Public Health Genomics. Author manuscript; available in PMC 2018 January 17.

Published in final edited form as:

Public Health Genomics. 2014 ; 17(4): 221–227. doi:10.1159/000363645.

Developing Clinical Cancer Genetics Services in Resource-Limited Countries: The Case of Retinoblastoma in Kenya

Li Qun He[a], **Lucy Njambi**[e], **Joseph M. Nyamori**[e], **Emmanuel M. Nyenze**[e], **Kahaki Kimani**[e], **Ibrahim Matende**[f], **Hilary Rono**[h], **Victor Njom**[g], **James Bett**[i], **Muchai Gachago**[e], **Mukiri Mukuria**[e], **Helen Roberts**[j], and **Helen Dimaras**[b,c,d]

[a]Human Biology Program, University of Toronto, Toronto, Ont., Canada

[b]Department of Ophthalmology and Vision Sciences, University of Toronto, Toronto, Ont., Canada

[c]Division of Visual Sciences, Toronto Western Research Institute, Toronto, Ont., Canada

[d]Department of Ophthalmology and Vision Sciences, The Hospital for Sick Children, Toronto, Ont., Canada

[e]Department of Ophthalmology, University of Nairobi, Nairobi, Kenya

[f]Lighthouse Eye Centre, Mombasa, Kenya

[g]Coast Provincial General Hospital, The Ministry of Health, Mombasa, Kenya

[h]Zonal Eye Surgeon (North Rift) and Kitale District Eye Hospital, Kitale, Kenya

[i]Moi Teaching and Referral Hospital, Eldoret, Kenya

[j]Kwale District Eye Centre, Kwale, Kenya

Abstract

Background/Aims—Clinical cancer genetics is an integral part of cancer control and management, yet its development as an essential medical service has been hindered in many low-and-middle-income countries. We report our experiences in developing a clinical cancer genetics service for retinoblastoma in Kenya.

Methods—A genetics task force was created from within the membership of the existing Kenyan National Retinoblastoma Strategy group. The task force engaged in multiple in-person and telephone discussions, delineating experiences, opinions and suggestions for an evidence-based, culturally sensitive retinoblastoma genetics service. Discussions were recorded and thematically categorized to develop a strategy for the design and implementation of a national retinoblastoma clinical genetics service.

Results—Discussion among the retinoblastoma genetics task force supported the development of a comprehensive genetics service that rests on 3 pillars: (1) patient and family counseling, (2) community involvement, and (3) medical education.

Helen Dimaras, Department of Ophthalmology and Vision Sciences, The Hospital for Sick Children, 555 University Avenue, Room 7260, Toronto, ON M5G 1X8 (Canada), helen.dimaras@utoronto.ca.

Conclusions—A coordinated national retinoblastoma genetics task force led to the creation of a unique and relevant approach to delivering comprehensive and accurate genetic care to Kenyan retinoblastoma patients. The task force aims to stimulate innovative approaches in cancer genetics research, education and knowledge translation, taking advantage of unique opportunities offered in the African context.

Keywords

Africa; Cancer; Clinical genetics; Genetic counseling; Genetic testing; Retinoblastoma

Introduction

Clinical cancer genetics is an integral part of cancer control and management [1], yet its development as an essential medical service has been hindered in many low-and-middle-income countries [2, 3] because of poverty, overburdened healthcare systems, and a focus on other significant health concerns [4]. For at-risk individuals to benefit from rapidly advancing cancer genetics technologies, clinicians must communicate and counsel effectively about genetics. However, there is a lack of guidance on effective patient genetic education and counseling, especially within the unique sociocultural context of African countries [3, 5].

Childhood eye cancer retinoblastoma was the first cancer for which a causal genetic mutation was discovered [6]. This seminal discovery initiated a course for genetic healthcare that has become a prototype for the management of other cancers and inherited disorders. A diagnosis of retinoblastoma in a family suggests that other family members may be at risk. Genetic counseling educates families on their cancer risks, and when molecular genetic testing is available, a more refined picture of risk emerges [7]. Testing of parents and siblings for this exact mutation can confirm or eliminate their risk [7]. The effective translation of genetic information supports at-risk individuals to comply with regular cancer surveillance for themselves and their at-risk offspring. It also empowers individuals to make informed reproductive decisions and follow a healthy lifestyle to mitigate second cancer risks. However, this important part of care is arguably more difficult to provide in low-resourced settings.

