Genetics in Medicine Open (2024) 2, 101879

ELSEVIER

Genetics in Medicine OPEN
An Official Journal of the ACMG

www.journals.elsevier.com/genetics-in-medicine-open

SPECIAL ARTICLE

"A Call To Action": The need for genetic counseling in Uganda

Tessa Adžemović[1,2,3,*], Kisakye Diana Kabbale[3], Eric Katagirya[4], John Mukisa[4], Misaki Wayengera[4]

[1]Department of Medicine, Brigham and Women's Hospital, Boston, MA; [2]Department of Pediatrics, Boston Children's Hospital, Boston, MA; [3]Infectious Disease Institute; [4]Dept of Immunology and Molecular Biology, Makerere University College of Health Sciences, Kampala, Uganda

ARTICLE INFO

Article history:
Received 4 February 2024
Received in revised form
22 July 2024
Accepted 24 July 2024
Available online 30 July 2024

Keywords:
Genetic counseling in Oman
Genetic counseling licensing
Scope of practice
Consanguinity
Genetic preventative measures

Introduction

Genetic conditions and congenital anomalies affect nearly 8 million children across the world, with approximately 94% of all births affected with congenital anomalies taking place in low- and middle-income countries (LMICs).[1] Importantly, these congenital abnormalities are a large contributor to under-5 mortality worldwide. In Uganda, a country in Africa with an estimated population of greater than 45 million, congenital anomalies are thought to affect 11.7% of neonatal deaths.[2,3]

However, despite the high maternal newborn and child mortality rate in Uganda, the exact burden of congenital and genetic disease is not currently well understood because of a paucity of data. The Unit of Genetics within the Department of Pathology at the College of Health Sciences at Makerere University, in the capital city of Kampala, was established in 1983 and serves as a referral center for children born with rare genetic disorders. Children and adults born with suspected genetic disorders are referred to the unit from specialized neurology, endocrine, neonatal, and pediatric outpatient clinics. The most common genetic disorders seen are hemoglobinopathies (sickle cell anemia and thalassemia), disorders of pigmentation (oculocutaneous albinism and vitiligo), disorders of sexual development, trisomy disorders, and spinal muscular atrophy. This unit is run by a single clinical geneticist, assisted by 3 medical research fellows, who provide medical genetics services, including limited genetic counseling to patients. The care of these patients in the clinic is greatly limited because of the lack of formalized genetic counseling in Uganda. For the purpose of this article, we use the National Society of Genetic Counselors definition of genetic counseling, which states that it is "the process of helping people understand and adapt to the medical, psychological, and familial implications of the genetic contributions to disease."[4]

The Article Publishing Charge (APC) for this article was paid by Tessa Adžemović.

Tessa Adžemović and Kisakye Diana Kabbale contributed equally to this work.

*Correspondence and requests for materials should be addressed to Tessa Adzemovic, Brigham and Women's Hospital, 75 Frances St PBB-B 428, Boston, MA 02115. *Email address:* tessa.adzemovic@gmail.com

doi: https://doi.org/10.1016/j.gimo.2024.101879

With increased recognition of the importance of genomic medicine in LMICs, significant strides have been made to increase inclusion of African populations in genomics and genetics research. The Human Heredity and Health in Africa consortium has been the largest and most collaborative initiative to address the under-representation of the sub-Saharan African population in genetics research to date.[5] The consortium has thus provided the platform for Uganda's involvement in population-based genetics and genomics research for both noncommunicable and communicable diseases.[6] Uganda has also benefited from augmented technical capacity in genomics, including the establishment of genomics laboratories to support genomics-related research. With large-scale advancements in genomic technologies and a surge in genomic studies taking place in LMICs, the fundamental ethical and legal framework to guide the implementation of human genetics and genomics research remains a neglected area.[7] One key component of this implementation is the field of genetic counseling.

