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The Role of Genomics in Arthritis Diagnosis in West Africa

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ABSTRACT

Arthritis, a complex group of musculoskeletal disorders characterized by joint inflammation and degeneration, presents significant diagnostic challenges, particularly in resource-limited settings like West Africa. The advent of genomic technologies has revolutionized the understanding of arthritis pathogenesis, offering novel diagnostic tools and biomarkers for early and accurate detection. This review explores the current and potential role of genomics in arthritis diagnosis across West African populations, highlighting the genetic basis of major forms such as rheumatoid arthritis (RA), osteoarthritis (OA), and juvenile idiopathic arthritis (JIA). It examines genetic studies conducted in or relevant to African populations, the implications of population-specific variants, and the integration of genomics with clinical practice. The review also discusses the limitations, ethical concerns, and infrastructural barriers impeding genomic research and application in the region. Finally, it emphasizes the need for increased investment in genomic surveillance, local research capacity, and personalized medicine approaches to enhance arthritis diagnosis and management in West Africa.

Keywords: Genomics, arthritis diagnosis, West Africa, rheumatoid arthritis, osteoarthritis.

INTRODUCTION

Arthritis is a collective term that refers to more than 100 different musculoskeletal conditions that affect the joints, surrounding tissues, and other connective tissues [1]. Among the most common forms are rheumatoid arthritis (RA), an autoimmune disorder that causes chronic inflammation of the joints and osteoarthritis (OA) a degenerative joint disease resulting from the breakdown of joint cartilage and underlying bone [2]. Globally, arthritis is one of the leading causes of disability, significantly impacting quality of life, productivity, and socioeconomic well-being. According to the World Health Organization, hundreds of millions of people suffer from arthritis-related conditions, with a considerable proportion being women and the elderly [3].

In West Africa, arthritis is an increasing public health concern. Factors such as an aging population, urbanization, sedentary lifestyles, obesity, and the rising incidence of non-communicable diseases (such as diabetes and cardiovascular disease) contribute to the growing burden of arthritis in the region [4]. As more people live longer and adopt Western-style dietary and lifestyle patterns, conditions like osteoarthritis and rheumatoid arthritis are becoming more prevalent, placing a significant strain on already under-resourced healthcare systems.

Despite this emerging challenge, arthritis remains underdiagnosed and undertreated in many parts of West Africa. The lack of awareness among both the general public and healthcare professionals, limited access to specialized diagnostic tools, and the shortage of trained rheumatologists and geneticists are major obstacles to timely and accurate diagnosis [5]. In rural and low-income settings, these issues are even more pronounced, often leading to delayed treatment, poor disease management, and severe disability.

In recent years, there has been a growing interest in the application of genomics, the comprehensive study of the full set of genes and their interactions for understanding the pathogenesis, diagnosis, and potential treatment of arthritis [6]. Genomic approaches, such as genome-wide association studies (GWAS), transcriptomic profiling, and whole-genome sequencing, have opened new pathways for identifying genetic variants and biomarkers associated

with arthritis susceptibility, disease progression, and treatment response. These advances have the potential to revolutionize arthritis diagnosis and management, allowing for personalized medicine and targeted therapies [7]. However, West Africa has lagged behind in genomic research, partly due to infrastructural limitations, high costs of genetic testing, ethical and policy-related challenges, and insufficient training in genomics and bioinformatics [8]. This gap underscores the urgency to evaluate and promote the role of genomics in diagnosing arthritis in the region, to help bridge health disparities and improve patient outcomes.

