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

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Key Words
Africa · Cancer · Clinical genetics · Genetic counseling · Genetic testing · Retinoblastoma

Abstract
Background/Aims: 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. 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. 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.

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-resource 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 back-
grounds, 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
<table>
<thead>
<tr>
<th>Challenges to understanding/compliance</th>
<th>Representative comments from the Task Force</th>
<th>Contributing factor(s)</th>
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</thead>
<tbody>
<tr>
<td><strong>Patient and family counseling</strong></td>
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<td>Belief that witchcraft or a curse caused disease</td>
<td>'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.'</td>
<td>Culture, education</td>
<td>Identify metaphors/language in Kenyan communities that could be used to describe common traits</td>
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<td>'You have to tie their beliefs [i.e. curse, witchcraft] into your explanation [i.e. of genetics].'</td>
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<td></td>
<td>'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.'</td>
<td>Culture, education</td>
<td>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</td>
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<td></td>
<td>'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.'</td>
<td>Culture, education</td>
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<td>Emotional distress, ineffective coping with diagnosis</td>
<td>'What we are doing right now is really not sufficient. We are not helping with the psychosocial needs of the mother.'</td>
<td>Education, psychosocial</td>
<td>Develop tools and incorporate a user-focused approach to counseling</td>
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<td><strong>Community involvement</strong></td>
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<td>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)</td>
<td>'Traditionally, society thinks the mother is to blame, just like infertility.'</td>
<td>Culture, society</td>
<td>Determine how local opinion leaders and traditional healers could complement the counseling approach</td>
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<td>'We need to involve fathers, because they are the decision makers. Especially when husbands support views opposite to those of the doctors.'</td>
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<td></td>
<td>'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].'</td>
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<td>'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.'</td>
<td>Culture, society</td>
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<td>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</td>
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<td>Families live too far from a clinic to attend frequent counseling sessions</td>
<td>'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.'</td>
<td>Socio-economic</td>
<td></td>
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Table 1. Common challenges to genetic counseling understanding and compliance as well as strategies to address them
Table 1 (continued)

<table>
<thead>
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</tr>
</thead>
<tbody>
<tr>
<td>Medical education</td>
<td>'We need someone who can sit down and spend the time to go through all the concerns of the mother.'</td>
<td>Healthcare system</td>
<td>Develop a multidisciplinary genetic counseling support team</td>
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<td>Lack of medical genetics training/positions</td>
<td>'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.'</td>
<td>Healthcare system</td>
<td>Develop tools to assist in knowledge and practice of medical genetics, testing, counseling</td>
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<td>Lack of genetic counseling training/positions</td>
<td>'We need to educate the health workers, e.g. nurses, medical officers, who interact with mothers everyday. They see a lot of people in one day.'</td>
<td></td>
<td>Pursue a global-to-local approach to make genetic services accessible to Kenyan families; connect to capacity building efforts in African genomics technologies</td>
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<td>Limited genetic testing facilities</td>
<td>'You can't tell them you have to go to India, Europe, etc.; they can't even go to Nairobi.'</td>
<td>Healthcare system</td>
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</table>

There is an urgent need to strengthen genetic education in Africa. There are no specialized training programs or recognized independent careers for medical genetics or genetic counselors. However, as the need for genetic services and professionals is growing, there are no specialized training programs or recognized independent careers for medical genetics or genetic counselors. The development of specialized curricula for medical genetics and testing is paramount. Postgraduate curricula with practical exposure in genetic counseling and testing will improve accessibility for the common retinoblastoma patient family and provide a rich opportunity for local laboratories to improve their capacity in testing.

Medical Education

There is an urgent need to strengthen genetic education 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 genetics. The development of specialized curricula for medical genetics and testing will improve accessibility for the common retinoblastoma patient family. (1) Ensure that parents are aware of the correct referral pathway to access the care they require; (2) establish a multidisciplinary team; and (3) connect the family with social assistance programs where available, that remove barriers to such care.
nity 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.

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