The National Burden of Sickle Cell Disorder

And The Way Forward

By

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Molineaux and co-workers[1], after some research in northern Nigeria, correctly summarised the situation when they wrote in 1979, "***There is no other known inherited disorder present at such high frequency in a large population and of comparable severity as sickle cell anaemia in Africa. With rising standards of living and control of malaria, sickle cell anaemia will become an immense medical, social and economic problem throughout the continent***." In sheer numbers, Nigeria has the largest burden of sickle cell disorder (SCD) in the whole world. Carriers of the sickle cell gene (Hb AS) have, over the past centuries, flourished and multiplied in tropical sub-Saharan Africa because their carrier status protected them from succumbing to the deadly falciparum malaria prevalent in the Region. In other words, they enjoyed a survival advantage over their peers who had not inherited the gene (Hb AA) and those who had inherited it from both parents and therefore had sickle cell anaemia (Hb SS).

Although countries around Nigeria also have an S gene carrier frequency of about 1 in 4 of their populations, Nigeria's large population has ensured that over 40 million Nigerians are healthy carriers of the S gene. This number of carriers far exceeds the total population of every other affected African country and indeed, of several of them put together. Consequently, about 150,000 Nigerian children are born each year with sickle cell anaemia (HbSS), the prevailing type of sickle cell disorder (SCD) in this Region. Survival of these children beyond

childhood is largely dependent on their access to appropriate care and as most of them are born into poor underprivileged families, very few of them survive childhood[2]. On the other hand, the survival of those with access to good care, at all ages, is steadily improving although many challenges to their quality of lives and life spans still remain.

In this paper, I shall attempt to examine Nigeria's response to the burden of SCD and discuss some of the issues pertaining to its management and control.

The Level of Awareness.

For an old disorder with a birth rate of 1 in 50 babies, the level of awareness is relatively low. This is not because of a lower birth rate previously, but because the affected children rarely survived childhood and were therefore less likely than now, to be encountered in secondary schools, in universities and in the workplace. Their peers and relatives now have the opportunity to observe their periodic pain crises and, sadly, occasionally, their premature deaths. This familiarity has heightened awareness. The other factor boosting awareness is the development of Sickle Cell Clubs, with attendant publicity, over the past two to three decades.

Response to the National Burden of SCD

Despite its 2% birth incidence[2], the estimated population of SCD affected persons in Nigeria is only about one million, owing to a high rate of premature

deaths. Unfortunately, the increasing awareness has not been matched by the development of a well resourced national policy. This has curtailed the dissemination of accurate and unbiased public information and education about the disorder and has fuelled the growth of myths, misinformation, inappropriate treatment, frustration and stigmatisation. The frustration has kindled the desire in many Nigerians to ***do something about sickle cell disorder***. What needs to be done often appears deceptively easy and is usually not fully thought out and remains a subject of confusion and controversy even among health care workers. In this regard, one frequently hears talk of eradication of the disorder while the wider context of management and control is invariably overlooked.

Selective Mating and the Eradication of SCD

Eradication of SCD by selective mating appears so logical and simple that many individuals, religious bodies and charitable organisations have tried to implement it themselves by screening young people and instructing those shown to be carriers to avoid choosing spouses who also have the sickle cell gene. Some churches even refuse to marry them.

This strategy is based on the false assumption that SCD can be easily eradicated by mass population screening of haemoglobin genotypes as a basis for coercive selection of spouses, The Military Administrator of Oyo State, had, in 1995, proposed a punitive edict aimed at prohibiting marriages between carriers of the sickle cell gene, but the conference of Solicitor–Generals in Nigeria thwarted its

introduction by declaring that it was unconstitutional and offended the human rights charter to which Nigeria is a signatory[3].

The reality however, is that enforced selective mating of couples has never been shown anywhere in the world to have reduced the incidence of any inherited disorder. Attempts to introduce it by the Church in Cyprus, for the control of thalassaemia (also an inherited haemoglobin disorder), there, led to increased anxiety and stigmatisation of affected persons and of healthy carriers of the gene[4, 5]. This, in turn, led to widespread denial and falsification of haemoglobin genotype results among carriers who wanted to marry each other. What the Church in Cyprus now does is to ensure that all couples have been counselled on thalassaemia before marriage.

