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GENETIC SCREENING, SICKLE CELL ANEMIA AND THE AFRICAN AND AFRICAN-AMERICAN PERSPECTIVES: THE ETHICS OF SCREENING AND COUNSELING

by

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Introduction

It is not all the time that a personal real life experience coincides with an issue that one is trying to explore in an academic paper. For me, this is one such occasion. Between the time that I submitted the draft of my paper, and the deadline for the final version, I got news of the passing of my cousin-in-law, a young woman of 25. She died during the birth of her second child, a year after she lost her second pregnancy. It was only after her death that I got to know that she had been a carrier. I am pretty sure that the husband did not know before they got married. Even if he did know, I am almost sure it would not have made a difference. Further still, I cannot be sure that she herself got appropriate diagnosis. Of course, there are clinics and teaching hospitals. Part of the problem has to do with access. General hospitals, which the majority of the population patronize, do not have the facilities. Patients' complaint of headache, fever, or stomach pain are treated almost immediately with pain killers, in most cases without any tests to determine the cause. In the event, serious ailments go undiagnosed and untreated till it is too late. That was the case with my cousin-in-law. In the following, I am not so much concerned with the cause and natural history of sickle cell anemia. Even if I want to, I cannot do justice to it, and I would rather leave that to the experts. My focus, therefore, is mainly on the ethics of screening and counseling, in the light of African and African-American world views and contemporary realities.

Sickle Cell Anemia

Experts tell us that there has been much confusion in the public perception of the reality of

sickle cell disease, and that the confusion of sickle cell trait (SCT) with sickle cell anemia (SCA) has led to much misinformation concerning the spread of the disease. Persons with SCT -Hb As- have a genetic condition in which an hemoglobin S (Hb S) is combined with hemoglobin Hb A. On the other hand, SCA is a condition in which a person has two hemoglobin S (Hb S). It has been determined that there is no significant difference in life expectancy between persons with AA (normal) and AS (SCT). Therefore, the rush to screen every one including those with SCT has been criticised as unwarranted, especially when it is not accompanied with appropriate counseling (Bowman, 1978).

Sickle Cell Anemia, according to one of the experts, " is characterized by the production of an abnormal molecule which has disastrous effects on the cells which elaborate it." (Neel, 1979, p.2) From this, Neel concludes that there is no satisfactory provision for therapeutic intervention: "Sickle cell anemia, unlike a variety of other genetic diseases, has simply not been a disease whose therapeutic time has come." (p. 2) Of course, it is not thereby concluded that screening and counseling are useless or inapproriate.

Incidence of SCA among African descendants

It has been observed that there is a high incidence of SCA among Africans and people of African descent. In a study of the incidence among the American Black population, it was discovered that the disease incidence was 1 in 400-800 while carrier frequency was 1 in 10-12. (President's Commission, 1983). Similar figures can be established for the continent. The question is why is there such a high frequency of the condition among African descendants? One answer- with particular reference to Africa, is the impact of falciparum malaria and the great resistance that SCA carriers have to it. This means that death from malaria is rare for SCA's and therefore if a large segment of the

population are carriers, they will survive the attack of malaria till adulthood and be able to pass on their genes, while those few who are not protected by the SCA condition will fall to malaria (President's Commission, 1983, p.20). One might consider it paradoxical that the SCA condition could have a protective role! Or more odd still, that, given the deleterious effect of malaria on African populations in general, this gene could serve any useful purpose! But when one observes that malaria is preventable and curable, but that there is no therapeutic alternative for SCA, the aburdity of those thoughts becomes apparent. A second factor identified is the prevalence of in-breeding among groups of African population which contributes to the reproduction of the genes from generation to generation. What then, is the attraction of screening, even when it appears there is no therapeutic alternative?

Genetic screening involves the deliberate testing of a population for the purpose of identifying those who might possess a particular genetic make-up. Screening is usually the first step toward a diagnosis. Anyone identified as having the condition for which the screening was done may then be further tested. In some cases, screening may also refer to the specific tests in individuals who have been determined to be at risk of a condition. For instance, a person determined to have been exposed to tuberculin infection may be tested for the condition. Screening may require just a blood test or it may involve tests on laboratory cells or, in the case of prenatal testing, the use of sonograph or fetoscopy.

Rationale for Screening Programs

There are several rationales for screening: to determine the connection between a specific genetic make-up and a medical condition; to identify the impact of certain environmental factors on genes; and to determine the incidence of a particular genetic condition among a population group.