The diagnosis and management of arthritis in West Africa face significant challenges that undermine effective patient care. A major issue is the lack of early and accurate diagnostic tools, especially in rural and resource-limited areas where access to healthcare infrastructure is minimal [9]. Additionally, there is limited awareness and understanding among clinicians and researchers about the genetic factors contributing to arthritis in these populations. This gap is compounded by the absence of large-scale genetic studies specifically targeting West African groups, resulting in a scarcity of region-specific biomarkers or risk models to guide diagnosis and treatment [10]. Furthermore, the high cost and technical complexity of genomic testing act as major barriers to its widespread adoption. Equally problematic is the poor integration of genomic research findings into national health policies and clinical protocols. Collectively, these challenges lead to delayed or incorrect diagnoses, suboptimal treatment approaches, and ultimately, worse health outcomes. Addressing these issues is essential to advance precision medicine and improve arthritis care in West Africa [11]. This study seeks to comprehensively explore the transformative potential of genomics in enhancing arthritis diagnosis within the healthcare systems of West Africa. Central to this investigation are several specific objectives that guide the research process. First, the study aims to assess the current landscape of arthritis diagnosis and management practices, identifying gaps and limitations within existing healthcare infrastructures. Understanding the baseline is critical for contextualizing the value genomics could add. Secondly, it will conduct a detailed review of genomic factors and biomarkers linked to arthritis, particularly focusing on rheumatoid arthritis and osteoarthritis, the two most prevalent and debilitating forms of the disease. This focus allows for identifying key genetic markers and molecular pathways that may serve as targets for early detection and prognosis. Thirdly, the study evaluates how genomics-based diagnostic technologies could improve accuracy and timeliness of arthritis diagnosis in West African populations, where diagnostic delays often lead to poor patient outcomes. Fourthly, the research aims to uncover the infrastructural, economic, social, and ethical barriers hindering the adoption of genomic tools in these settings, including challenges related to resource constraints, lack of trained personnel, and concerns around privacy and consent. Finally, it proposes strategic recommendations for integrating genomics into public health policies and clinical practice to foster personalized, effective arthritis care. Guided by pertinent research questions addressing diagnostic challenges, genetic markers, technology applicability, and policy frameworks, this study holds significant value. It will bridge the existing knowledge gap arising from the predominance of non-African genomic data, thereby tailoring arthritis diagnostics to the genetic profiles of West African populations. Moreover, it aims to improve patient outcomes through early diagnosis, strengthen healthcare systems via targeted capacity-building, and promote collaborative genomic research. Equally important, the study addresses ethical considerations, ensuring culturally sensitive and equitable implementation of genomics in West Africa's healthcare landscape. Collectively, this research lays a vital foundation for harnessing genomic medicine as a powerful tool in combating arthritis and improving health equity in the region.

Genetic Basis of Arthritis

Genetic predisposition is a fundamental factor influencing the development and progression of various types of arthritis, including rheumatoid arthritis (RA), osteoarthritis (OA), and juvenile idiopathic arthritis (JIA) [12]. Research conducted predominantly in European and Asian populations has revealed several key genes linked to these conditions. For instance, the HLA-DRB1 and PTPN22 genes have been strongly associated with increased susceptibility to RA, while genes such as GDF5 and COL2A1 are implicated in OA. Juvenile idiopathic arthritis also has identifiable genetic components, although the exact mechanisms remain under investigation. Despite these advancements, there remains a significant lack of genetic data specific to African populations, including those in West Africa. This gap in knowledge presents a major obstacle to the development of effective, targeted treatments and personalized medicine approaches for arthritis in these populations. African populations are known to exhibit greater genetic diversity compared to other continental groups, which means that findings from non-African studies may not be directly applicable or fully representative. Therefore, it is imperative to conduct population-specific genetic studies within Africa to better understand the unique genetic factors influencing arthritis in these communities [13]. Such research will be crucial for improving diagnosis, management, and therapeutic interventions tailored to the genetic backgrounds of African patients.