Here, we outline the need for genetic counselors in Uganda, in both clinical genetic services and research initiatives, which go hand in hand in this growing and dynamic field. We review this critical need and suggest approaches to establishing the genetic counseling profession in Uganda.

Current state of genetic counseling in Uganda

Although 94% percent of births with severe congenital anomalies happen in LMICs, paradoxically, these countries also have the fewest genetic counselors.[8] It is known that genetic counseling has a crucial role to play in performing genetic risk assessments, educating patients on their illnesses, and providing information on genetic testing and in the interpretation of results from genetic tests.

Despite their universal importance to the field of human genetics and genomics research, almost all genetic counselors are situated in North America and Western Europe, with few providers in other areas.[9] At this time, South Africa is the only country in Africa that has genetic counselors, with no accredited genetic counselors in Uganda.[7,10]

The absence of genetic counselors in Uganda has created a void when it comes to access and the interpretation of family and medical histories, and results from genetics tests. Without this workforce, there continues to be a knowledge gap and power differential that afflicts the most vulnerable populations. Uganda currently has 2 practicing clinical geneticists with formal training in genetic counseling, one of whom works at the Unit of Genetics and Genomics. Efforts to train health care workers in genetic counseling services in Uganda have been minimal, including a 1-week training for a small cohort of clinicians and nurses, supported by Human Heredity and Health in Africa in 2019.[11] In contrast, in North America and many parts of Western Europe, genetic counseling training is at master's level. In Africa, at the time of writing, Masters of Science programs in genetic counseling only exist in South Africa and Ghana.

We describe below the crucial role of genetic counselors and why they are important for the health of Uganda's people, in the domains of interpretation, education, and counseling as detailed by the National Society of Genetic Counselors.

Interpretation

Genetic counseling is tasked with the "interpretation of family and medical histories to assess the chance of disease occurrence or recurrence."[4]

We use the examples of sickle cell disease and familial cancer syndromes in Uganda to illustrate the importance of interpretation performed by genetic counselors. It is approximated that 75% of global annual sickle cell births occur in Sub-Saharan Africa. In Uganda, there is a high national burden of sickle cell anemia, estimated at 0.9%, with some postulating that it is one of the countries with the highest sickle cell burden in the world.[12,13] To date, Uganda lacks a universal newborn screening program for sickle cell anemia, although it does have cost-effective screening that is increasing in scope. Screening for sickle cell disease was initiated for a subset of infants exposed to HIV, in high-burden districts, under the Early Infant Diagnosis Program for HIV.[14] Recent initiatives within the Ugandan Ministry of Health are pushing for universal screening programs.[15] Moreover, because of the high burden of sickle cell disease in Uganda, prospects for gene therapy trials are emerging, which could potentially save and prolong the lives of patients living with the disease.

Sickle cell disease is a condition in which interpretation of the patient's family history and education on inheritance is paramount to care. Because individuals with sickle cell trait are at risk of having a child with sickle cell disease, prenatal and postnatal genetic counseling is of great importance.[16] We argue that screening programs and gene therapy cannot be appropriately nor adequately implemented without the use of genetic counselors to discuss the condition, its inheritance pattern, the risks of therapy, and the psychosocial needs of the patient.

Another important example of the necessity of interpretation of family and medical histories is in the setting of familial cancer syndromes. Understanding one's hereditary predisposition to cancer can drive early screening and detection. A recent study in Uganda reported a high prevalence of inherited breast cancer, arguing for the integration of genetic counseling in breast cancer and other cancer screening programs.[17]

Education

The second task of genetic counseling is, "education about inheritance, testing, management, prevention, resources, and research."[4] The cornerstone of genetic counseling is educating the patient and family on the disease in question. With most clinical conditions, there is significant trepidation

associated with a diagnosis. The task of education is both challenging and broad. A genetic counselor must educate the patient/family on inheritance pattern, risks of recurrence of the disease process, medical management and surveillance, prognosis, reproductive options, and resources for support, financial assistance, and educational needs. Education provided by genetic counselors is its own artform because it requires technical skills (comprehension of the medical genetics/genomics), as well as teamwork, emotional intelligence (an understanding of health literacy and competency), and good communication skills. Given that genetic counseling is a niche and complex specialty, we argue that nonspecialized counselors, used in many genomic studies in Uganda, cannot fulfill the mandate of genetics counselors to provide contextual education.