Another popular but false assumption, is that, all marriages between carriers of the S gene are contracted in ignorance. As health workers in the specialty, we in fact see many marriages contracted between carriers who are both well educated and informed about their Hb status, and between them and persons affected with sickle cell anaemia. I know at least two Nigerian doctors who have married their SCD affected patients. One of the doctors was himself a carrier of the S gene. Although I knew the other doctor quite well I did not bother to ask whether he was also a carrier, as that would have seemed intrusive and perhaps, even judgemental. A few marriages contracted between affected individuals (e.g. SS and SS; SS and SC) have also been reported in Nigeria and elsewhere.

A research study of the effect on choice of marriage partner of informing young people and their families of their carrier status was carried out in the Arta area of Greece, where 20% of the population carry either thalassaemia or sickle cell gene. All young people of marriageable age were screened and counselled, and counselling contact was maintained for a two-year period. When the pattern of marriages was assessed at the end of the period, screening had had no measurable effect on choice of partner [6].

These examples simply go to show that there is more to marriages than Hb genotypes. We must ask ourselves whether we have got the moral right to choose spouses for others or simply to allow them to make informed choices. **Given our present state of knowledge, eradication of SCD can only be feasible by the unthinkable genocide of all healthy carriers of the gene or their enforced banishment from the country!**

Stigmatisation

Stigmatisation and discrimination are products of ignorance and insensitivity. They are not only felt and encountered by affected persons and their families, but also, to a lesser degree, by carriers of the S gene. Some examples may blow your minds.

A secondary boarding school student stopped taking his daily medication because his schoolmates derided him for doing so. He was too ashamed to inform them of his condition and need to take daily medication.

A University of Lagos lecturer with two affected children himself was trying to organise a Sickle Cell Club on the campus for the benefit of affected students. He publicised his mission asking for affected students to come forward. Many did not do so, for fear of identification and stigmatisation. Nobody wants to be different or be regarded as an object of pity, or feel by implication of widely expressed “solutions” that his birth was an unwanted mistake which should not have happened if only both parents had known better and avoided marrying each other. ‘*What a pity that these people were born. Let us inform everyone to avoid marrying so called “incompatible spouses” who can cause the births of others like the”*. Some of these statements and sentiments expressed by their peers are, of course, well-meaning, but, on reflection, highly insensitive and borne out of ignorance of some realities. So, the affected student feels like an unwanted alien and would like to hide in denial. You can imagine the resulting loss of confidence and self-esteem.

A young Nigerian woman was devastated when the overbearing father of her fiancée prevented their marriage on discovering that, she, like his son, was a carrier. She told us that, since then, she had changed her Hb genotype to AA.

In early May 2009, the Punch Newspaper reported that a 10 year old girl was found starved and sleeping on the streets in Yenagoa, Bayelsa State. Some kind-hearted people rescued her and learnt that when her parents separated she was taken by her mother. When her mother went to live with her new boy friend, he rejected keeping her as soon as he found out that she had sickle cell anaemia. She was ejected, even though her father had assured her mother that he would continue to bear the cost of her maintenance and education.

While still trying to fathom that cruelty to an innocent child, a friend of mine in Lagos told me that he had offered to foster, accommodate and pay all the living and educational expenses of the 7 year old son of a recently deceased friend. Another friend who was inexplicably winking at him while he was offering to do so later told him why he was trying to attract his attention. It was to warn him against fostering the boy whom he knew had sickle cell anaemia.

The cruelty factor notwithstanding, the last two cases would suggest that many people are not aware that people with SCD can live long useful lives, and, in this country, are found in all the usual civic professions as well as in politics, where, I know at least two, who have risen to the positions of State Governor and State Commissioner respectively.

Marketing of Worthless Remedies

Charlatans and other adventurers have, not unexpectedly, flooded the market with drugs that are worthless to people with SCD. **These drugs have been condemned by the Nigerian Society for Haematology and Blood Transfusion (see their Communiqué on page 44 of The Punch of August 28, 2008).** Many people buy these drugs because they bear NAFDAC registration numbers on their packages. When in 2008 we met Professor Dora Akunyili, the former Director-General, she insisted that **NAFDAC registration was meant only to assure the public of the safety of the drug for human consumption and is not an approval of its efficacy in the treatment of the condition or disease for which it is advertised.** This is hardly understood by members of the public and the Sickle Cell Foundation Nigeria is still in discussion with NAFDAC over this.

