Chapter 4
Global Genomic Response to Neonatal Screening

ABSTRACT

In the age of genomic science, neonatal screening for sickle cell anemia involves critical ethical legal, social, and financial implications. The screening for early identification of children most likely to face the complications associated with sickle cell disease was designed to enable parents utilize the state of the art scientific curative care to enable their children to live economically productive lifestyle. However, screening must not be used for discrimination and refusal to insure patient with sickle cell disease. The availability of bone marrow transplant and gene-therapies are clinical options to assist at-risk patients.

PART I: ETHICS OF NEONATAL SCREENING FOR SICKLE CELL ANEMIA

Ethical, Legal, Social, and Financial Implications of Neonatal Screening for Sickle Cell Anemia in Sub-Saharan Africa in the Age of Genomic Science

This project was designed to identify the legal implications associated with the management of sickle cell in newborns and to discuss the socio-cultural implications in the management of sickle cell in newborns. In assessing the ethical components of neonatal screening for sickle cell disease one must consider the implications for screening at-risk subject bearing in mind those there relevant therapeutic interventions for the management of the disease. Besides, conscientious efforts are needed to determine the ethical and the financial implications of treating newborn with sickle cell anemia as a public health priority among physicians, administrators and health educators in Sub-Saharan African (SSA) nations.

Introduction

Sickle cell anemia as a major disorder occurs from a change in valine to glutamine substitution in the sixth amino acid position of the beta globin chain. From biochemical analysis, this results in quaternary structure of hemoglobin molecule which under conditions of deoxygenation creates aggregation of

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hemoglobin into long polymers which align themselves into rigid paracrystalline gels and distorts the red blood cell into sickle shape. If one of the two beta globin genes is affected, the ensuing condition is characterized as sickle cell trait (SCT) and if both genes are involved the resulting situation is sickle cell disease. From over four decades of clinical practice, Konotey-Ahulu (1974) provided succinct clinical definition of sickle cell disease. He characterized sickle cell disease as " the possession of two abnormal allelemorphic genes related to hemoglobin formation, at least one of which is the sickle cell gene, the genotype constituting sickle cell disease being SS, Sc, S Thal, SE SF 'high gene' and SD" (Konotey-Ahulu, 1974). Multiple comparisons of normal hemoglobin and sickle cell variants are illustrated in Figure 1. The clinical abnormality caused by sickle cell anemia includes manifestations of severe pain, leg ulcers, and swellings of the joints, excruciating pains in the abdomen, arms, fatigue, and sometimes death (Calloway & Harris, 1977; Ebomoyi & Srinivasan, 2008).

The observable differences between sickle cell disease and SCT were discovered 64 years ago ((Paulings, Itano, Singer, & Wells, 1949). The differences lie in the quantity of erythrocytes of SCT and sick cell disease and the involvement of greater reduction in the partial pressure of oxygen which is required for a significant quantity of the trait to sickle than sickle cell disease. In SCT, a person inherits one normal hemoglobin gene (A) from one parent and one abnormal gene (S) from the other parent. With regards to sickle cell disease, this hereditary disorder follows the Mendelian pattern. The distribution of sickle cell variants in SSA is illustrated in Figure 2.

In the 21st century, the occurrence of sickle disease has become a worldwide public health burden and the case fatality from sickle cell anemia has attained epidemic proportion which makes SSA nations the sickle cell epi-center of the world. Reports from the World Health Organization (WHO) (2006) have confirmed how sickle cell disease is particularly common among people whose ancestors come

Figure 1. Various shapes of hemoglobin and sickle cells