Genomic Studies in African and West African Populations

Genomic studies focusing on African and West African populations are gradually unveiling unique genetic factors that influence disease susceptibility, particularly in autoimmune conditions like arthritis. Although research in this area remains limited compared to studies conducted in European or Asian populations, emerging evidence highlights that African-specific genetic variants may play a distinct role in modulating the risk and progression of arthritis [14]. One notable example is the variation in human leukocyte antigen (HLA) alleles, which are key genetic markers involved in immune system regulation. These alleles show significant differences in prevalence and expression patterns between African and non-African populations, potentially explaining divergent autoimmune disease manifestations and outcomes. Addressing the historic underrepresentation of African populations in genomic research, initiatives like the Human Heredity and Health in Africa (H3Africa) consortium have been pivotal in advancing this field. H3Africa provides critical funding and infrastructure to support genomic studies that focus on diseases disproportionately affecting African populations, including rheumatic and autoimmune diseases. Through collaborative efforts across multiple countries and institutions, H3Africa aims to improve understanding of the genetic architecture underlying these conditions, which could lead to more effective, population-specific diagnostic, preventive, and therapeutic strategies. As genomic data from African populations expand, they promise to enrich global knowledge of disease mechanisms and enhance health equity in genomic medicine [15].

Genomic Tools and Biomarkers in Arthritis Diagnosis

Recent advancements in genomic technologies have significantly transformed the landscape of arthritis diagnosis, offering promising alternatives to traditional diagnostic methods. Techniques such as next-generation sequencing (NGS), genome-wide association studies (GWAS), and transcriptomics have facilitated the identification of specific molecular signatures, including gene expression profiles, microRNAs, and epigenetic alterations like DNA methylation patterns, which are increasingly recognized as reliable biomarkers for early arthritis detection [16]. These genomic tools enable clinicians to detect disease onset at a molecular level, often before clinical symptoms become pronounced. In regions like West Africa, where access to conventional diagnostic tools such as advanced imaging and serological testing may be constrained by infrastructure and cost limitations, genomic biomarkers present a valuable opportunity for enhancing diagnostic accuracy. Their potential lies in providing non-invasive, rapid, and individualized diagnostic information, which could be pivotal in initiating timely interventions and improving patient outcomes. However, realizing this potential requires substantial investment in local genomic research, capacity building, and integration into healthcare systems. With appropriate policy support, training of healthcare professionals, and community engagement, the application of genomics in arthritis diagnosis could play a transformative role in addressing diagnostic challenges and advancing personalized medicine in resource-limited settings [17].

Challenges and Barriers in West Africa

The integration of genomics into arthritis diagnosis in West Africa faces numerous challenges that limit its practical application and scalability. Infrastructural limitations are a major concern, as only a few institutions in the region are equipped with advanced laboratories and bioinformatics facilities necessary for conducting comprehensive genomic analyses. This is compounded by funding constraints, given that genomic research requires substantial financial investment and often depends on sporadic external grants rather than consistent local funding mechanisms. Furthermore, there is a significant shortage of skilled professionals such as geneticists, rheumatologists, and molecular biologists, which hampers both research and clinical translation. Ethical and cultural concerns also present substantial barriers; many communities harbor mistrust toward genetic research due to fears around privacy, misuse of genetic information, and inadequate understanding of consent processes [18]. These issues collectively create a complex landscape that restricts the widespread adoption and impact of genomics in arthritis care across West Africa.

Opportunities and Future Directions

To fully harness the transformative potential of genomics in improving arthritis diagnosis in West Africa, a multifaceted and forward-thinking strategy is essential. First, capacity building must be prioritized through substantial investments in genomics and bioinformatics training programs, laboratory infrastructure, and workforce development. Establishing well-equipped research centers and fostering skilled professionals will provide a solid foundation for sustainable progress. Second, collaborative networks at both regional and international levels are vital for accelerating scientific advancement. These partnerships can facilitate the sharing of data, technologies, and best practices, enabling West African researchers and clinicians to tap into global expertise while addressing local health needs [19]. Third, community engagement is a cornerstone of successful implementation. Involving local communities in the research process, ensuring informed consent, and maintaining transparency will build trust, reduce stigma, and enhance the relevance and acceptance of genomic studies. Finally, the integration of genomic insights with clinical data can advance precision medicine offering tailored diagnostic and therapeutic strategies that

reflect the unique genetic diversity of West African populations. Together, these strategies can not only improve early detection and management of arthritis but also position West Africa as a key player in the global genomics landscape, fostering innovation and equity in healthcare delivery.

