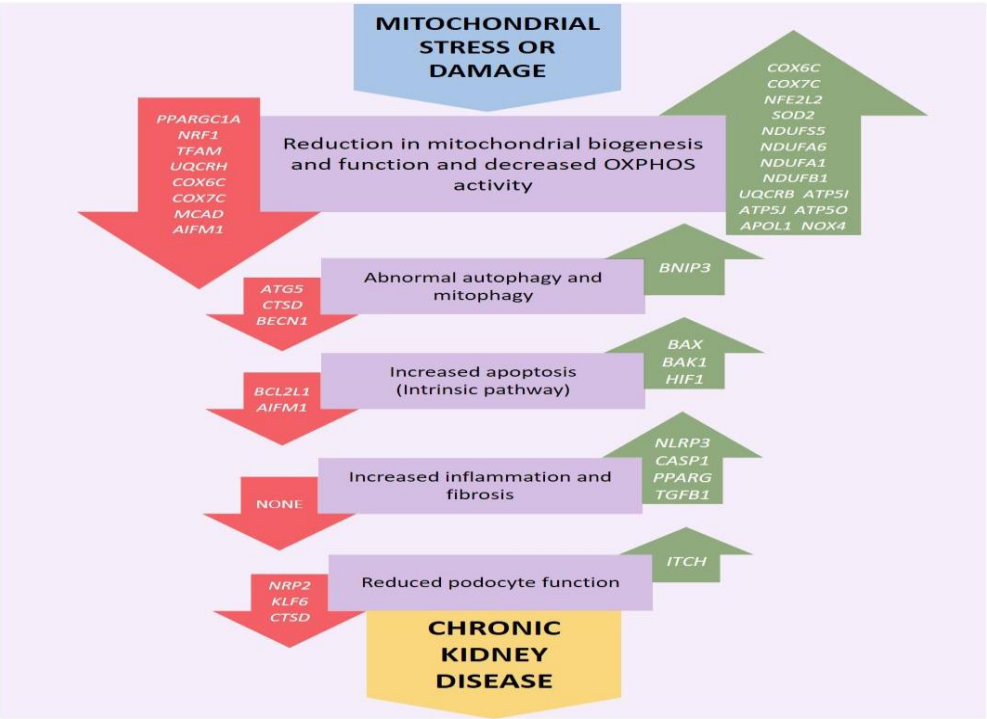

Figure 2: Gene-Regulated Mitochondrial Stress Responses in Chronic Kidney Disease (Cañadas-Garre *et al.,* 2019).

2.2 Findings from Global and African Genomic Studies

The discovery of genetic regions linked to chronic illnesses, including chronic kidney disease (CKD), has been greatly advanced by genome-wide association studies (GWAS) and modern sequencing technologies. Genome-wide association studies involve scanning millions of single nucleotide polymorphisms (SNPs) across diverse populations to identify genetic variations that influence disease susceptibility, progression, and treatment response (Chen *et al.,* 2024). These studies have

illuminated how genetic diversity contributes to differences in disease prevalence and outcomes among populations. Globally, GWAS have identified numerous SNPs associated with chronic conditions such as cardiovascular diseases, diabetes, and neurodegenerative disorders. Each identified variant serves as a potential genetic biomarker, offering insights into disease mechanisms and identifying molecular targets that could inform new therapies.

In Africa, genomic research is beginning to reveal population-specific genetic variants that contribute to CKD and related chronic illnesses. The remarkable genetic diversity among African populations presents an opportunity to uncover novel alleles that are absent in other regions. For example, variations in the apolipoprotein L1 (APOL1) and non-muscle myosin heavy chain 9 (MYH9) genes, which occur more frequently in people of African ancestry, have been strongly linked to increased susceptibility to kidney disease and faster progression to end-stage renal failure (Ekulu *et al.,* 2021). Expanding GWAS and sequencing studies in African and Nigerian populations is therefore essential to address their historical underrepresentation in global genetic research. Combining genomic discoveries from both African and international cohorts supports the development of precision medicine strategies that tailor disease prevention, diagnosis, and treatment to population-specific genetic profiles. Such efforts will ensure that genomic medicine contributes equitably to improving the prediction, management, and overall control of CKD in Nigeria and across the African continent (Joshi *et al.,* 2023).