Screening may also be done specifically for research purposes-- to test a new screening technique. There are both genetic and non-genetic screening programs. The latter is also directed to uncovering the need for medical intervention and includes, for example, blood pressure tests or tuberculin tests which are not based on assumptions about genetic constitution.

The main goal of screening is to determine the need for medical intervention, or to alert the individual as to the risks of genetic disease. In new born screening, the objective is to discover a genetic condition for which it has been suspected that the baby is susceptible. In prenatal carrier screening, the objective is to alert the parents of the risks of the fetus carrying some abnormal genes. In genetic screening, to a larger extent, than in routine medical screening, the information from the results of the procedure is also important for other people beside the individual screened. Thus there is the moral problem of disclosure. Screening for reproductive reasons is used to help couples as well as individuals make decisions about their reproductive choices.

Screening and Counseling

For most cases, screening and counseling go, or ought to go, together because of the obviously challenging situation faced by a family discovering that their prospective child is susceptible to a genetic disease, or a couple discovering that one of them has a genetic condition that may affect their offsprings. Genetic screening falls into three major kinds: newborn screening for detecting genetic conditions in newborns with a view to avert serious health problems through medical intervention; carrier screening for identifying individuals whose genetic constitution may include some abnormal genes that may affect their offsprings; and prenatal screening, for examining the fetus through the amniotic fluid for the presence of genetic disease.

New-born screening is used to determine the existence of certain "inborn errors of metabolism", as in sickle cell anemia (SCA). These are conditions in which there exists an abnormal or missing enzyme or other protein, resulting from a defective gene. In such conditions, there is a 25% chance that any subsequent pregnancy will result in a child with that condition. In carrier screening, on the other hand, people of reproductive age are provided with relevant information to the health of any children they might have. This is especially the case with screening for SCA which occurs at a higher frequency among people of African descent. Carrier screening of members of this group, it is thought, can help them to detect the existence of the disorder prior to their producing a child, and relatives may benefit by using the result to determine their own susceptibility.

Screening and Counseling for the Sickle Cell condition

The historical purpose of screening in public health has normally been to detect those who have contagious disease to prevent its spread and allow for timely medical intervention. The justification for screening has therefore relied on the availability of treatment procedures. Screening for genetic conditions, including SCA does not appear to be based on this premise. Here, screening is not to combat an identified virus or bacteria, but to prevent the birth of humans with the condition. This is one of the moral dilemmas of screening for SCA. On the one hand, a carrier, who gets to know his/her condition, may be in a position to make informed decisions about their future choices. On the other hand, it appears that such an individual may become a victim of this (public) knowledge of his/her condition. It has therefore been argued that, since SCA is not a disease, the fight against it is fight against people, both real and potential (Murray, 1978). It is more difficult if one considers the plight of the people with this condition, who have to live with the realization that they are not

appreciated as full human beings. Screening means the identification of individuals, and it often creates anxiety because of the stigmatization that it leads to, especially when, in most cases, counseling comes after screening and identification. In addition, the black community had problems with mandatory screening programs because of the past history of discrimination and unjustified medical and research intervention in their lives. For instance, it was feared that the information from the screening procedures may be used to deny them employment and insurance protection. This fear proved justified in some cases. For instance, people with SCA were denied employment in the armed forces, and with airlines (as flight attendants). In some cases, it raises the moral problem of public disclosure of non-partenity when both spouses have no trait and are not carriers and their child has the condition.

From all these, it follows that appropriate genetic counseling must go together with the screening procedure. As Murray explains, genetic counseling "is the process of communicating all of the factors that relate to the disease or condition in question, including the manifestations of the disease, the prognosis of the disease, the genetics of the disease and the alternatives of one or another course of action for those at risk." (cited by Scott-Emuakpor,1979 p.16) The aim of counseling in SCA has been to provide information to the public about the condition and regarding the available options in the hope of reducing the incidence of the condition. The success of this approach can be determined by reduction in the number of SCA cases. It is not clear whether the results are positive or negative thus far. However, in view of the socio-cultural beliefs of the people of Africa and their descendants in the United States, it may not be difficult to discover the answer.

