



Genetic Disorder: Albinism Counselling through a Mobile Phone Platform in Developing Countries

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ABSTRACT A congenital disorder (albinism) is characterized by a complete or partial absence of pigment in the skin, hair and eyes without effective counselling and education to the patients in Africa is of great concern. This paper investigated how people living with albinism are counseled and educated in South Africa. The study employed a case study approach in data collection. It was revealed that the number of trained genetic nurse counsellors in South Africa are few and this makes it extremely difficult for the counsellors to offer counselling support to albinos. Majority of the albinos travel long distances to city clinics for help. It was further discovered that almost ninety percent of both the counsellors and albinos own mobile or smart phones which can be used as a communication tool to counsel the albinos. However, this is not the case. The findings lead to the proposal of Genetic disorder counselling mobile phone platform – (GDCMPP) to enhance interaction between albinos and genetic nurse to improve education, and counselling services to the albinos.

INTRODUCTION

The general objective of genetic health service is to “help people with a genetic disorders to live and reproduce as normally and responsibly as possible” (Irakiza and Mutesa 2014). A genetic disorder is any disease that is caused by an abnormality in an individual’s genome or the person’s entire genetic makeup (Krause and Greenberg 2008). The abnormality can range from minuscule to major one, from a discrete mutation in a single base in the DNA of a single gene to a gross chromosome abnormality involving the addition or subtraction of an entire chromosome or set of chromosomes. Some genetic disorders are inherited from the parents, while other genetic diseases are caused by acquired changes or mutations in a pre-existing gene or group of genes. Mutations can occur either randomly or due to some environmental exposure. There are many forms of genetic disorders including, Down Syndrome, neural tube defects and albinism (Christianson et al. 1995).

Albinism in human being is a congenital disorder characterized by a complete or partial absence of pigment in the skin, hair and eyes (Boissy et al. 1996). Albinism is associated with a number of vision defects, such as photophobia, nystagmus, and amblyopia. The lack of skin pigmentation makes for more susceptibility to sunburn and skin cancers. In rare cases, albinism may be associated with deficiencies in the trans-

portation of melanin granules. This also affects essential granules present in immune cells leading to increased susceptibility to infection (Kaplan et al. 2008).

People with albinism often face social and cultural challenges (even threats), as the condition is often a source of ridicule, discrimination, or even fear and violence. It is socially stigmatized in many African societies (Magna 2014), including South African society. The discrimination and ridicule affect the self-esteem and self-confidence of the albinos. Self-esteem according to Smith and Mackie (2007), reflects an individual’s overall subjective emotional evaluation of his or her own worth. It is the decision made by an individual as an attitude towards the self. It can result in positive or negative evaluations of one’s self. In the case of Albinos, it always result into negative evaluation of themselves because of society discrimination and ridicule.

In December 1997, a white paper on an Integrated National Disability Strategy was launched from the Deputy President’s Office. It provides guidelines to government departments on how to include disability related programmers in their functions including albinism disorders (DoH 1997). For the provinces to achieve this, the South African Government introduced genetic counselling to assist individuals and families with genetic disorder. Genetic counselling is a communication process which aims to assist individuals or families with a genetic disorder or birth

defect to understand the medical implications, diagnosis, prognosis and management of the disorder; the inheritance pattern and risk of recurrence; the options available to deal with the risk and the choices and actions required; and how to make the best possible daily life adjustment to the disorder.

The question is how effective is this genetic counselling being offered to people with albinism as there are few trained genetic counsellors in the whole country, South Africa. What compounds the problem is that many of these albinos live in rural communities in South Africa and the few trained genetic counsellors live in the urban cities which makes it extremely difficult to offer counselling service to the rural communities.

However many of these albinos own mobile phone which can be used by the counsellors as a means of effective communication to educate the albinos but they do not use this for such purposes. Mobile phones have a wide range of facilities that may be useful in a health care context such as web access, video, photo messaging and SMS text messaging but are not utilized for such purposes (Ramanathan et al. 2013).

The objective of this paper is to investigate how Albino people are counseled and educated by genetic nurse counsellors and based on the findings propose a genetic disorder counselling mobile phone platform – (GDCMPP) to enhance and improve the counselling process.

The proceeding sections of this paper is presented as follows: related work, methods, results and discussion, proposed Genetic disorder counselling mobile phone platform – (GDCMPP) and finally the conclusion.