2.3 Implications of Genomic Profiling for Early Detection

Genomic profiling has emerged as a transformative tool for the early detection and diagnosis of kidney diseases, particularly in identifying genetic causes that might otherwise remain undetected through conventional testing. Evidence from large-scale studies demonstrates that exome sequencing (ES) achieves a diagnostic yield of around 39% in patients with suspected genetic kidney disease, with higher rates in children and those with early disease onset. These findings underscore the power of genomic testing to provide precise molecular diagnoses, clarify or reclassify clinical assessments, and directly influence patient management, in many cases, genomic profiling negated the need for invasive renal biopsies, informed treatment plans, and

guided surveillance and family counseling, thereby improving patient care and cost-effectiveness. Early genomic testing allows for the identification of disease at a molecular stage before irreversible damage occurs supporting personalized interventions and family-based screening. However, broader implementation requires addressing barriers such as workforce training, testing costs, and access to genetic counseling (Jayasinghe *et al.,* 2021; Jefferis & Mallett,2024)

3. The Role of Bioinformatics in CKD Research and Early Detection

3.1 Overview of Bioinformatics Tools and Databases in CKD Research

Computational tools extract meaningful and useful information from large amounts of data and help researchers comprehend large datasets. Information on genetic variations associated with diseases enables researchers to understand the underlying genetic factors, discover potential biomarkers, and uncover pathways for targeted interventions in various diseases (Mohd Athar et al., 2024). Genetic variation information explains the interplay between diseases and genetic elements. For example, the Gene Expression Omnibus (GEO) is a public database that provides a platform for the storage and retrieval of high-throughput gene expression and molecular abundance data (Mohd Athar et al., 2024). Researchers worldwide can access and analyze a diverse range of genomic datasets, contributing to the advancement of genomics and biomedical research. In one study, Barrios et al. (2025) utilized GEO datasets to perform an integrated bioinformatics analysis comparing diabetic and non-diabetic CKD, identifying differentially expressed genes such as TGFB1, FN1, and PPARGC1A, which are involved in fibrosis, inflammation, and mitochondrial dysfunction. Using computational tools like DAVID, STRING, and Cytoscape, they uncovered critical molecular pathways contributing to CKD progression, demonstrating how bioinformatics databases and tools can enhance understanding of the genetic and molecular mechanisms underlying kidney disease.

3.2 Applications in Biomarker Discovery and Predictive Modeling (Revised)

Genetic biomarkers serve as powerful indicators for chronic diseases, providing insight into disease susceptibility, progression, and potential therapeutic targets. In

the context of chronic kidney disease (CKD), the use of computational and bioinformatics tools has made it possible to uncover complex molecular patterns and identify biomarkers that can improve diagnosis and prognosis. Traditional approaches relying only on measures of renal function, such as serum creatinine or estimated glomerular filtration rate, often fail to capture early molecular changes that precede functional decline. Therefore, integrating genomic and computational analyses offers a more precise means of predicting CKD progression and tailoring interventions to individual genetic profiles (Lai *et al.,* 2024).A key example of this approach is the use of the Gene Expression Omnibus (GEO) microarray dataset, specifically dataset GSE45980, to identify differentially expressed genes (DEGs) among patients with progressive and stable CKD. Through this analysis, researchers identified the C–C chemokine receptor type 7 (CCR7) gene as a significant biomarker candidate. CCR7 plays an essential role in immune response regulation, and its ligands such as C–C chemokine ligand 19 (CCL19) and C–C chemokine ligand 21 (CCL21) were also identified as differentially expressed genes in CKD patients. This finding suggests that the CCR7–CCL19/CCL21 signaling axis may influence disease progression.