CONCLUSION

The integration of genomics into arthritis diagnosis holds transformative potential for healthcare in West Africa. With a rising burden of arthritis-related conditions, early and precise diagnosis is crucial to improving patient outcomes. Genomic tools offer a promising alternative to conventional diagnostic methods by identifying genetic markers specific to the West African population, thereby paving the way for more personalized and effective interventions. However, numerous barriers ranging from infrastructural deficits and funding constraints to ethical concerns and limited expertise continue to impede progress. Addressing these challenges through strategic investments in local capacity building, research infrastructure, and cross-border collaborations will be key. Equally important is the need for inclusive, community-informed research that fosters trust and ethical integrity. By leveraging genomics in a culturally sensitive and scientifically robust manner, West Africa can enhance arthritis diagnostics, contribute to global health knowledge, and move closer to achieving equitable, precision-based healthcare solutions.

REFERENCES

1. Alum, E. U. Ugwu, O. P. C. Nutritional Strategies for Rheumatoid Arthritis: Exploring Pathways to Better Management. *INOSR Scientific Research.* 2023; 10(1):18-26. https://doi.org/10.59298/INOSRSR/2023/3.2.47322
2. Chauhan, K., Jandu, J.S., Brent, L.H., Al-Dhahir, M.A.: Rheumatoid Arthritis. In: StatPearls. StatPearls Publishing, Treasure Island (FL) (2025)
3. Ibiam, U. A., Ugwuja, E. I., Aja, P. M., Igwenyi, I. O., Offor, C. E., Orji, O. U., Ezeani N. N, Aloke, C., Egwu, C. O. Antioxidant effect of *Buchholziacoriacea* ethanol leafextract and fractions on Freund's adjuvant-induced arthritis in albino rats: A comparative study. Slovenian Veterinary Research. 2022; 59 (1): 31–45. doi: 10.26873/svr-1150-2022.
4. Gouda, H.N., Charlson, F., Sorsdahl, K., Ahmadzada, S., Ferrari, A.J., Erskine, H., et al: Burden of non-communicable diseases in sub-Saharan Africa, 1990–2017: results from the Global Burden of Disease Study 2017. The Lancet Global Health. 7, e1375–e1387 (2019). https://doi.org/10.1016/S2214-109X(19)30374-2
5. Inchingolo, F., Inchingolo, A.M., Fatone, M.C., Avantario, P., Del Vecchio, G., Pezzolla, C., et al.: Management of Rheumatoid Arthritis in Primary Care: A Scoping Review. International Journal of Environmental Research and Public Health. 21, 662 (2024). https://doi.org/10.3390/ijerph21060662
6. Sharma, S.D., Leung, S.H., Viatte, S.: Genetics of rheumatoid arthritis. Best Practice & Research Clinical Rheumatology. 38, 101968 (2024). https://doi.org/10.1016/j.berh.2024.101968
7. Aloke, C., Ibiam, U. A., Obasi, N. A., Orji, O. U., Ezeani, N. N., et al. Effect of ethanol and aqueous extracts of seed pod of Copaifera salikounda (Heckel) on complete Freund's adjuvant-induced rheumatoid arthritis in rats. J Food Biochem. 2019 Jul;43(7):e12912. doi: 10.1111/jfbc.12912. Epub 2019 May 23. PMID: 31353723.
8. Ivanga, M., Iroungou, B.A., Bigoundou-Nzamba, S., Bisseye, C.: Accelerating cancer genomics research in Sub-Saharan Africa. Front Oncol. 15, 1531799 (2025). https://doi.org/10.3389/fonc.2025.1531799
9. Azevedo, M.J.: The State of Health System(s) in Africa: Challenges and Opportunities. Historical Perspectives on the State of Health and Health Systems in Africa, Volume II. 1–73 (2017). https://doi.org/10.1007/978-3-319-32564-4_1
10. Reay, W.R., Clarke, E.D., Albiñana, C., Hwang, L.-D.: Understanding the Genetic Architecture of Vitamin Status Biomarkers in the Genome-Wide Association Study Era: Biological Insights and Clinical Significance. Advances in Nutrition. 15, 100344 (2024). https://doi.org/10.1016/j.advnut.2024.100344
11. Hanna, M.G., Pantanowitz, L., Jackson, B., Palmer, O., Visweswaran, S., Pantanowitz, J., et al.: Ethical and Bias Considerations in Artificial Intelligence/Machine Learning. Modern Pathology. 38, 100686 (2025). https://doi.org/10.1016/j.modpat.2024.100686
12. Ibiam, U. A., Orji, O. U., Aja, P. M., Ezeani, N. N. and Ekpono, E. U. Anti- Inflammatory Effects of *Buchholzia coriacea* Ethanol Leaf-Extract and Fractions in Freund's Adjuvant-Induced Rheumatoid Arthritic Albino Rats. *Indo American Journal of Pharmaceutical Sciences (IAJPS).* 2018; **5** (7): 6341- 6357. https://doi.org/10.5281/zenodo.1311167.