Related Work

Albinism as a Genetic Disorder

In South Africa the more serious common monogenic or single-gene disorders that occur in all groups include Huntington disease (Hayden 1981; Krause and Greenberg 2008), Marfan syndrome, myotonic dystrophy, neuro-fibromatosis (Beighton et al. 1991), polycystic kidneys, tuberous sclerosis, and osteogenesis imperfecta, among others (Beighton 1976). In addition, those that occur at higher rates than usual in specific groups include: oculocutaneous albinism in the Black population (Kromberg and Jen-

kins 1982); cystic fibrosis in the White groups (Denter et al. 1992); Tay Sachs and Gaucher disease in the Jewish population (Jenkins et al. 1977; Goldblatt and Beighton 1979); familial hypercholesterolemia and porphyria in the Afrikaans (of Dutch origin) population (Jenkins 1990); beta thalassemia in the Greek, and various thalassemia in the Indian populations (Krause 1994).

Oculocutaneous albinism (OCA) is generally the result of a biological inheritance of genetically recessive genes passed from both parents to an individual. A mutation in the human gene may result in the deregulation of melanocyte tyrosinase enzymes, a change that is hypothesized to promote brown versus black melanin synthesis, resulting in a third oculocutaneous albinism *genotype* (Boissy 2016). Some rare forms are inherited from only one parent. There are other genetic mutations which are proven to be associated with albinism. All alterations, however, lead to changes in melanin production in the body (Boissy 2016). Some of these are associated with increased risk of skin cancer.

The chance of offspring with albinism resulting from the pairing of an organism with albinism and one without albinism is low. However, because organisms (including humans) can be carriers of genes for albinism without exhibiting any traits, albinistic offspring can be produced by two non-albinistic parents. Albinism usually occurs with equal frequency in both sexes (Boissy 2016). An exception to this is ocular albinism, which it is passed on to offspring through X-linked inheritance. Thus, ocular albinism occurs more frequently in males as they have a single X and Y chromosome, unlike females, whose genetics are not like so (Hutton, and Spritz 2008). Humans beings with albinism often face social and cultural challenges (even threats), as the condition is often a source of ridicule, discrimination, or even fear and violence. Albinos are socially stigmatized in many African societies. A study conducted in Nigeria on albino children stated that “they experienced alienation, avoided social interactions and were less emotionally stable. Furthermore, affected individuals were less likely to complete schooling, find employment, and find partners” (Magna 2014). Many cultures around the world have developed beliefs regarding people with albinism.

In African countries such as Tanzania (BBC News 2008) and Burundi (BBC News 2009) there has been an unprecedented rise in witchcraft-

related killings of people with albinism in recent years, because their body parts are used in potions sold by witchdoctors.

Self-esteem

Many early theories suggested that self-esteem is a basic human need or motivation. American psychologist Abraham Maslow included self-esteem in his hierarchy of human needs. He described two different forms of “esteem”: the need for respect from others in the form of recognition, success, and admiration, and the need for self-respect in the form of self-love, self-confidence, skill, or aptitude (Maslow 1987). Respect from others was believed to be more fragile and easily lost than inner self-esteem. According to Maslow, without the fulfillment of the self-esteem need, individuals will be driven to seek it and unable to grow and obtain self-actualization. Maslow also states that the healthiest expression of self-esteem “is the one which manifests in respect we deserve from others, more than, fame and flattery”. Modern theories of self-esteem explore the reasons humans are motivated to maintain a high regard for themselves. Sociometer theory maintains that self-esteem evolved to check one’s level of status and acceptance in one’s social group. According to Terror Management Theory, self-esteem serves as protective function and reduces anxiety about life and death (Greenberg 2008).

Self-esteem is important because it shows ourselves how we view the way we are and the sense of our personal value. Thus, it affects the way we are and act in the world and the way we are related to everybody else (Bonet 1997).

Carl Rogers (1902-1987), an advocate of humanistic psychology, theorized the origin of many people’s problems to be that they despise themselves and consider themselves worthless and incapable of being loved (Wickman and Campbell 2003). This is why Rogers believed in the importance of giving unconditional acceptance to a client and when this was done it could improve the client’s self-esteem (Bonet 1997). In his therapy sessions with clients, he offered positive regard no matter what (Wickman and Campbell 2003). Indeed, the concept of self-esteem is approached since then in humanistic psychology as an inalienable right for every person, summarized in the following sentence:

“Every human being, with no exception, for the mere fact to be it, is worthy of unconditional respect of everybody else; he deserves to esteem himself and to be esteemed (Bonet 1997).