Further structural and functional insights reveal that CCL21 contains a unique 32-amino acid C-terminal tail, enriched in basic residues that facilitate binding to glycosaminoglycans and other extracellular molecules. While CCL21 primarily mediates cell migration through CCR7, CCL19 complements this action by promoting non-migratory processes, such as enhancing cell survival. Both ligands were upregulated in CKD samples, reinforcing their biological relevance. Notably, CCR7 expression correlates with CKD severity but is independent of patient age or sex, indicating its robustness as a prognostic biomarker across diverse populations. Together, these findings highlight CCR7 and its associated ligands as promising molecular targets for predicting CKD progression and developing novel therapeutic interventions (Lai *et al.,* 2024).

3.3 Integration of Bioinformatics and Genomic Data in Clinical Research

There are various genetic tests available, and it is essential to understand the distinctions between the sequencing methods used to generate data and the specific

analyses performed (Halbritter *et al.,* 2025). The scope of analysis has significant implications for patient care. While our understanding of genetic variants and detection technologies grows, it remains limited; therefore, a negative test does not exclude the use of genetics. Interpretation of negative test results must consider the specific gene selection used, the limitations, and the year the test was performed (Halbritter *et al.,* 2025). The broader the analysis, the higher the chance of incidental findings, which may impact the care of the patient (Halbritter *et al.,* 2025).

Patients who are better informed about the trajectory of their diseases and therapies may be more likely to make necessary lifestyle and behavior changes, from increased exercise to compliance with personalized medicine decisions (Recharla *et al.,* 2023). Patients who are more engaged may get diagnosed earlier, follow treatment regimens more closely, suffer fewer emergency events, and ultimately lose fewer healthy person-years (Recharla *et al.,* 2023).

4. Nigeria and the African Genomic Landscape

4.1 Current Status of Genomics and Bioinformatics Research in Nigeria

Genomics and bioinformatics are relatively new but rapidly expanding scientific fields in Nigeria. Over the past two decades, there has been growing engagement by Nigerian researchers, supported by both national and international collaborations. Nigeria's genetic diversity, with over 250 ethnic groups and 500 languages, provides a unique opportunity for studies in population genetics, disease susceptibility, and biodiversity conservation. Since the launch of initiatives such as the Human Heredity and Health in Africa (H3Africa) Consortium, the generation of genomic data and collaborative research capacity across African institutions has significantly increased (Fatumo *et al*., 2020).

Several key institutions are leading genomic and bioinformatics research in the country, including the Nigerian Institute of Medical Research (NIMR), the Centre for Genomics Research and Innovation (CGRI) at the National Biotechnology Development Agency (NBDA), the African Centre of Excellence for Genomics of Infectious Diseases (ACEGID) at Redeemer's University, and the African Collaborative Centre for Microbiome and Genomics Research (ACCME). Other

notable projects include the Cassava and Yam Genomics Projects at the International Institute of Tropical Agriculture (IITA), Ibadan (Fatumo *et al.,* 2020)

The establishment of the Nigerian Bioinformatics and Genomics Network (NBGN) in 2019 has further consolidated national efforts by connecting researchers and promoting collaborations among Nigerian bioinformaticians and genomic scientists. Through conferences, workshops, and training programs, NBGN supports capacity building and research skill development across universities and research institutions. These collective efforts have resulted in a steady rise in genomic publications and capacity development in recent years, highlighting Nigeria's growing influence in African genomics research (Choudhury *et al.,* 2020; Fatumo *et al.,* 2020).

Despite progress, the current level of infrastructure, expertise, and funding remains below national research needs. However, ongoing initiatives such as NBGN and institutional partnerships continue to strengthen Nigeria's position as a regional hub for genomics and bioinformatics advancement. (Choudhury *et al.,* 2020).

4.2 Challenges Hindering Advancement in Nigeria

Despite growing interest, the expansion of genomics and bioinformatics in Nigeria faces numerous structural and systemic barriers. The most significant is limited and inconsistent research funding. National research budgets rarely prioritize molecular sciences, resulting in dependence on external grants that are often short-term and project-specific (Fatumo *et al.,* 2022). This restricts long-term sustainability and hinders local ownership of genomic programs.