A Strategy for Retinoblastoma Genetics in Kenya

Since its inception in 2008, the multidisciplinary Kenyan National Retinoblastoma Strategy (KNRbS) group aims to optimize retinoblastoma care in eastern Africa [8]. Members include clinicians and community healthcare workers, policymakers and patient families, among others. The group meets annually to discuss and plan actions to comprehensively address retinoblastoma medically and socially, and to hold in-depth workshops on treatment, pathology and genetics. These efforts are supported largely through the efforts of the nongovernmental organization Daisy's Eye Cancer Fund (www.daisyfund.org). Specialty task forces within the KNRbS group meet on an ad hoc basis between formal annual meetings to advance efforts towards specific programs and projects. One major endeavor of the KNRbS group is to build a comprehensive, nationwide genetic testing and counseling

program for retinoblastoma. Recommendations for genetic counseling in the presence and absence of testing were developed and adopted in the first clinical care guidelines, which have been endorsed by the Kenyan Ministry of Medical Services (in press).

The work described in this paper is now our response to implement the guidelines that had been agreed upon. We initiated a series of in-person and telephone discussions among 35 members of the KNRbS genetics task force. Participants offered experiences, opinions and suggestions for the development of an evidence-based, culturally sensitive and effective retinoblastoma genetics service. Discussions were recorded and thematically categorized to develop a strategy for the design and implementation of a national retinoblastoma clinical genetics service.

Challenges and Opportunities for Clinical Cancer Genetics Services in Kenya

The incidence of retinoblastoma in Kenya is consistent with global published figures, at 1/17,030 live births [9]. Most children have advanced disease at presentation [9], and the cumulative 3-year survival is 26.6% [10]. At the country's main referral hospital, approximately 60–80% of ophthalmology ward beds are occupied by retinoblastoma patients [11]. To our knowledge, no data on the national burden of genetic conditions and birth defects are available for Kenya.

Communication between clinicians and parents at retinoblastoma diagnosis is laden with emotions: parents are told about the malignant nature of the disease, many are advised that the removal of their child's eye(s) is necessary to save life, palliative treatment is initiated as needed. Genetic counseling takes place in this eventful, fast-paced and stressful setting. In Kenya, there is no genetic counseling discipline to support the treating healthcare team. The physician must inquire about the family history of cancer and/or eye conditions, examine parents and young siblings of the diagnosed child, and inform parents about the cancer risks for future pregnancies, usually in the absence of genetic testing for retinoblastoma, which gives more detailed information on the diagnosis. Although molecular diagnostic services are offered by some private Kenyan laboratories, retinoblastoma genetic testing is not available. Kenyan families who have accessed such testing have done so privately, at their own expense, via international laboratories.

In the absence of formalized genetic training or a counseling protocol, physicians provide counseling based on their own knowledge and styles. In turn, families come from diverse educational, religious and cultural backgrounds, influencing how genetic information is understood, internalized and used. This is further complicated by competing views and information families receive in their communities. The common challenges we have faced in Kenya are outlined in table 1.

To address these challenges, we support the development of a comprehensive genetics service that rests on 3 pillars: (1) patient and family counseling, (2) community involvement, and (3) medical education. Program design and research directions are described for each pillar and summarized in table 1.

Patient and Family Counseling

Counseling families about the nature, cause and genetic implications of retinoblastoma is complex, and creative counseling methods are needed to facilitate true understanding by parents (fig. 1). Simplifying the message, perhaps by using metaphors related to the daily experiences of counselees, may be one effective approach. The use of culturally relevant terminology can increase understanding and potentially reduce stigma associated with heritable diseases [12]. For rural families, concepts of inheritance can be explained by using farming metaphors, such as disease resistance in crops or livestock [13]. Focus groups of survivors and patient families will delineate the appropriate terminology for counseling. Actively exploring the origin and reasoning behind an individual's belief can provide cues to strategically refute or incorporate these into counseling, without disrespecting or discounting personal beliefs [14]. Counselors specialized in psychosocial support could be recruited to help patient families cope with feelings of guilt and stress. Those counseling should be mindful of the choice of words used, as they may have implications that undermine the initial aim of counseling (table 1). Repeated counseling over a longer time period, rather than one intensive session at diagnosis, is ideal for sustained understanding and compliance.