Multiplicity of Sickle Cell NGOs

Diverse sickle cell NGOs, some merely duplicating rather than consolidating efforts, and others, worryingly with dubious motives and unclear capacity, have started to emerge. Many are driven by an emotional over simplified vision of eradication without training in genetic counselling or in any aspect of its management. This tendency, if unchecked will disseminate mixed messages that can confuse members of the public and thus become counter-productive.

The Way Forward

Coordination and Harmonisation of Institutions and Programmes

The Sickle Cell Foundation Nigeria (SCFN) was established to address the problem of SCD in Nigeria in a systematic, scientific and sustainable manner. It has achieved its first goal of developing, in accordance with WHO recommendation [7], a comprehensive National Sickle Cell Centre in Lagos. The intention is for each State to have a collaborating Sickle Cell Centre that will supervise and coordinate the programmes and activities of all Sickle Cell Clubs within that State.

Training and Capacity Building

The National Sickle Cell Centre is engaged in training various cadres of health care personnel in order to improve the standard of preventive and curative care available to persons with SCD.

Genetic Counselling

Genetic counselling is not marriage counselling. Sickle cell disorder should be sensitively managed within the community. Otherwise, people living with SCD or who are healthy carriers of the gene may feel stigmatised and be tempted to conceal, deny or falsify their haemoglobin (Hb) genotype status. This state of affairs can be counter-productive to efforts aimed at managing the disorder in the community. The importance of genetic counselling in preventing these undesirable outcomes is well established.

In counselling, the client is a person with SCD, his close family member or a healthy carrier of the sickle cell gene or the gene of some other haemoglobin variant such as Hb AC. The role of the counsellor is to ensure that the clients are

given accurate unbiased information necessary to assist them to reach their own decisions on reproductive behaviour or any other course of action related to the disorder. Their decision must be respected and supported by the counsellor and confidentiality must be maintained. Failure to do so usually alienates the client to the detriment of the relationship between counsellor and client.

Effective counselling is informative, confidential, non-directive and supportive. It requires skills that are best acquired through appropriate training and experience. Doctors who see many patients with SCD should endeavour to acquire the necessary skills and, if they have many patients, employ the services of one or more trained nurse-counsellors.

The Principles of Genetic Counselling

Informative

Genetic counselling should provide accurate and unbiased information about the disorder. To do this effectively, it is necessary to find out the prior knowledge and perceptions of the client concerning SCD. Any myths, wrong or superstitious beliefs should be uncovered, corrected and then replaced with accurate information. Depending on the level of understanding and the cultural background of the client, this informative process should be conducted in an appropriate language with illustrations and imagery to enhance the spoken word. More than one counselling session may be necessary and a summary of the session should be augmented with appropriate written materials, which the client can take home.

Confidential

Confidentiality must be maintained so that the client will be assured that his privacy is respected and kept. This will encourage full disclosure and create a trusting and beneficial relationship with the counsellor.

Non-directive

Counselling is not about telling clients what to do. It is about giving them accurate information to help them reach their own decisions. Directing them to take a particular course of action is often counter-productive. If the client later regrets the decision he or she will resent the directive and blame the counsellor for it. Where the client decides to ignore the directive the relationship with the counsellor becomes threatened and may cease to exist. It is unethical for the counsellor to subtly direct the client by providing unbalanced and biased information.

Supportive

The counsellor should empathise with and support the decisions taken by the client. The counsellor should help the client explore self, feelings, attitudes, and values in relation to the diagnosis. The client should feel secure enough to return and discuss possible consequences of his decision. The counsellor should refer the client to where he can obtain further information, social services or appropriate health care advice.