Generally, many Africans choose to ignore the scientific prediction regarding the probability of a child with SCA being born into the family. One reason is the cultural and religious belief in

predestination and the power of God and extra human forces to intervene on one's behalf. Even if the services were available and access was easy, I cannot be sure that my cousin-in-law would have taken full advantage of it. For it is difficult to get a typical African person to go to the clinic for the determination of his/her genetic condition. Secondly, the causes of illness as conceived by Africans differ from European conception, and therefore the scientific prediction may not be trusted by them. People will not entertain any counsel against natural reproduction because of the belief that one should bear one's own children. I will discuss each of these factors in turn, drawing from Gbadegesin (1994).

African (Yoruba) Perspectives

For Western scholars who attempt to understand traditional African belief systems, the first problem encountered is the number of non-physical (or quasi-physical) entities that populate the African universe: God, gods, ancestral spirits, nature spirits, and how these are constantly invoked in the causal explanation of events. There is no denying the fact that these entities are believed to have particular roles to play in the lives of Africans. This is why they are taken seriously by a typical African, including the scientifically minded ones. Thus, as Mbiti remarks sometimes ago, the general impression of the African is that he/she lives in a religio-spiritual universe. The existence of these entities in the African universe is largely responsible for their conceptions of cause and effect. Generally, there is a belief in a supreme being, **Olodumare, Onyame, Chukwu** (for the Yoruba, Akan and Igbo respectively). This is the architect (not creator) of the world. The world is made out of some pre-existing stuff. In the making of the world, there is a division of labor: one lesser deity is responsible for the molding of the body, another responsible for supplying the part essential for

determining the course of events for a person (**ori** for the Yoruba), while the life principle is supplied by the supreme deity. Given this world-view, it is easy to understand why causal explanation should take the form of appeal to a world beyond human control. The human person is subject to a variety of forces, seen and unseen, in and outside nature, which account for the events that happen to him/her. This basic structure of Yoruba reality, which shares many similarities with other African perspectives, can be summed up in a few propositions which can then be used to define their conceptions of health and disease.

First, there is the belief in the existence of Olodumare, the supreme deity, who controls the events in the universe. Everything that happens is ultimately traced to the will of God (***amuwa Olorun***).

Second, there are other deities, a few of whom are regarded as peers of Olodumare. These include the arch divinity, who is the molder of human body; Orunmila, the wisdom deity; and Esu, the deity of order. Others include ogun, orisa oko, shango etc. Each of these has a role to play in the maintenance of order and the promotion of human good.

Third, there are extra-human forces, mainly evil (***ajogun***) whose concern is to disrupt the order of the universe and create problems for people. These include the forces of witchcraft, sorcery, diseases, misfortune, curse etc. There is a constant struggle between these forces and the deities whose concern is the promotion of the good.

Fourth, each person has, in addition to the body and life principle which is spiritual, a personality soul (**ori inu**) which is the bearer of destiny. The meaning of this is that we are all here on a mission to deliver the message of destiny which we carry from the pre-natal world. Our live experiences reflect the content of this message, and when we stray away from the message, we do

not do well. Some messages are bad and one may need to appease the appropriate deities to improve on one's destiny. In the final analysis, however, it may not be possible to change an unwanted destiny. In some cases, a good destiny may be obstructed by the forces of evil- super-natural.

Fifth, human beings have to find a way of avoiding the forces of evil. Fortunately, they can depend on the support of ancestral spirits to help them in this struggle because the ancestors have acquired greater power in the land of the dead. To assure themselves of this support and protection, descendants are expected to live well, obey moral injunctions and honor the memory of the ancestors. Sixth, all of these elements in the Yoruba universe- God, lesser deities, ancestors, nature spirits, destiny-are therefore recognized as causal agents, and explanations of events, especially extraordinary ones, are made by appeal to them.

Seventh, it follows that the causes of illness may be traced to the action of these causal agents, and that success in maintaining good health depends on how far one is able to live in harmony with the various forces, with nature and one's social environment. Usually illness occurs when there is a disruption of the balance, and good health will be restored only by restoring the balance.

In traditional Africa, there is a holistic conception of health. A person is healthy when they are able to perform their normal daily routines; otherwise they are not. To be healthy is to be physically, mentally, spiritually and socially balanced. A person who is able to perform his/her daily routines may have contracted a disease without knowing, but this does not make him unhealthy, according to this conception, since to be unhealthy is to be ill and this requires the consciousness of the individual.