A wide range of research efforts and tool development can aid clinicians in providing effective genetic counseling. Various tools, from picture-rich pamphlets to more advanced e-health solutions, can be explored for their efficacy in supporting knowledge uptake and use. It is also crucial to develop tools that evaluate if counseling has provided accurate information and provided assistance to dispel fear, confusion or blame. Incorporating a user-focused approach, to find out what challenges might be faced at home, may help develop a Kenyan structure of counseling that is iterative and more effectively addresses these challenges. We are undertaking such research activities to refine our approach to counseling and achieve optimal outcomes for patients and their families.

Community Involvement

In many African societies, information is often disseminated orally through social circles. This means that the advice of family, elders or community leaders may supersede genetic counseling information, even if it is effectively explained and understood by parents (table 1). We aim to expand the scope of genetic counseling by involving local opinion leaders (e.g. community health workers, village chiefs, and traditional healers [13, 14]) to support patient families with their genetic diagnosis. Previous research on the knowledge, attitudes and practices of traditional healers and eye conditions in Kenya indicated that 55.5% of traditional healers surveyed would opt to refer ocular tumor cases to hospitals [15]. It follows then that this observed tendency to refer patients could be nurtured to expand on the traditional understanding of genetic conditions (i.e. 'disease in the blood') and improve collaboration on the genetic follow-up of families. Further research to determine how local opinion leaders could best complement the counseling approach, including how their current beliefs mesh with genetic concepts, is necessary (table 1). Parents of retinoblastoma patients in Kenya have expressed interest in developing a peer-to-peer genetics education program, where more 'senior' parents advise and support parents of newly diagnosed children. This

'buddy-system' could be key to ensuring the retention of genetic information and compliance with the screening of at-risk offspring.

Kenya is experimenting with a national community strategy approach that may be relevant to the implementation of our clinical genetics service [16]. In this scheme, a single health worker is assigned as the primary contact for 20 households and is the first link in a cascade of health facilities (i.e. from the local dispensary to the tertiary referral center) to ensure the families have access to care and are not lost to follow-up. The strategy has been shown to improve practices in maternal and neonatal care, and it suggests that empowering communities can result in significant health gains [17]. Thus, the 'first-link' health worker for each retinoblastoma-affected family could be recruited and trained to (1) provide complementary genetic counseling for families, (2) ensure that parents are aware of the correct referral pathway to access the care they require, and (3) connect the family with social assistance programs, where available, that remove barriers to such care.

Medical Education

There is an urgent need to strengthen genetic education in low-and-middle-income countries at all levels of medical training [2, 18]. In Kenya, to our knowledge, there are no specialized training programs or recognized independent careers for medical geneticists or genetic counselors. However, as the need for genetic services and professionals is growing [19], this is an opportune time to shape the direction of medical training in genetic counseling and testing. The development of specialized academic curricula with practical exposure is paramount. Postgraduate research interest and participation in basic genetics will advance the knowledge of the molecular development of retinoblastoma, and possibly new therapies. There are no genetic testing services for retinoblastoma in Kenya. If a family has the financial means to do so, they may access testing available outside the country; however, this is rare. A global-to-local approach for retinoblastoma gene testing, in which laboratories with global expertise partner with local labs seeking to improve their capacity in genetic testing, will improve accessibility for the common retinoblastoma patient family and provide a rich opportunity for Kenyan healthcare teams to acquire the additional knowledge and skills required to interpret and disclose genetic testing results and counsel families effectively. With improved access to testing, training in the interpretation of tests and counseling, healthcare providers will be able to convey more conclusive and sound information on heritability of each patient, significantly reducing the probability of families relying on an 'optimistic approach' adopted as a means of coping, but rooted in denial.