13. Sibomana, O., Bukuru, J., Saka, S.A., Uwizeyimana, M.G., Kihunyu, A.M., Obianke, A., Damilare, S.O., Bueh, L.T., Agbelemoge, B. of G., Oveh, R.O.: Routine malaria vaccination in Africa: a step toward malaria eradication? Malaria Journal. 24, 1 (2025). https://doi.org/10.1186/s12936-024-05235-z
14. Omotoso, O.E., Teibo, J.O., Atiba, F.A., Oladimeji, T., Adebesin, A.O., Babalghith, A.O.: Bridging the genomic data gap in Africa: implications for global disease burdens. Globalization and Health. 18, 103 (2022). https://doi.org/10.1186/s12992-022-00898-2
15. Ibiam,U. A., Ugwu, O. P. C, Alum, E. U., A Comprehensive Review of Treatment Approaches and Perspectives for Management of Rheumatoid Arthritis. *INOSR Scientific Research.* 2023; 10(1):12-17.https://doi.org/10.59298/INOSRSR/2023/2.2.13322
16. Dave, K., Patel, D., Dave, N., Jain, M.: Genomic strategies for drug repurposing. Journal of the Egyptian National Cancer Institute. 36, 35 (2024). https://doi.org/10.1186/s43046-024-00245-z
17. Khan, A., Barapatre, A.R., Babar, N., Doshi, J., Ghaly, M., Patel, K.G., Nawaz, S., Hasana, U., Khatri, S.P., Pathange, S., Pesaru, A.R., Puvvada, C.S., Billoo, M., Jamil, U.: Genomic medicine and personalized treatment: a narrative review. Ann Med Surg (Lond). 87, 1406–1414 (2025). https://doi.org/10.1097/MS9.0000000000002965
18. Michel G M, Paul F, Camille H, Jean C, Georges H, Philippe R M. *In vitro* studies on the Fc-receptor function of mononuclear phagocytes in rheumatoid arthritis: Relation between the Fc-receptor blockade and the concanavalin A …*Journal of clinical immunology*, *6*, *442-456*.
19. Stephen, S., Aigbavboa, C.: Enhancing Academia–Industry Partnerships for Sustainable Building: A Change Framework for Research and Innovation Integration in Sub-Saharan Africa. Sustainability. 17, 3863 (2025). https://doi.org/10.3390/su17093863

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