For the Yoruba, for instance, a person has Alaafia (health) when she is physically, socially, psychologically and spiritually well-balanced. The absence of health is "aisan", the state of not being

well or alright. It is the whole body that is well or ill and this is the object of attention of the traditional doctor. The latter does not diagnose specific parts; his interest is in what could be responsible for disrupting the balance. This usually takes him from the world of nature, to the spiritual world in which the various forces referred to earlier are active agents. The doctor is also interested in the patient's background including social and spiritual relations. For it is believed that most illnesses occur when there is a breakdown in social relations. A person who is suddenly sick after an illicit affair with a friend's spouse will have to confess and pay the necessary damages. Since there is a general belief, based on the views about reality, that such a conduct attracts the wrath of the ancestors, the guilt conscience and anxiety syndrome may be responsible for much of the illness.

It is to be noted, however, that the traditional doctor also has their own account of germ theory. Therefore, the attribution of illness to spiritual forces does not exclude diagnosis in terms of germs. The Yoruba term for germ is "kokoro" and herbs are prepared to get rid of them. Indeed, this is the first level of analysis and it is usually undertaken with an examination of the social-spiritual relations.

The examination of social-spiritual relations may lead the healer to assign some role to supernatural causes in the explanation of an illness. Thus, it may be suggested that a deity needs to be appeased, or that an ancestor is angry or that the patient's "ori"- personal deity- has to be placated. This is where the traditional healer presumably runs counter to the tenets of scientific medicine. It is usual for the healer to combine primary (non-mechanistic) with secondary (mechanistic) explanation and to combine remedies suggested by both. However, the fact that he has invoked a supernatural cause is enough to cause concern for our scientific spirit. Yet, there is at least a comparative procedure in scientific medicine, in the realization that stress from whatever sources may reduce the

body's resistance to disease.It is also realized that the state of mind of the patient regarding her belief in the efficacy of the drug prescribed has a serious effect on what she gets out of it. This is not radically different form the presupposition of the traditional African doctor. The importance of the patient's psychological disposition, which derives from her assumptive frame of reference, suggests a combination of both primary and secondary explanation models in the diagnosis of illness.

In the specific case of sickle cell disease, these cultural factors play a significant role in the reaction of patients and relations to the disease. Many traditional Nigerians suffering from the disease find it difficult to reconcile themselves to the scientific prognosis, ascribing their conditions to various factors, including machination of enemies, wrath of gods and fate. In such a situation, genetic counseling may have to take account of the assumptive frame of reference of the people. For instance, the phenomenon of children who die shortly after birth on a repetitive basis with a particular mother, is refered to as "abiku" by the Yoruba and "ogbanje" by the Igbo. This phenomenon may in fact represent cases of the sickler child who does not survive to adulthood. There is also the concept of "abiku agba" (adult abiku), and this may also refer to the case of the adult sickler who survives childhood only to die as a teenager. The belief is that an abiku child, like every other person, has a spiritual partner in the spiritual world. However, unlike other persons, the abiku's spiritual partner wants them to be together and therefore recalls her prematurely. Also, following the belief in destiny, the abiku child may be said to have chosen to be born and die prematurely over and over again. The remedy that the parents revert to, given this mind-set, is the traditional doctor, who attempts a spiritual cure, based on the shared perspective. In the final analysis, an untimely death which defies any naturalistc explanation available in the world view, is attributed to *amuwa olorun (*will of God).

In the traditional communities of many African societies, the idea of a pre-marital screening

for sickle cell disease is not common, at least, not in the Western sense. Though families screen the family background of their prospective in-laws, these are done to ensure that their child is not marrying into a family that has been afflicted with cases of insanity, leprosy and such other diseases that are considered dreadful. Since the symptoms associated with sickle cell disease are not usually visibly demonstrated in entire extended families, they are not the focus of traditional screening. It is therefore difficult to have traditional people understand the implications of such screening. For them, it is like undermining the power and authority of God. In any case, many traditional people do not have absolute confidence in the western health system.

Among Westernized Nigerians who understand the ramifications of the disease and its implications for a happy married life, it is also difficult to accept the need for screening. For one thing, many in this group get into a relationship because they are in love. The fear of discovering a fact that could put a clog in the wheel of love is therefore real for some people. Second, among this group, there is a growing number of "born-again" Christians, who would rather not doubt God's plan for them by going through a screening procedure which , in their belief, does just that.

