SIERRA LEONE SICKLE CELL DISEASE INITIATIVE: A WHITE PAPER

BACKGROUND

Sickle cell disease (SCD) is one of the most common inherited blood disorders in the world. SCD is caused by the inheritance of two copies of the gene encoding hemoglobin S, a protein that results from a missense mutation in the β-globin subunit of hemoglobin A, or the co-inheritance of the gene for hemoglobin S and another abnormal or nonfunctional hemoglobin gene.

The worldwide burden of SCD is rising. It is estimated that more than 300,000 babies with two copies of the sickle hemoglobin gene are born every year. It is also estimated that in 2050, the number of newborns with SCD will increase to over 400,000 per year. These estimates do not reflect the incidence of other sickle hemoglobinopathies (hemoglobin SC, SE, Sß thalassemia, etc.) nor numbers of all age-affected individuals. Between 2010 and 2050, it is expected that about 14.2 million affected babies will be born worldwide. This is probably an underestimate, but it provides the best current approximation of patient populations afflicted with the disease. Perhaps more importantly, more than three-quarters of those affected with SCD will be born in sub-Saharan Africa.

Patients with SCD have a multitude of complications, including pain, infections, stroke, sepsis, acute chest syndrome, multi-organ damage, and early death. In high-income countries including the United States, which house less than 10% of the global burden, early mortality has been significantly reduced over the past few decades, with more than 90% of people with sickle cell surviving beyond 20 years of age. This can be attributed to widespread implementation of a range of interventions, including newborn screening, penicillin prophylaxis (because sickle cell increases susceptibility to pneumococcal infection), pneumococcal vaccination and parental education. By contrast, the World Health Organization (WHO) estimates mortality of those younger than five to be more than 50% in low-income countries including Sierra Leone; and has estimated that SCD accounts for up to 9% of all deaths in children under five in sub-Saharan Africa. Causes of mortality in these patients include malaria-associated severe anemia, pneumococcal and other bacterial infections. In addition, the disease is associated with significant health challenges including recurrent debilitating pain, chronic anemia and stroke.

The morbidity and mortality associated with SCD in sub-Saharan Africa is a consequence of **limited health resources and infrastructure**. Examples include a widespread lack of **newborn screening programs, pneumococcal vaccines, and penicillin prophylaxis regimens**. In addition, while high regional disease prevalence would be expected to facilitate epidemiologic, translational, and clinical research, most sub-Saharan African nations lack the means and capacity required to pursue such investigations.

PURPOSE

This white paper proposes the establishment of a Sickle Cell Disease (SCD) Initiative in Sierra Leone by establishing a collaborative agreement between the University of Cincinnati and the Jericho Road Community Health Center. The collaborative agreement will provide the basis for healthcare capacity building activities, specifically the development of a comprehensive plan to establish an infrastructure upon which a pilot SCD screening, health maintenance and wellness programs would be developed. The collaborative agreement will also provide the framework within which SCD-related epidemiologic, translational, and clinical research and educational initiatives will be developed. The collaborative agreement will between a clinical partner (Jericho Road Community Health Center) and an academic partner (University of Cincinnati**). It is anticipated that additional collaborative agreements and partnerships will be developed with other academic partners viz., Cincinnati Children's Hospital Medical Center (Ohio) and Medical College of Georgia (Georgia).**

OBJECTIVES & GOALS

The collaborative agreement undergirds the following objectives:

1. Create and implement a pilot sickle cell disease program in Kono District (Sierra Leone).
2. Create and implement a pilot educational and training program for local medical, nursing, and other healthcare providers caring for children with sickle cell disease in Kono District (Sierra Leone).
3. Develop research initiatives to demonstrate the disparities in SCD outcomes seen in children residing in Sierra Leone compared to children with SCD in the United States.

To advance these objectives, the following goals will pursued:

1. Create a pilot centralized, electronic, patient-consented, sickle hemoglobinopathy database that will facilitate registration and follow-up of SCD patients and serve as the backbone of the SCD clinical and research programs.
2. Develop SCD standards of care appropriate to the national (Sierra Leone) clinical needs and resource availability.
3. Organization of clinical skill development activities appropriate to national (Sierra Leone) needs and resource availability
4. Implementation of preventive/therapeutic practices (i.e., newborn screening, genetic counseling, pneumococcal vaccines, penicillin prophylaxis, etc.) appropriate to national (Sierra Leone) needs and resource availability.

5. Organization of research skill development activities and planning for SCD cohort studies.
6. Collaborate with international, national, and local SCD support groups to advance SCD programs in Sierra Leone.
7. Collaborate with local, regional, and national governmental agencies to advance SCD programs in Sierra Leone.
8. Seek extramural funding to support SCD clinical and research programs in Sierra Leone.

CALL TO ACTION

This White Paper acknowledges that limited resources for diagnosis and treatment, combined with a dearth of extant strategies to combat SCD, have led to a paucity of care for children affected by the disease in sub-Saharan African nations. In Sierra Leone, in particular, SCD remains an important but largely neglected risk to child survival. The probability of early death among children born with SCD in Sierra Leone might be as high as 90% in rural areas where access to health care is limited, but closer to 50% in populations with somewhat better access to health care and lower exposure to infectious diseases.

The situation in Sierra Leone is also compounded by the lack of public understanding of SCD, which perpetuates social stigma and myths about disease causation and results in few people seeking appropriate treatment. In 2010, the WHO Regional Office for Africa commissioned a strategy document that spelled out guidelines for actionable steps to combat SCD, but these are yet to be translated into government action in several sub-Saharan African countries. Consequently, the burden of SCD will continue to rise, placing an increasing strain on limited national healthcare resources.

This White Paper also acknowledges the need to find affordable, evidence-based solutions that can be integrated into existing healthcare systems to ensure their sustainability. And that action is needed on several fronts as follow:

- First, public health programs, particularly newborn screening, health education, and immunization are urgently needed.
- Second, pilot programs including the careful collection and analysis of SCD data and publication of outcomes are essential.
- Third, studies to quantify the public health burden of SCD in Sierra Leone need to be conducted. Such studies require interdisciplinary collaboration among different types of clinicians, researchers and cooperation among different public health programs conducting representative population surveys, including malaria, and nutrition.

- Fourth, there is the urgent need for public-awareness campaigns to combat stigma and misunderstandings about SCD, and to provide opportunities for genetic counseling and initiatives that promote disease avoidance.

CONCLUSION

Many effective interventions for SCD are affordable and within grasp. However, the public will to act needs to be harnessed. Academic organizations, healthcare providers, grassroots advocacy organizations both nationally and internationally should be emboldened to place sickle-cell disease high on their agenda. Collaborative arrangement and broad partnerships as proposed in this White Paper are critical to implementing sustainable solutions that may reduce the burden of SCD in Sierra Leone.

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