

Genomic research on Esophageal Cancer in African populations suboptimal: Funding needed to address disease burden



Source: Image by [Anton Johnson](#) from [Pexels](#)

African Esophageal Cancer Corridor

Esophageal Cancer (EC) is an aggressive and fatal malignancy constituting a major health burden in high incidence areas. It is the 6th most common cause of cancer mortality worldwide.¹ Over 80% of EC cases and deaths are reported in LMI countries.¹ In Africa, its prevalence is disproportionately higher in the African Esophageal Cancer Corridor which stretches from East to Southern Africa.² The mortality to incidence ratio (cancer deaths in relation to new cases) in this region is high (97.2%), showing the fatality of an EC diagnosis.¹ There is ongoing work looking at EC genomics, however little is known about the role of genetic factors in EC development and progression among African populations.²⁻³

Esophageal cancer genomics

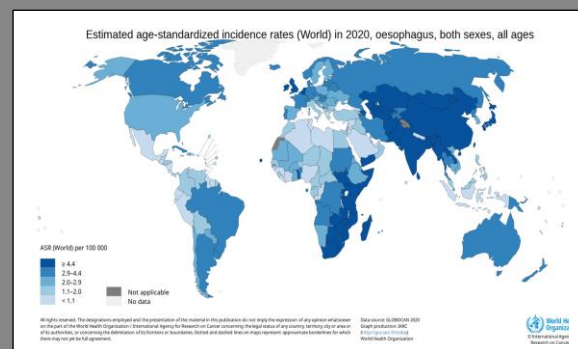
A systematic review² and review³ of EC genetic and genomic studies conducted by Scientists in South Africa, as well as a PhD Thesis⁴ from 2018-2021, revealed a dearth of EC genomic studies in African Populations. This lack leads to a poor understanding of its causes and subsequently renders implementation of genomic medicine for EC elusive. Genomic studies can reveal abnormalities in genes that drive the development and growth of cancer. It can identify variants, pathways and biomarkers associated with increased risk and mortality, to be used in precision medicine.

The lack of financial investment in genomic medicine in Africa has led to most genomic medicine knowledge being founded on genomes of European ancestry, despite African populations displaying higher levels of genetic diversity. Prioritization of EC genomic medicine in African populations will help fill this gap. It will also generate evidence that can be translated to prevention and therapeutics.

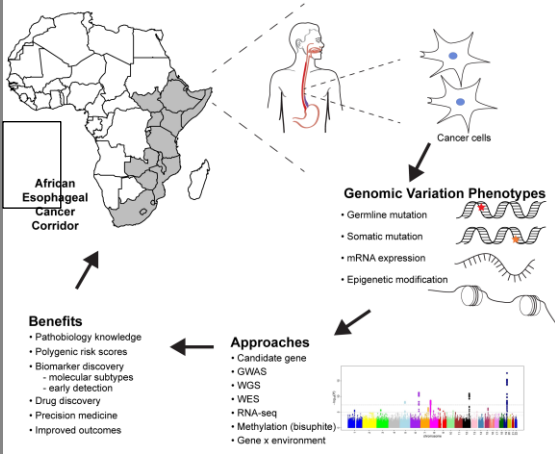
There is therefore an urgent need to invest in genomic research for EC in Africa, to improve health outcomes of affected populations. The BMGF is the most appropriate funder to invest in this important field given its priority to save lives, improve health through disease prevention and treatment in Africa.

The Global Health Division of the Bill and Melinda Gates Foundation should prioritise:

- 1** **Funding** studies looking at Esophageal Cancer genomics research in Africa
- 2** **Funding** Bioinformatics training in Africa and genomic medicine training programs for health professionals
- 3** **Leveraging** existing collaborations such as the African Esophageal Cancer Consortium (AfrECC)⁵, Evolving Risk Factors for Cancers in African Populations (ERICA-SA)⁶ study, Johannesburg Cancer Study⁷, and the South African Society for Bioinformatics (SASBi) to facilitate Esophageal Cancer genomics research in Africa



Esophageal cancer incidence rates for 2020 Source : GLOBOCAN ¹



Future of esophageal cancer genomic research in Africa. Source: Simba et al 2022³

Implications

Why is funding EC genomics research important?