Mobile Phone

Mobile phones have the potential to improve access to health care services (WHO 2008) particularly in developing countries where mobile phones are more prevalent than ICTs such as landlines, telephones and the internet (Kinkade and Verclas 2008). The exploration of the potential of mobile phones in health service delivery has led to the establishment of mobile phone-based projects for health care and the development of mobile phone-based applications for health care services in developing countries such as South Africa, India, Rwanda, Peru, Uganda (Vital Wave Consulting 2009). These applications fall in the following utilization areas: Education and Awareness, Remote Data Collection, Remote Monitoring, Communication and Training for healthcare workers, Disease and Epidemic Outbreak Tracking, and Diagnostic and treatment support (Vital Wave Consulting 2009). White (2005) and Fynn et al. (2006) note that these applications take advantage of the mobility and flexibility that mobile phones provide, in combination with other ICT technologies such as the internet and database technologies, the potential to overcome many of the barriers of distance and cost facing developing countries.

The use of mobile phone In health (mHealth) provides the potential to deliver health behavioural interventions tailored to a person’s baseline characteristics, such as disease, demographic, and frequently changing environmental contexts (Patrick et al. 2008).

The communication between a healthcare provider and a patients through a mobile phone can take place in two platforms, Message-based interaction (MBI) or Remote Procedural Call (RPC) platform. A message-based interaction includes one mobile phone element creating a message that details the service required for another mobile phone element (Sommerville 2010). In MBI approach, unavailability of sender or the receiver can be tolerated as the message simply stays in a queue until the receiver becomes available. Secondly it does not require the sender and receiver of the message to be aware of each other.

On the other hand, Remote Procedural Call (RPC) platform requires the communicators on both mobile phone components to be present. One component calls another component as if it was a local procedure. Using RPC communication platform requires both the sender and the receiver to be present at the time of communication. Example of these platforms include video calls and online discussion platform.

The researcher has chosen MBI platform because of poor bandwidth internet connectivity in Southern Africa.

METHODOLOGY

In order to realize the objectives of this research paper two provinces, Gauteng and Kwa-Zulu Natal provinces were selected. These two provinces were selected because of availability of trained genetic nurse counsellors and high prevalence of albinism in nearby villages. Two health care clinics were selected from each of the two province making a total of 4 health care clinics. For confidentiality which was requested by these clinics, the names of the clinics are anonymized in this research paper. Given the nature of the study, a case study approach provided the most effective method for data collection. Participants for the study were drawn from the entire population genetic nurse counsellor and albinos. These genetic nurse counsellors provide counselling to the albinos and the albino are the recipients of the counselling services. Two genetic nurse counsellors and two albinos from each clinic were selected for participation. A total of four genetic nurse counsellors and four albinos volunteered to take part in the study. Data was collected using semi-structured, open-ended interviews. This data collection method was used because it provides both flexibility and wider focus in data collection. The interviewees were asked to respond to the following questions in their own words:

1. What is the current practice of providing counselling and education to albinos?
2. How often is counselling and education provided to the albinos per month?
3. What technologies do you use to assist in providing or receiving the counselling service
4. Do the albinos and counsellors have access to mobile phones?

5. Is the mobile phone used to support the counselling process? If not what is the mobile phones usually used for?

The interviews lasted for one hour with each interviewee and were audio-recorded and transcribed by the researcher. Transcripts were coded using content analysis techniques as prescribed by Fraenkel et al. (1993) and others. After the initial coding, an independent researcher and the main researcher met to check the consistency of their interpretation. The researcher then coded the final transcripts, identified the main themes, and traced possible relationships. The different broad categories that were noted are discussed below and they include; current practice of providing counselling and education to albino people; and current technologies used and access to mobile phones by albinos and genetic counsellor

OBSERVATIONS AND DISCUSSION

Current Practice of Providing Counselling and Education to Albino People

The genetic counselors indicated that they are few counselors who serve many genetic disorder patients. This is confirmed by Kromberg et al. (2006), that genetic counselors are few but provided service to patients from all the population groups with an average of about eighty (percent) of the patients seen from the state health-care sector of other provinces.