Disorders of sexual development, such as androgen insensitivity syndrome, an X-linked condition, are frequently encountered in the genetics clinic. Dissemination of the results from such genetics tests can only be done by a provider with training in genetic counseling who is equipped to provide information about the mode of inheritance, risk of the recurrence in another pregnancy, refer the cases for specialized surgical and medical interventions, and offer psychosocial support.

Additionally, Uganda has seen the increased uptake of assisted reproductive technology (ART) over the past decade. Culturally, fertility holds great importance in traditional Ugandan society. Genetic counseling and education before ART is important because undiagnosed genetic disorders may contribute to infertility. Second, education of individuals during ART to provide information on genetic tests performed on the embryo before the implantation of embryos is crucial.

Genetic counselors also play an integral role in the interpretation and dissemination of research results in the Global North; however, there remains little instruction on how to disseminate Genetics and Genomic Research results to research participants in Africa.[18] Not surprisingly, there is little to no dissemination of genetic results to participants, despite research to suggest that participants would like their results to be returned to them.[19] Providers cite feeling that genetic research results should be returned, as long as it is done by a trained provider in a culturally sensitive way.[20] Within the sole genetics clinic in Uganda, return of results is currently done by a clinical geneticist and 3 research fellows who have training in genetics and some aspects of genetic counseling; however, in most settings in Uganda, it is unknown whether providers partaking in genomics research have undergone any formal training in genetics.

Counseling

Lastly, we discuss "counseling to promote informed choices and adaptation to the risk or condition," which is contingent on the empathetic provision of guidance from a client-centered perspective.[4]

Three things need to be considered when counseling a patient on their genetic condition: competency, support, and empathy. With regard to competency, it is assumed the individual is capable of caring for their own emotional well-being when provided with accurate information in the presence of psychological and emotional support. The counselor must provide support by assisting individuals and their families in coping with the difficulties of a new diagnosis and in facing dilemmas. Lastly, they must provide empathy, be nonjudgmental and respect autonomy.

Recently, there has been an increased demand for paternity testing services in Uganda. Unexpected results have led to confusion and considerable grief, as well as psychosocial distress.[20] Because of the absence of genetic counselors, individuals performing paternity tests and receiving results do not have access to adequate support, leading to confusion, sadness, and insecurity, not only for individuals but also for families.

Certain tribal populations in Uganda continue to partake in consanguineous relations. Education on the consequences of consanguineous marriages through genetic counseling is therefore a key step to recognizing and mitigating the risks of consanguineous births, which can have devastating consequences.[21]

Challenges

Of course, the implementation of genetic counseling in Uganda is not without its challenges. Efforts to introduce genetic counseling in Ethiopia, for example, have identified implementation and sustainability as major barriers to the initiation of genetic counseling in similar environments.[1]

First as detailed above, a primary challenge in Uganda will be the lack of adequate funding in the setting of a volatile economy and stagnant economic policy. Second, although the adult literacy rate in Uganda has greatly improved, approximately 1 in 5 adults are still illiterate. Genetics and genomics is a new and complex field in Uganda that will require the translation and interpretation of genetic terms across many different dialects. Third, cultural values and beliefs are a key component of genetic counseling. Given the ethnic, socioeconomic, and religious diversity of Uganda, providers will have to be adept in navigating varying beliefs.

The way forward

Uganda's health care system currently prioritizes communicable diseases, and largely overlooks noncommunicable diseases, including genetic disorders, as outlined above. Simultaneously, this increasing prevalence of noncommunicable diseases is concurrent with mortality that "outstrips existing health investments."[22] The paucity of genetic counselors in Uganda is directly related to the low health budget for noncommunicable diseases.[23] A crucial

first step is the allocation of funding for the essential field of genetic counseling, and moreover, genetic counselors.