It is recognized that a variety of healthcare providers can effectively cater to the genetic counseling of families. In Cameroon, a multidisciplinary team of gynecologist-obstetricians, psychologists and medical geneticists together decides on the approach to counsel each case of prenatal diagnosis [20]. In South Africa, the postgraduate genetic counseling program was initiated on the experiences of a medical social worker [21]. Similarly, in Kenya, nurses, general hospital counselors and/or social workers could be recruited to support physicians in genetic counseling for retinoblastoma. This multidisciplinary approach not only alleviates the time demand for ophthalmologists, but also wields the collective communication skills of the team to better support the families' psycho-emotional needs. Genetic training, therefore,

may be diversified and tailored to individual healthcare providers' genetics background to allow effective learning.

The Way Forward

While the strategy as presented here stems from targeted discussions with the genetics task force (fig. 2), the ideas are inextricably linked and influenced by the interdisciplinary discussions at multiple annual meetings of the broader KNRbS group, with the overall aim of advancing care and outcomes for children with retinoblastoma. The group's process is dynamic and iterative, striving to create an evidence base where there is none. The approach has been research based since the beginning, with members actively applying for and acquiring research funds to develop and test various initiatives [22, 23]. Working closely with partners in civil society organizations and the public sector, we are engaging decision makers throughout the research process, so that lessons learned can be formalized within the existing healthcare channels in the long-term.

Conclusions

A 3-pronged approach to developing cancer genetics services in Africa is proposed, focusing efforts on innovative patient and family counseling, community involvement, and enhanced genetics training and research among healthcare workers. The coming together of this genetics task force represents a tremendous achievement. We aim to stimulate innovative approaches in cancer genetics research, education and knowledge translation, taking advantage of unique opportunities offered in the African context.

Acknowledgments

We would like to acknowledge the valuable contributions to the discussions from ophthalmology residents from the University of Nairobi, Kenya. We thank Prof. Dunera Ilako, Chair of the Department of Ophthalmology, University of Nairobi, for her support in this project. L.Q.H. was supported by the Center for International Experience, University of Toronto, through the Association of Universities and Colleges in Canada (AUCC). AUCC had no role in the study design, data collection and analysis, decision to publish, or preparation of the manuscript. We thank Daisy's Eye Cancer Fund Kenya for hosting L.Q.H. during her internship, and for their dedication in facilitating the development of the Kenyan National Retinoblastoma Strategy group.

References

1. Stopfer JE. Genetic counseling and clinical cancer genetics services. Semin Surg Oncol. 2000; 18:347–357. [PubMed: 10805957]
2. Wonkam A, Tekendo CN, Sama DJ, Zambo H, Dahoun S, Béna F, Morris MA. Initiation of a medical genetics service in sub-Saharan Africa: experience of prenatal diagnosis in Cameroon. Eur J Med Genet. 2011; 54:e399–e404. [PubMed: 21473937]
3. World Health Organization. Medical Genetics Services in Developing Countries – The Ethical, Legal and Social Implications of Genetic Testing and Screening. Geneva: 2006.
4. Morhason-Bello IO, Odedina F, Rebbeck TR, Harford J, Dangou JM, Denny L, Adewole IF. Challenges and opportunities in cancer control in Africa: a perspective from the African Organisation for Research and Training in Cancer. Lancet Oncol. 2013; 14:e142–e151. [PubMed: 23561745]
5. Todd C, Haw T, Kromberg J, Christianson A. Genetic counseling for fetal abnormalities in a South African community. J Genet Couns. 2010; 19:247–254. [PubMed: 20135211]