Genetic nurse counselors proved services on counseling, teaching, research, marketing, and administration. The counselor further stated that they contribute a great deal to the provision of genetic services in South Africa. They counsel approximately one-third of all the cases sent for genetic counseling, and the numbers are increasing. Apart from these, they indicated that they play many other roles that are essential to the smooth and efficient running of genetic service. They indicated that counselors in genetic counseling sessions elicit a relevant family and medical history, construct an accurate pedigree, and then calculate risk status of the individual and family and offer choices for dealing with this risk. In the process they obtain an appropriate social and psychosocial history of the individual and family. They interpret special investigations associated with the genetic condition, and apply theoretical scientific knowl-

edge, sensitively. As part of this process, they also adapt to change and tolerate uncertainty. One counselor said *"we plan and execute a range of interviewing and counseling techniques, and also manage the clients, patients, and their families with empathy, caring, compassion, sensitivity, acceptance of diversity, and respect for their dignity, privacy, and confidentiality"* One of the albinos also stated that *"we are rejected from the society and this affect our emotional status, we need more counselling to establish our self-esteem and confidence."* One of the counselors also confirmed that *"because we are few, we are unable to serve all the people with genetic albinism."*

It was further revealed that South Africans who are affected by a genetic albinism are confronted with unique circumstances and barriers, including poverty and minimal access to resources. Affected individuals and families are unable to afford information and interventions to address basic health issues, let alone to maximize their quality of life. These members from rural communities' travel hundreds of kilometers to city clinic to obtain comprehensive genetic services. Genetic counseling as indicated by a nurse, takes place through a translator with no training in genetics or counseling, leading to possible misunderstanding between the patient and the counselor.

The services of genetic counselling are done through face to face approach.

Current Technologies Used and Access to Mobile Phones by Albinos and Genetic Counsellor

The patients who go for counselling indicated that they are provided with pamphlets which explains the levels and status of their conditions. The counsellors who give education and counselling to the patients use flipcharts, smart boards and verbal expiations to educate the patients.

The albinos and the counselors' were asked if they have access to mobile phones? This was followed by another question, is the mobile phone used to support the counselling process? If not, what is the mobile phones usually used for? The purpose of these questions was to ascertain how mobile phones can be used in the counselling and to support the people living with albinism through counselling and education. All the interviewee (100%) indicated that they own

mobile phone or smart phones. This was not a strange response because Global Economic Prospects (2007) confirms that over eighty percent of all youth and adults own one mobile phone or more. Cassidy et al. (2009) reiterates that mobile phone can be a platform to promote health education and counseling if it is properly harnessed.

On the question of how mobile phones are used to support counseling process, the counselors and the albinos unanimously said they do not use mobile phone instead the albinos travel to the clinic for support. The counselors, on the other hand, are unable to travel to the villages to give support to the patients because they are few in number and serve a huge number of albinos in the country. They further indicated that mobile phones are not used for counselling purposes because no special application has been created to send interactive message or SMS to patients. Patients and counselors alike use their mobile phones to communicate with their friends and relative on issues outside HIV/AIDS counselling. However, mobile phones have a wide range of facilities that may be useful in a health care context such as web access, Video, photo messaging and SMS text messaging (Ramanathan et al. 2013).

The Need for Genetic Disorder Counselling Mobile Phone Platform – (GDCMPP)

Based on these findings, the researcher proposes a Genetic disorder counselling mobile phone platform – (GDCMPP) to enhance interaction between albino patients and counsellors to improve education, and counselling practices to the albinos. This findings has been motivated by the fact that there are few trained genetic counsellors in South Africa and these counsellors are situated in few regions like Gauteng province and KwaZulu Natal. The platform will be linked to a standalone Telecom Mobile System. The patient uses the mobile phone to send a code for service. The code is routed to the Genetic Communication software system. The patient is then presented with the menu to select a service. The patient then selects educational or counsellor services for the specific clinic. The system then notifies the counsellor about the request which prompts the counsellor to uses his or her code to access the system to provide the service required by the patient. The counsellor then sends either a text message or

voicemail to the albino patient. Figure 1 shows the Proposed Genetic disorder counselling mobile phone platform – (GDCMPP).

As indicated in Figure 1, the proposed system incorporates genetic disorder communication software system which has a service menu and communication interface. The service menu provides options for the albinos to enter codes which are transmitted to the counsellor. The system again has a database which provides both the counsellor and the patients a record base of their mobile numbers and codes. These codes are used by both the albinos and the counsellor to access the system. In other to keep the system on an internet web page, the system is linked to a mobile network provider web “Telecom system”.

The principles underlying the development of the GDCMPP system will be the ability of the system to;

- Provide security exchange data (encryption services).
- Use structured, widely-adopted, updated ways of capturing and storing data.
- Use standardized, widely-adopted health information exchange protocols (standard messaging and transport capability) and adapt to advances in technology.