With this additional funding, there must be certain priorities for growing genetic counseling in Uganda. First, we propose the establishment of accredited genetic counseling degree-training programs to grow the workforce to meet the needs in Uganda. Second, we recommend the creation of regional and national regulatory bodies that will provide support and supervision and ensure consistency of genetic counseling across providers in Uganda. Moreover, given the necessity of genetic counselors in ongoing genomics research, we assert that research budgets should inherently include funding for genetic counselors.

Given that these interventions will take adequate infrastructure and long-term capacity building, in the interim, we recommend an intermediate solution with provision of genetic counseling courses for those providers already providing clinical genetics services or involved in human genomics research.

Conclusion

We have highlighted the critical need to introduce the field of genetic counseling in the era of the genomics revolution in Uganda both for the betterment of clinical care, as well as for the ethical conduction of research. We propose the Unit of Genetics at Makerere University as an initial platform to provide training for future genetic counselors. As the global burden of noncommunicable diseases continues to grow and childhood mortality due to communicable diseases continues to decrease, Uganda will inevitably continue to see an increase in patients with genetic conditions. Therefore it is imperative that genetic counseling is urgently integrated into Uganda's health care system.

Data Availability

The authors are happy to supply data or materials on request.

Acknowledgments

The authors thank the Unit of Genetics within the Department of Pathology at the College of Health Sciences at Makerere University and all the patients whom they have cared for.

Funding

Tessa Adžemović was supported by the National Institute of Neurological Disorders and Stroke and the Fogarty International Center of the National Institutes of Health under grant #D43TW009345 awarded to the Northern Pacific Global Health Fellows Program.

Author Contributions

Conceptualization: K.D.K., T.A., M.W.; Methodology: K.D.K., T.A.; Writing-original draft: K.D.K., T.A.; Writing-review and editing: M.W., E.K., J.M.

Ethics Declaration

This paper does not involve human subjects and did not require Institutional Review Board approval.

Conflict of Interest

The authors have no conflicts of interest.

References

1. Quinonez SC, O'Connor BC, Jacobs MF, et al. The introduction of genetic counseling in Ethiopia: results of a training workshop and lessons learned. *PLoS One*. 2021;16(7):e0255278. http://doi.org/10.1371/journal.pone.0255278
2. Uganda profile. Uganda Bureau of Statistics. Accessed February 1, 2024. https://www.ubos.org/uganda-profile/
3. Maternal and newborn health disparities. Uganda. Accessed May 1, 2024. https://data.unicef.orgcountryprofile_UGA
4. National Society of Genetic Counselors' Definition Task Force, Resta R, Biesecker BB, et al. A new definition of Genetic Counseling: National Society of Genetic Counselors' Task Force report. *J Genet Couns*. 2006;15(2):77-83. http://doi.org/10.1007/s10897-005-9014-3
5. Adoga MP, Fatumo SA, Agwale SM. H3Africa: a tipping point for a revolution in bioinformatics, genomics and health research in Africa. *Source Code Biol Med*. 2014;9:10. http://doi.org/10.1186/1751-0473-9-10
6. Fatumo S, Mugisha J, Soremekun OS, et al. Uganda Genome Resource: a rich research database for genomic studies of communicable and non-communicable diseases in Africa. *Cell Genom*. 2022;2(11):None. http://doi.org/10.1016/j.xgen.2022.100209
7. Ochieng J, Kwagala B, Barugahare J, et al. Perspectives and ethical considerations for return of genetics and genomics research results: a qualitative study of genomics researchers in Uganda. *BMC Med Ethics*. 2021;22(1):154. http://doi.org/10.1186/s12910-021-00724-1
8. Congenital anomalies: fact sheet N° 370. World Health Organization. Accessed February 1, 2024. http://www.who.int/mediacentre/factsheets/fs370/en/
9. Ormond KE, Laurino MY, Barlow-Stewart K, et al. Genetic counseling globally: where are we now? *Am J Med Genet C Semin Med Genet*. 2018;178(1):98-107. http://doi.org/10.1002/ajmg.c.31607
10. Abacan M, Alsubaie L, Barlow-Stewart K, et al. The global state of the genetic counseling profession. *Eur J Hum Genet*. 2019;27(2):183-197. http://doi.org/10.1038/s41431-018-0252-x
11. Nembaware V, African Genomic Medicine Training Initiative, Mulder N. The African genomic medicine training initiative (AGMT):