6. Friend SH, Bernards R, Rogelj S, Weinberg RA, Rapaport JM, Albert DM, Dryja TP. A human DNA segment with properties of the gene that predisposes to retinoblastoma and osteosarcoma. Nature. 1986; 323:643–646. [PubMed: 2877398]
7. National Retinoblastoma Strategy Canadian Guidelines for Care. Stratégie thérapeutique du rétinoblastome guide clinique canadien. Can J Ophthalmol. 2009; 44(suppl 2):S1–S88. [PubMed: 20237571]
8. Dimaras H, White A, Gallie B. The Kenyan National Retinoblastoma Strategy: building local capacity in the diagnosis and management of pediatric eye cancer in Kenya. Ophtamol Rounds. 2008; 6(4)
9. Nyamori JM, Kimani K, Njuguna MW, Dimaras H. The incidence and distribution of retinoblastoma in Kenya. Br J Ophthalmol. 2012; 96:141–143. [PubMed: 22028473]
10. Nyawira G, Kahaki K, Kariuki-Wanyoike M. Survival among retinoblastoma patients at the Kenyatta National Hospital, Kena. JOECSA. 2013; 17:15–19.
11. Kimani K, Ouma B, Gallie BL, White A, Dimaras H, et al. Rati's challege: a vision for Africa. Report from the First Kenyan National Retinoblastoma Strategy Meeting, Daisy's Eye Cancer Fund. 2009:39.
12. Tekola F, Bull S, Farsides B, Newport MJ, Adeyemo A, Rotimi CN, Davey G. Impact of social stigma on the process of obtaining informed consent for genetic research on podoconiosis: a qualitative study. BMC Med Ethics. 2009; 10:13. [PubMed: 19698115]
13. Marsh VM, Kamuya DM, Mlamba AM, Williams TN, Molyneux SS. Experiences with community engagement and informed consent in a genetic cohort study of severe childhood diseases in Kenya. BMC Med Ethics. 2010; 11:13. [PubMed: 20633282]
14. Long KA, Thomas SB, Grubs RE, Gettig EA, Krishnamurti L. Attitudes and beliefs of African-Americans toward genetics, genetic testing, and sickle cell disease education and awareness. J Genet Couns. 2011; 20:572–592. [PubMed: 21748660]
15. Nyenze E, Ilako D, Kimani K. KAP of traditional healers on treatment of eye diseases in Kitui district of Kenya. East Afri J Ophtamol. 2007; 13:6–11.
16. Kenyan Ministry of Health. Taking the Kenya Essential Package for Health to the Community: a strategy for the delivery of level one services. Nairobi: Afya House; 2006.
17. Wangalwa G, Cudjoe B, Wamalwa D, Machira Y, Ofware P, Ndirangu M, Ilako F. Effectiveness of Kenya's Community Health Strategy in delivering community-based maternal and newborn health care in Busia County, Kenya: non-randomized pre-test post test study. Pan Afr Med J. 2012; 13(suppl 1):12.
18. Wonkam A, Njamnshi AK, Angwafo FF 3rd. Knowledge and attitudes concerning medical genetics amongst physicians and medical students in Cameroon (sub-Saharan Africa). Genet Med. 2006; 8:331–338. [PubMed: 16778594]
19. Kromberg JG, Wessels TM, Krause A. Roles of genetic counselors in South Africa. J Genet Couns. 2013; 22:753–761. [PubMed: 23723047]
20. Nguefack CT, Brulet C, Njiengwe E, Sandjon G, Onomo M, Kamgaing JT, Kolesnikov A, Epopa A, Wamba B, Doualla C. Douala prenatal diagnosis staff (Cameroon): four years of activity. Prenat Diagn. 2012; 32:94–96. [PubMed: 22470935]
21. Kromberg JG, Sizer EB, Christianson AL. Genetic services and testing in South Africa. J Community Genet. 2013; 4:413–423. [PubMed: 22711384]
22. Dimaras H, Dimba EA, Gallie BL. Challenging the global retinoblastoma survival disparity through a collaborative research effort. Br J Ophthalmol. 2010; 94:1415–1416. [PubMed: 20679076]
23. Dimaras H, Kimani K, Dimba EA, Gronsdahl P, White A, Chan HS, Gallie BL. Retinoblastoma. Lancet. 2012; 379:1436–1446. [PubMed: 22414599]

Color version available online

Fig. 1.

Genetic counseling session. A genetic counseling session at Kenyatta National Hospital. An ophthalmologist talks with the mother of a child with retinoblastoma about the heritable nature of the disease.

Color version available online

Fig. 2.

Retinoblastoma Genetics Working Group. The KNRbS facilitated a group to discuss the challenges and opportunities in delivering a comprehensive genetics service for retinoblastoma families.