Infrastructure gaps also remain a major obstacle. Many laboratories lack essential facilities such as next-generation sequencing (NGS) platforms, reliable power supply, and high-performance computing systems required for bioinformatics research. Where equipment exists, maintenance challenges and inadequate technical support frequently disrupt research continuity. Furthermore, access to stable internet and digital storage for large genomic datasets is limited, impeding data analysis and sharing. Another persistent barrier is shortage of trained professionals. Although regional initiatives like H3Africa have created training opportunities, the number of skilled bioinformaticians and molecular geneticists in Nigeria is still insufficient for

the growing research demand. The migration of trained scientists to institutions abroad has further worsened this shortage, creating a capacity drain that undermines local expertise.

Additionally, issues of data access, ownership, and governance also present additional challenges. Data-sharing restrictions and concerns over misuse of African genomic information often discourage collaboration and delay publication of results (Choudhury *et al.,* 2020). This limits Nigeria's visibility in global genomics databases and reduces the potential for comparative multi-population studies. Finally, the weak link between research and clinical translation continues to hinder progress. Genomic discoveries have yet to be incorporated into Nigeria's healthcare practice, largely due to a lack of policy frameworks connecting research institutions with health ministries and clinical laboratories (Oluniyi *et al.,* 2021).

4.3 Opportunities for Sustainable Growth and Collaboration

Despite these challenges, Nigeria holds significant opportunities to strengthen its genomics and bioinformatics landscape. The declining cost of sequencing technologies has made genomic analysis more accessible to developing countries. Portable sequencing tools and cloud-based bioinformatics platforms now allow high-quality genomic studies to be conducted with reduced infrastructure requirements (Fatumo *et al.,* 2022). Leveraging these technologies can democratize access and increase participation in large-scale genomic initiatives. The establishment of regional genomic databases and biobanks represents another opportunity for sustainable growth. Integrating Nigerian datasets into Pan-African repositories such as H3ABioNet enhances representation of African genetic diversity in global research and supports the identification of population-specific disease markers (Choudhury *et al.,* 2020). This inclusion is essential for improving the precision and equity of genomic medicine.

Furthermore, international and regional collaborations offer avenues for capacity building. Continued partnerships with global institutions can provide technical expertise, mentorship, and access to advanced analytical tools. Centers such as ACEGID and the African Genomic Centre demonstrate that with adequate support,

local scientists can lead world-class genomic surveillance and bioinformatics projects (Oluniyi *et al.,* 2021). Nigeria can also capitalize on emerging training programs in bioinformatics and data science to expand human capital. Academic institutions are increasingly incorporating computational biology into their postgraduate curricula, a move that will gradually reduce reliance on foreign expertise (Fatumo *et al.,* 2020). In addition, collaborative frameworks between universities, government agencies, and industry can promote innovation, translating genomic findings into diagnostic tools, therapeutics, and precision medicine applications relevant to Nigerian populations (Choudhury *et al.,* 2020).

Integrating genomics into national health policy could enhance disease surveillance and prevention. By linking genomic research with epidemiological data, Nigeria can adopt a precision public-health approach to better predict and manage diseases such as CKD, malaria, and diabetes. This strategy aligns with Africa's broader vision of harnessing genomics to achieve sustainable health equity (Fatumo *et al.,* 2022).

5. Future Perspectives and Conclusion

5.1 Strengthening Genomic and Bioinformatics Capacity in Nigeria

National and cooperative programs like the Nigerian Bioinformatics and Genomics Network (NBGN), which was founded to link researchers and advance data-driven genomics research throughout the nation and Africa, have played a significant role in the development of bioinformatics and genomics capacity in Nigeria (Fatumo *et al.,* 2020). Through webinars, workshops, conferences, and mentorship programs, the NBGN has successfully connected almost 3,000 members, enhancing training partnerships and research capacity.

To empower Nigerian researchers in genomic data science and precision medicine, the second NBGN conference (NBGN21), themed "Leveraging Bioinformatics and Genomics for the Attainment of the Sustainable Development Goals," highlighted the necessity of infrastructure development, capacity building, and policy integration. In order to improve local proficiency in analyzing genomic data, the one-day Genomic Analysis Workshop on Genome-wide Association Study (GWAS) and Polygenic

Risk Score (PRS) analysis was created as a research capacity-building initiative, supporting SDG 4 – Quality Education (Fatumo *et al.,* 2022).