showcasing a community and framework driven genomic medicine training for nurses in Africa. *Front Genet*. 2019;10:1209. http://doi.org/10.3389/fgene.2019.01209
12. Hernandez AG, Kiyaga C, Howard TA, et al. Trends in sickle cell trait and disease screening in the Republic of Uganda, 2014-2019. *Trop Med Int Health*. 2021;26(1):23-32. http://doi.org/10.1111/tmi.13506
13. Piel FB, Patil AP, Howes RE, et al. Global epidemiology of sickle haemoglobin in neonates: a contemporary geostatistical model-based map and population estimates. *Lancet*. 2013;381(9861):142-151. http://doi.org/10.1016/S0140-6736(12)61229-X
14. Mvundura M, Kiyaga C, Metzler M, et al. Cost for sickle cell disease screening using isoelectric focusing with dried blood spot samples and estimation of price thresholds for a point-of-care test in Uganda. *J Blood Med*. 2019;10:59-67. http://doi.org/10.2147/JBM.S186528
15. Hernandez AG, Kiyaga C, Howard TA, et al. Operational analysis of the national sickle cell screening programme in the Republic of Uganda. *Afr J Lab Med*. 2021;10(1):1303. http://doi.org/10.4102/ajlm.v10i1.1303
16. Pecker LH, Naik RP. The current state of sickle cell trait: implications for reproductive and genetic counseling. *Blood*. 2018;132(22):2331-2338. http://doi.org/10.1182/blood-2018-06-848705
17. Adedokun B, Zheng Y, Ndom P, et al. Prevalence of inherited mutations in breast cancer predisposition genes among women in Uganda and Cameroon. *Cancer Epidemiol Biomarkers Prev*. 2020;29(2):359-367. http://doi.org/10.1158/1055-9965.EPI-19-0506
18. H3 Africa website. Accessed January 29, 2024. https://h3africa.org/wp-content/uploads/2018/05/H3Africa%20Feedback%20of%20Individual%20Genetic%20Results%20Policy.pdf>
19. Nabukenya S, Waitt C, Senyonga R, et al. Research participants' preferences for individual results of pharmacogenomics research: a case of a Ugandan HIV Research Institute. *J Empir Res Hum Res Ethics*. 2023;18(4):218-232. http://doi.org/10.1177/15562646231187434
20. Rutakumwa R, de Vries J, Parker M, Tindana P, Mweemba O, Seeley J. What constitutes good ethical practice in genomic research in Africa? Perspectives of participants in a genomic research study in Uganda. *Glob Bioeth*. 2019;31(1):169-183. http://doi.org/10.1080/11287462.2019.1592867
21. Sereke SG, Berhe SE, Bongomin F. Harlequin fetus: a mayhem in a consanguineous marriage? *Clin Case Rep*. 2021;9(7):e04540. http://doi.org/10.1002/ccr3.4540
22. Govt permits only 2 labs to do DNA tests. Daily Monitor. Accessed February 1, 2024. https://www.monitor.co.ug/uganda/news/national/govt-permits-only-2-labs-to-do-dna-tests-4299846
23. Third national development plan (NDPIII) 2020/21 – 2024/25. Ministry of Health Republic of Uganda. Accessed February 1, 2024. https://www.health.go.ug/cause/third-national-development-plan-ndpiii-2020-21-2024-25/