These religio-cultural beliefs do not, however, limit the impact of the condition on its victims, with dire social and psychological consequences. Patients lose their self-esteems, parents go through emotional stress, and in the end may lose their children only to blame it on destiny or the machination of the enemy, sometimes splitting extended families. I had to intervene in a tragic case involving another first cousin and my aunt. My cousin had lost his wife to cancer. He remarried after two years, but apparently the new wife has sickle cell disease, and my cousin has the trait. Their first child died before he turned one. The second was still- born. My cousin then accused my aunt of having a hand in these tragedies, and he packed his belongings away from the family house. When I intervened, he

reminded me of the various incidencies, and how her own two brothers had died at very young ages and her sister was an invalid till she died at child birth. There are many such cases, and these cases would seem to point to a serious need for genetic counseling.

As discussed above, there are cultural factors that impinge on the usefulness of genetic counseling. But there are also structural problems. The majority of Nigerians live in rural areas. Many of them do not have access to hospital facilities, or they do not take advantage of such facilities. Those who take advantage do not have access to genetic counseling because of the limitations on the services provided in such hospitals which are, basically, primary health care facilities. Still, it is important that physicians and pediatricians take up the role of genetic counseling in the local hospitals in lieu of trained counselors. Local priests and religious leaders also have important roles to play here, though in many cases religion is a limiting factor. I know of a university professor whose spirituality prevented him from accepting the need for hospital care for himself or the family.

Traditional Yoruba and other groups in Nigeria have ambivalent attitudes to the use of genetic knowledge. There are some aspects of the implications of genetic knowledge which they can relate to. In most cases, an extremely "malformed" new born baby will be quietly disposed of without qualms, but mild malformations will be spared. A genetic problem discovered late will be taken as God's purpose. The use of genetic knowldge for choice of sex is not looked upon favorably because it is considered as tampering with the work of God. Artificial insemination is not available to the majority because of the cost, not because of any cultural belief. Surrogate parenthood is not all that strange since, in some cases, the tradition recognizes the practice in polygynous marriages.

The suspicion of African Americans about screening programs for SCA arises from their suspicion of the system in general. From the Tuskegee experiment to the horror of involuntary

sterilization, African Americans have cause to doubt the good faith of the establishment. The furor over the screening programs for SCA is due in part to this. It is also due, however, to the negative propaganda, erroneous information and confusion surrounding the prevalence of the condition. In the light of the strong cultural and spiritual ties that bind African Americans to the motherland, it is to be expected that many of these perspectives are also shared by them.

Conclusion: A thicket of moral quandary

Is mass screening really justified? Must it be obligatory or voluntary? What principles must guide any screening program? And what standard of reasonable care ought to be established? From the benefit of hindsight, it seems clear that many of the screening programs established in response to the public awareness of SCA were developed in haste and without adequate counseling program.(Westring and Andrews, 1974). The first point worthy of note then is that the screening programs ought to take the interests of the community into account. This is in accordance with the important value of well-being, a derivative of the principle of beneficence. It stands to reason to expect that a screening program that is aimed at benefitting individuals will take their interest and well-being into consideration. This will certainly require respecting them as persons irrespective of their conditions. It also follows therefore that the results of such screening should be held in confidence and that the privacy of the screenees must be protected to avoid stigmatisation and discrimination. In any case, considering the primacy of the principle of autonomy, such screening tests cannot be mandatorily imposed. Individuals ought to have the right to determine whether or not they want to make themselves available. Such decisions, needless to say, can and, perhaps, ought to be made in conjuction with family and extended family members in the tradition of African family system. And though, some might see this as an infringement on individual autonmy, it is not quite so,

especially in such a case with repercussions for the family. In the case of SCA, it does not appear that the larger society has a more pressing interest than the families to warrant the imposition of mandatory screening . Therefore, there is no obvious conflict between individual and social welfare.

Other moral problems are not easily dismissible. Consider the position of the physician who just discovered an apparent case of non-partenity after a child tested positive and the father is not a carrier. Should he/she reveal this information? How about the dire consequences for the family which has appeared stable? And suppose they were not given the true information, what if the couple decides against having more children, thinking that they are both responsible for the positivity of the child's test? Are there absolute values here that cannot be overridden? It is not clear what an acceptable moral course should be, except that the physician has to take into consideration the interest of the child and of the apparent stability of the family, without losing sight of his/her professional responsibility. Untangling this thicket of moral quandary is itself a major repsonsibility.

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