Genomics is an invaluable approach in providing information that could be used to predict cancer risk through biomarker discovery, screen asymptomatic individuals, diagnose more accurately and develop targeted treatments. Without adequate financial investment, we will remain far from being able to implement genomic medicine for EC in Africa. This means the EC will remain a health burden in high incidence areas. Funding for EC genomic research will allow for more studies on EC, create a critical mass of African bioinformaticians, and health professionals well versed in genomic medicine.

African genomes in genomic medicine

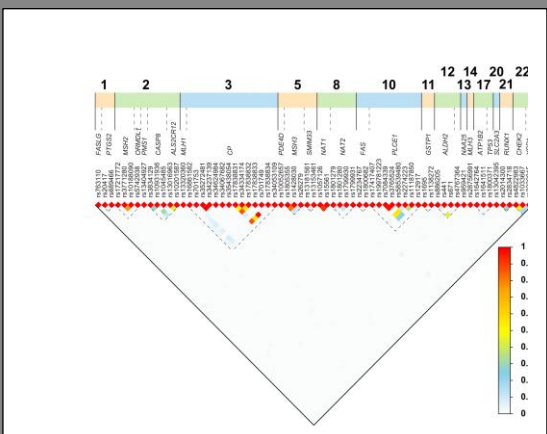
It is important to note that African genomes harbor the most genetic diversity and variation, and yet are the least genetically characterized. Genetic variants of medical relevance therefore remain unknown. For EC this impedes progress on applying genomics in understanding cancer development, tailored screening, and therapeutic interventions, promoting health equity and ultimately reducing the burden of EC in the African Esophageal Cancer corridor.

What are the benefits?

Furthering genomic medicine in Africa requires leveraging existing infrastructure, collaborations learning from the extensive experience of current genomic medicine implementations in other countries. The BMBF can partner with existing collaborations currently working of EC research in Africa mainly under the AfrECC consortium umbrella. \$USD 2 million is needed to support this research for 5 years. Funding will also address the health disparities that currently existing due to Esophageal Cancer genomic medicine lagging in Africa. This is line with the BMGF commitment to tackle inequities in the world. This is therefore an opportunity for the BMBF to use genomics as a tool to address health disparities in EC, bring long awaited answers to this EC hotspot region and reduce the EC disease burden.

Key Findings

- 1 There is lack of genomic studies on Esophageal Cancer in African populations
- 2 The majority of the existing studies are not generalisable and difficult to ascertain true effect.
- 3 There is a lack of financial investment needed to further genomic medicine research and capacity building on Esophageal Cancer in high incidence areas of Africa



Linkage disequilibrium plot showing the number (n=77) of DNA variants (SNPs) investigated in African studies. Simba. Source: Simba et al 2019²

References

1. Ferlay, J. et al (2021). Cancer statistics for the year 2020: An overview. International journal of cancer, 149(4), 778-789.
2. Simba, H., et al (2019). Systematic Review of Genetic Factors in the Etiology of Esophageal Squamous Cell Carcinoma in African Populations. Front. Genet. 10, 642.
3. Simba H, et al (2022) Esophageal Cancer Genomics in Africa: Recommendations for Future Research. Front Genet. 25;13:864575.
4. Simba, H. et al (2021). The Role of Genetic and Environmental Factors in the Aetiology of Esophageal Cancer PhD (Public Health) Stellenbosch University. <https://scholar.sun.ac.za/handle/10019.1/123653>.
5. Van Loon, K et al. (2018). The African Esophageal Cancer Consortium: A Call to Action. Jgo, 1–9
6. SAMRC. Evolving Risk Factors for Cancers in African Populations (ERICA-SA). <https://www.samrc.ac.za/intramural-research-units/evolving-risk-factors-cancers-african-populations-erica-sa> (2021).
7. Chen, W. C., et al. (2020). Johannesburg Cancer Study (JCS): Contribution to Knowledge and Opportunities Arising from 20 Years of Data Collection in an African Setting. Cancer Epidemiol. 65, 101701.

This Issue Brief was developed with support from the Center for Evidence based Health Care (CEBHC) at Stellenbosch University. Credits: Contributors include Hannah Simba, Helena Kuivaniemi, Gerard Tromp, and Nasreen Jessani



Contact:

Name: Dr Hannah Simba
 Email: simbah@iarc.who.int
 Address: Department of Global Health, FMHS, Stellenbosch University, South Africa / ENV Branch, International Agency for Research on Cancer (IARC-WHO), France
 Phone: +33 76 66 89 02 55