Table 1

Common challenges to genetic counseling understanding and compliance as well as strategies to address them

Challenges to understanding/compliance	Representative comments from the Task Force	Contributing factor(s)	Strategies
Patient and family counseling			
Belief that witchcraft or a curse caused disease	'We need to separate retinoblastoma from taboo, tradition and spirituality. Don't deny people's beliefs. Don't tell people they are a fool. Explain to them, then let them realize it themselves.' 'You have to tie their beliefs [i.e. curse, witchcraft] into your explanation [i.e. of genetics].'	Culture, education	Identify metaphors/language in Kenyan communities that could be used to describe common traits
Difficulty in grasping genetic concepts (e.g. 'mutation', 'percent risk', 'inheritance')	'You ask for family history, parents won't know, because they won't include a child with retinoblastoma in the clan record if they think it's witchcraft.'	Culture, education	Conduct focus groups with patient families to identify and address: • origin and reasoning behind beliefs that affect how families understand genetic disease, and • psychosocial needs of families during genetic diagnosis/counseling
Lack of direct translation in local dialects for words such as 'genes', 'chromosomes', etc.	'Explain that eye color and heights are controlled by genes which you get from dad and mom. These genes result in diseases. Retinoblastoma is one of these diseases.'	Culture, education	
Emotional distress, ineffective coping with diagnosis	'What we are doing right now is really not sufficient. We are not helping with the psychosocial needs of the mother.'	Education, psychosocial	Develop tools and incorporate a user-focused approach to counseling
Community involvement			
Inferior role of women in some societies/households may affect how a mother discloses a genetic diagnosis to her family, or how a family internalizes a diagnosis (i.e. blames the mother for 'causing' the disease)	'Traditionally, society thinks the mother is to blame, just like infertility.' 'We need to involve fathers, because they are the decision makers. Especially when husbands support views opposite to those of the doctors.' 'Mothers don't want to be associated with "running in the family" [inheritance], or else husbands can just blame her and remarry. We need to decide carefully how much information is released to the parents [in terms of who carries the mutation].'	Culture, society	Determine how local opinion leaders and traditional healers could complement the counseling approach Design and test a counseling/educational approach that includes fathers, extended family, local opinion leaders, etc. Develop and test a peer-to-peer genetic education program, with outreach components to serve those who live away from a clinic
Extended family (e.g. grandparents, uncles, aunts, other elders) beliefs/understanding of a disease determines the compliance with treatment/screening; parents of an affected child obey the instruction of elders, whether or not they understand/wish to comply with a medical opinion	'People around the mother influence her decisions a lot … so it is important to have opinion leaders and healthcare workers in the community to support the mother.'	Culture, society	

Challenges to understanding/compliance	Representative comments from the Task Force	Contributing factor(s)	Strategies
Families live too far from a clinic to attend frequent counseling sessions	'If the parents come to Nairobi once, and we don't find any disease this time. Then the parents will think the child will always be all right. Then they will never come back. So asking the parents to always come to Nairobi is not possible.'	Socioeconomic	
Medical education			
Timing of counseling is at or near diagnosis, and too stressful to facilitate uptake/understanding by parents	'We need someone who can sit down and spend the time to go through all the concerns of the mother.'	Healthcare system	
Lack of genetic counseling training/positions	'In every discipline in Africa, doctors are overwhelmed. It's not an excuse. We can use counselors and other healthcare providers. You have a team that you work with, that's what happened with diabetes.' 'We need to educate the health workers, e.g. nurses, clinical officers, who interact with mothers everyday. They see a lot of people in one day.' 'There isn't a formal program or institution which can provide the counseling. We [treating physicians] sometimes base our counseling on parents' attitudes. When the parents resist, we'll give them some time, then try again. And we also tell the nurses, and let the nurse convey the information.'	Healthcare system	Develop a multidisciplinary genetic counseling support team Develop tools to assist in knowledge and practice of medical genetics, testing, counseling Pursue a global-to-local approach to make genetic services accessible to Kenyan families; connect to capacity building efforts in African genomics technologies
Lack of medical genetics training/positions	'Even doctors don't understand it [retinoblastoma genetics], so they don't embrace it. They feel that it is a burden and resist it.' 'Counseling we do. But if we can map the genetics of the parents, that will be better. If we can really see if there's a predisposition.'	Healthcare system	
Limited genetic testing facilities	'You can't create all the genetic information without giving them the services. You can't tell them you have to go to India, Europe, etc.; they can't even go to Nairobi.'	Healthcare system	