Objectives of genetic counselling

The counsellor should

1. ensure that the client has obtained an accurate diagnosis of his condition

2. ensure that the client is feeling well and comfortable and not at that moment in need of medical attention

3. ensure that the client knows what the session is about and how long it might last

4. take a family history pertaining to sickle cell or other inherited haemoglobin disorder

5. establish the prior awareness, knowledge and perceptions of the client about SCD and sickle cell trait

6. tactfully but firmly dispel myths and misinformation held by the client

7. ensure that the client understands the inheritance of SCD and how it might affect him or his children depending on the Hb genotypes of their parents

8. ensure that the client understands the likely clinical course and treatment needs of patients with SCD

9. ensure that the client understands that carriers of the sickle cell gene are healthy persons who are unlikely to fall ill because of their status.

10. inform the client about existing facilities for health and social care and provide a referral where necessary

11. provide the client with a spectrum of careers that are compatible with living with SCD.

12. encourage affected clients and their parents by referring to successful role models living with SCD in the community

13. summarise the session, allow for questions, handover educational materials and make appointment for a follow up session

Research, Research, Research

The importance of research is underestimated in Nigeria due largely perhaps to our under-development. There would be no progress without research into all aspects of sickle cell disorder. But for the research carried out in America, the present state of knowledge which has improved the quality of life and life-expectancy in sickle cell anaemia would not have been possible. As the country with the largest burden and blessed with many bright scientists, we need the financial resources to devote to research that should convert sickle cell anaemia to a disorder like hypertension which is compatible with normal duration and quality of life.

Development of a National Policy on SCD

The SCFN has constituted a **Nigerian Sickle Cell Expert Advisory Committee**. The committee's first objective is to consider, initiate and revise policy and strategies appropriate to the management, prevention and control of sickle cell disorder in Nigeria. Other objectives and decisions can be found on the SCFN website www.sicklecellfoundation.com.

The First World Sickle Cell Day – 19 June, 2009

Following decades of advocacy, the United Nations in December 2008 resolved to recognise sickle cell anaemia as a public health problem and to mark the World Sickle Cell Day (WSCD) on June 19, every year, starting from 2009. The intention is that the WSCD will create awareness and draw attention of

governments, donors and all stake holders every year, to what has been achieved and what needs to be done to address SCD in each of the affected countries.

Immediate Goals and Prospects

Our immediate goal would be for Nigerians with Hb SS to attain the average life expectancy in affected Americans (53 years) and eventually, with appropriate research and service programmes, surpass it and make the pain crisis a rare event, which can be rapidly relieved when it occurs. The USA now has 3 million healthy carriers of the sickle cell gene; 2,000 children born annually with sickle cell disorder and about 100,000 persons living with sickle cell anaemia. It is instructive that in the USA the average life expectancy for someone with sickle cell anaemia (Hb SS) was 7 years in 1974 and it is now 53 years. The average quality of life has also improved and these have been attributed to their development of sickle cell centres and the introduction, through research, of prophylactic and therapeutic interventions.

Nothing good comes easy and lofty goals must be adequately funded. It is remarkable that over 80% of the annual budget of the Sickle Cell Association of America is derived from thousands of small individual donations rather than from a few large corporate donations.

Many Nigerians agree that we should tackle our national burden of SCD, but not many regard themselves as part of the solution. A friend donated an unsolicited cheque of half a million naira to the SCFN last year. Another couple spearheaded the contribution of a large generator to the National Sickle Cell Centre. Contribution of professional services of solicitors, auditors, health educationists and publicity specialists are also welcome. We hope that with support to the National and State Sickle Cell Centres as in Owerri and Benin and others yet to be developed, we can all help show that Nigerians are capable of competently and sustainably addressing their problems. We hope that many Nigerians will thus be encouraged to contribute to reducing the national burden of sickle cell disorder in our country by supporting and sustaining the effort of the Sickle Cell Foundation and accredited collaborators in each State of the Federal Republic of Nigeria.

Enquiries about training in counselling and other services can be obtained from the Sickle Cell Foundation Nigeria, National Sickle Cell Centre, Ishaga Road (*opposite LUTH*), Idi-Araba P O Box 3463. Surulere, Tel +234 803 584 6666, +234 17621522; 01 725 3957 – 8; website www.sicklecellfoundation.com email: **info@sicklecellfoundation.com**; scf-ng@hotmail.com

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