CONCLUSION

This paper examined how counselling and education are provided to people who are albinos in South Africa. The investigation unearthed the current practices of genetic counselling to albinos and further explore the technologies and tools applied by genetic counsellors during the process of counselling and education of patients.

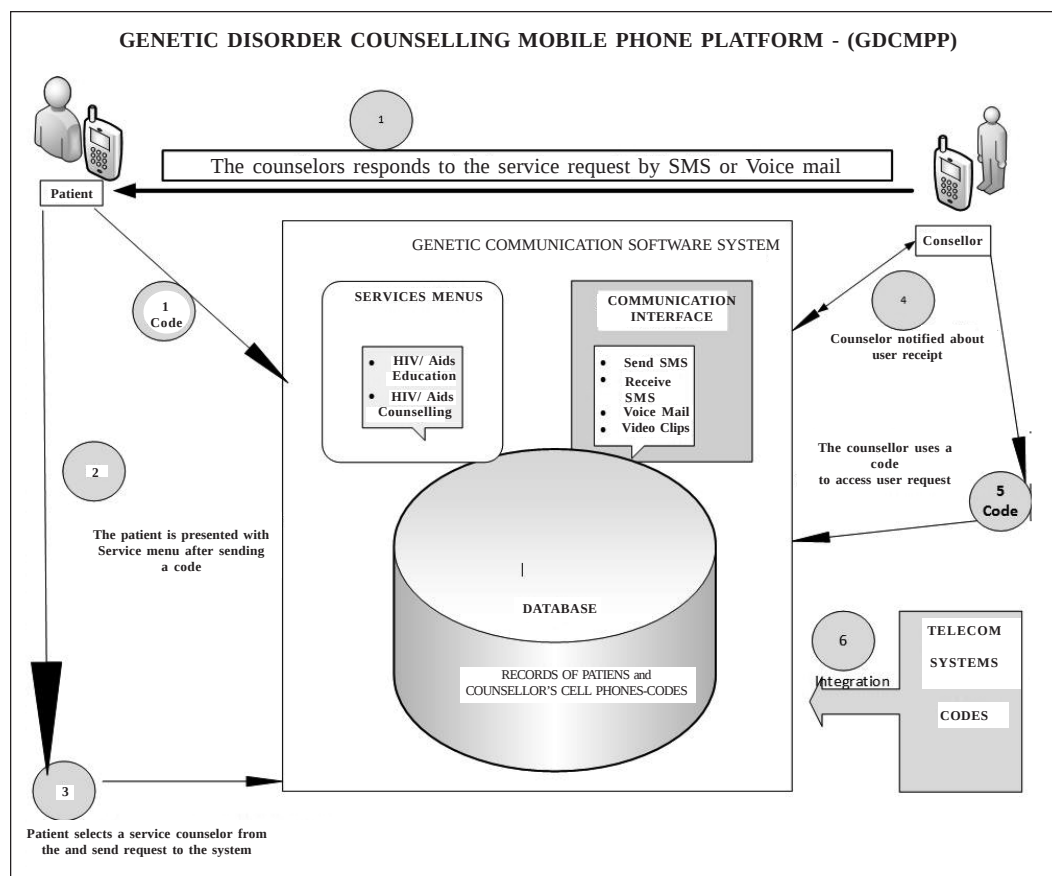


Fig. 1. Proposed Genetic disorder counselling mobile phone platform – (GDCMPP)

Source: Author

It was revealed that the number of trained genetic nurse counsellors in South Africa are few and this makes it extremely difficult for the counsellors to travel to villages around the city clinics to offer counselling support to albinos. Some of the patients travelled about 50-150 km to such city clinics for help. It was further discovered that almost ninety percent of both the counsellors and albinos own mobile phones of which eighty percent are smart phones.

The findings lead to the proposal of Genetic disorder counselling mobile phone platform – (GDCMPP) to enhance interaction between albino patients and genetic nurse counsellors to improve education, and counselling to the albinos. The platform also serves as structure to assist government and non-governmental organizations who intend to contribute to support albinos who are discriminated against and attacked in Africa.

RECOMMENDATIONS

It is recommended that the department of health in many developing countries install a separate database which is able to store records of albinos, notify the counsellor about request of albinos and prompts counsellor to uses code to access the system to provide the service required by the albinos. It is also recommended that the database is annually updated.

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