The significance of genomics and bioinformatics in tackling non-communicable diseases (NCDs) via population-specific genetic research was also emphasized at the conference. Understanding African genetic diversity is essential to identifying disease-susceptibility variants that could guide improved clinical translation and policy action (Bentley et al., 2020). Strengthening genomic education and institutional research infrastructure was highlighted as crucial to enable timely interventions for diseases like chronic kidney disease (CKD) and other NCDs common in African populations (Fatumo *et al.,* 2022).

5.2 Future Research Direction and Policy Implications

The availability of services for end stage kidney disease (ESKD) care varies greatly worldwide, with low-income countries facing the most limited access to diagnostic tests and treatments for blood pressure, anaemia, bone disease, and electrolyte disorders. While most countries can measure haemoglobin and provide iron therapy, fewer can offer advanced treatments such as non-calcium-based phosphate binders and cinacalcet. Nephrologists are the main providers of ESKD care in most countries, but global workforce shortages persist, particularly among nephrologists, interventional radiologists, dialysis access surgeons, and transplant surgeons. The number of nephrologists generally increases with national income levels, and there has been slight improvement since the previous survey. Most nations have established dialysis and transplant registries, though fewer track acute kidney injury or chronic kidney disease. Around half of countries regularly report patient outcomes and quality indicators such as blood pressure, haemoglobin, and survival rates. Oversight of ESKD care is mainly handled by national governments, while others rely on hospitals, regional authorities, or NGOs. Access to kidney replacement therapy also differs between adults and children, especially in low-income countries. Compared to the 2017 survey, the 2019 data reveal broader global coverage, improved dialysis and transplant availability, and more countries developing registries for kidney disease (Bello *et al.,* 2021).

Genomic research has revealed genetic predispositions to many chronic conditions, and advances in sequencing technologies are paving the way for truly personalized medicine. Future studies will continue exploring rare genetic variants, non-coding genomic regions, and epigenetic modifications, ultimately enabling more targeted and effective therapies tailored to individual genetic profiles (Kessler, 2018).

5.3 Study Limitation

This review adopts a narrative synthesis approach, which allows for broad conceptual integration of genomic and bioinformatics evidence but does not employ a formal systematic search protocol or quantitative meta-analytic framework. As such, while the discussion captures key developments in CKD genomics and precision medicine, it does not provide pooled effect estimates or structured quality grading of included studies. In addition, much of the genomic data referenced derives from pan-African or global cohorts, reflecting the current structure of available evidence and underscoring the need for larger Nigeria-specific genomic datasets. Future research may build upon this foundation by conducting systematic reviews, longitudinal cohort studies, and predictive validation analyses within Nigerian populations to further strengthen clinical translation and policy alignment.

5.4 Conclusion: Toward Precision Medicine for CKD in Nigeria

Chronic kidney disease remains a major public health challenge in Nigeria due to late diagnosis, limited access to advanced care, and reliance on conventional biomarkers that detect disease only after significant renal damage has occurred. This review highlights the critical role of integrating genomics and bioinformatics in addressing these limitations by enabling earlier detection, improved risk stratification, and personalized management of CKD.

Evidence from global and African genomic studies demonstrates that population-specific genetic variants, particularly those prevalent among individuals of African ancestry, significantly influence CKD susceptibility and progression. Bioinformatics tools and public genomic databases further enhance biomarker discovery and predictive modeling, offering opportunities for precision-based prevention and treatment strategies.

Although Nigeria has made notable progress through national institutions and continental collaborations, challenges such as inadequate funding, limited infrastructure, workforce shortages, and weak translational frameworks persist. Addressing these gaps through strengthened policy support, sustained investment in research capacity, and integration of genomic data into clinical practice will be essential. Advancing precision medicine approaches for CKD in Nigeria has the potential to improve early diagnosis, optimize patient outcomes, and promote equitable kidney healthcare across diverse populations.

REFERENCES

1. Aderinto, N., Olatunji, G., Kokori, E., Ogieuhi, I. J., Babalola, A. E., Ayodeji, K. B., … Ishola, I. V. (2024). *Genomic insights into renal diseases: Advancements and implications. The Egyptian Journal of Internal Medicine, 36*(1), 73. https://ejim.springeropen.com/articles/10.1186/s43162-024-00341-5

2. Adoga, M. P., Fatumo, S. A., & Agwale, S. M. (2014). *H3Africa: A tipping point for a revolution in bioinformatics, genomics and health research in Africa. Source Code for Biology and Medicine, 9*, 12. https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4019950/

3. Alobaidi, S. (2025). Emerging Biomarkers and Advanced Diagnostics in Chronic Kidney Disease: Early Detection Through Multi-Omics and AI. *Diagnostics*, *15*(10), 1225. https://doi.org/10.3390/diagnostics15101225

4. Barrios, C., Riera, M., Rodríguez, E., Márquez, E., Del Risco, J., Pilco, M., ... & Crespo, M. (2025). Identification of Differentially Expressed Genes and Pathways in Non-Diabetic CKD and Diabetic CKD by Integrated Human Transcriptomic Bioinformatics Analysis. International Journal of Molecular Sciences, 26(15), 7421.

5. Bello, A. K., Levin, A., Lunney, M., Osman, M. A., Ye, F., Ashuntantang, G., Okpechi, I. G., Jha, V., Feehally, J., Harris, D. C. H., Kalantar-Zadeh, K., Luyckx, V. A., & the ISN–GKHA Working Group. (2021). *Status of care for end stage kidney disease in countries and regions worldwide: International Society of*

Nephrology Global Kidney Health Atlas (ISN–GKHA) 2019. Kidney International Supplements, 11(2), e1–e150. https://doi.org/10.1016/j.kisu.2021.01.001

6. Bentley, A. R., Callier, S. L., & Rotimi, C. N. (2020). *Evaluating the promise of inclusion of African ancestry populations in genomics. npj Genomic Medicine, 5*, 5. https://www.nature.com/articles/s41525-019-0111-x

7. Cai, M., Li, X., Zha, Y., Luo, T., & Ding, X. (2018). *Genetic variants associated with chronic kidney disease and related traits: A comprehensive review. Kidney Diseases, 4*(1), 1–12.

8. Cañadas-Garre, M., Anderson, K., Cappa, R., Skelly, R., Smyth, L. J., McKnight, A. J., & Maxwell, A. P. (2019). *Genetic susceptibility to chronic kidney disease – Some more pieces for the heritability puzzle. Frontiers in Genetics, 10*, Article 453. https://doi.org/10.3389/fgene.2019.00453

9. Cavalcanti Brandão, L. A., Rodrigues de Moura, R., Rodrigo Assunção, B., Del Vecchio, C., d'Adamo, A. P., Ratzinger, G., ... & Crovella, S. (2025). *Genomic profiling in hidradenitis suppurativa: InterOmics pipeline for DNA-RNA sequencing highlights HLA variants, keratin-associated mutations and extracellular matrix alterations as contributing factors to HS pathogenesis. PLoS One, 20*(6), e0326458.

10. Choudhury, A., Aron, S., Sengupta, D., et al. (2020). *High-depth African genomes inform human migration and health. Nature, 586*(7831), 741–748. https://www.nature.com/articles/s41586-020-2859-7

11. Ekulu, P. M., Nkoy, A. B., Adebayo, O. C., et al. (2021). *A focus on the association of APOL1 with kidney disease in children. Pediatric Nephrology, 36*, 777–788. https://doi.org/10.1007/s00467-020-04553-z

12. Fatumo S, Ebenezer TE, Ekenna C, et al. The Nigerian Bioinformatics and Genomics Network (NBGN): a collaborative platform to advance bioinformatics and genomics in Nigeria. Global Health, Epidemiology and Genomics. 2020;5:e3. doi:10.1017/gheg.2020.3

13. Fatumo, S., Chikowore, T., Choudhury, A., Ayub, M., Martin, A. R., & Kuchenbaecker, K. (2022). *A roadmap to increase diversity in genomic studies. Nature Medicine, 28*(2), 243–250. https://www.nature.com/articles/s41591-021-01672-4

14. Frąk, W., Kućmierz, J., Szlagor, M., Młynarska, E., Rysz, J., & Franczyk, B. (2022). *New insights into molecular mechanisms of chronic kidney disease. Biomedicines, 10*(11), 2846. https://doi.org/10.3390/biomedicines10112846

15. Good, D. M. (2010). *Discovery and validation of urinary peptide biomarkers for chronic kidney disease using capillary electrophoresis mass spectrometry. Clinical Proteomics, 6*(1–2), 33–46.

16. Govender, M. A., Brandenburg, J.-T., Fabian, J., & Ramsay, M. (2021). *The use of 'omics for diagnosing and predicting progression of chronic kidney disease: A scoping review. Frontiers in Genetics, 12*, 682929. https://www.frontiersin.org/articles/10.3389/fgene.2021.682929/full

17. Haas, M. E., Aragam, K. G., Emdin, C. A., Bick, A. G., International Consortium for Blood Pressure, Hemani, G., et al. (2018). *Genetic association of albuminuria with cardiometabolic disease and blood pressure. American Journal of Human Genetics, 103*, 461–473. https://doi.org/10.1016/j.ajhg.2018.08.004

18. Halbritter, J., Figueres, L., Van Eerde, A. M., Capasso, G., Hoorn, E. J., Nijenhuis, T., Pérez-Gómez, M. V., Sayer, J. A., Simons, M., Walsh, S., Zagorec, N., Müller, R.-U., & Cornec-Le Gall, E. (2025). *Chronic kidney disease of unexplained cause (CKDx): A consensus statement by the Genes & Kidney Working Group of the ERA. Nephrology Dialysis Transplantation*

19. Jayasinghe, K., Stark, Z., Kerr, P.G. *et al.* Clinical impact of genomic testing in patients with suspected monogenic kidney disease. *Genet Med* **23**, 183–191 (2021). https://doi.org/10.1038/s41436-020-00963-4

20. Jefferis, J., & Mallett, A. J. (2024). Exploring the impact and utility of genomic sequencing in established CKD. Clinical kidney journal, 17(3), sfae043.

21. Joshi, E., Biddanda, A., Popoola, J., Yakubu, A., Osakwe, O., Attipoe, D., ... & Salako, B. (2023). *Whole-genome sequencing across 449 samples spanning 47 ethnolinguistic groups provides insights into genetic diversity in Nigeria. Cell Genomics, 3*(9). https://doi.org/10.1016/j.xgen.2023.100378

22. Kent, D. M., Steyerberg, E. W., & van Klaveren, D. (2018). *Personalized evidence-based medicine: Predictive approaches to heterogeneous treatment effects. BMJ, 363*, k4245. https://www.bmj.com/content/363/bmj.k4245

23. Lai, J., Shan, H., Cui, S., Xiao, L., Huang, X., & Yun, Y. (2024). *Bioinformatics analysis reveals CCR7 as a potential biomarker for predicting CKD progression. Medicine, 103*(30), e33705.

24. Lew, A. R., Kellermayer, T. R., Sule, B. P., and Szigeti, K. (2018). Copy number variations in adult-onset neuropsychiatric diseases. Curr. Genomics 19, 420–430. doi: 10.2174/1389202919666180330153842

25. Mohd Athar, M., Manhas, A., Rana, N., & Irfan, A. (2024). *Computational and bioinformatics tools for understanding disease mechanisms. Biocell, 48*(5–6), 1–10.

26. Oluniyi, P. E., Ajogbasile, F. V., & Oguzie, J. (2021). *Africa's genomic response to COVID-19 and emerging infectious diseases. Nature Communications, 12*, 4456.

27. Patera, F., Gatticchi, L., Cellini, B., Chiasserini, D., & Reboldi, G. (2024). Kidney fibrosis and oxidative stress: from molecular pathways to new pharmacological opportunities. *Biomolecules*, *14*(1), 137.

28. Recharla, M., Nuka, S. T., Chakilam, C., Chava, K., & Suura, S. R. (2023). *Next-generation technologies for early disease detection and treatment: Harnessing intelligent systems and genetic innovations for improved patient outcomes. Journal of Advanced Biotechnology Research, 10*(s), 1923–1930