Research Article

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Sickle Cell Anemia in pediatrics African

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1. Abstract

Sickle cell disease is a genetic blood disorder affecting Red-blood cells. Is an umbrella term for a group of life-long debilitating auto somal recessive disorders that are caused by asingle-point mutation (Glu→Va) that results in polymerization of hemoglobin (Hb) and reversible sickle-shape deformation of erythrocytes. The most common form of SCD is caused by homozygosity for the -globin S gene mutation (SS disease). The United Nations has recognized SCD as a global public health concern, and the World Health Organization (WHO) recommends that 50% of member states will have established SCD control programs by 2020 (World Health Organization, 2006) [1] is the central path ophysiology of this disease, although the importance of chronic anemiah emolysis and vasculopathy has been establish. One of the main problems of sickle-cell disease in children is the development of cerebrovascular disease and cognitive impairment, and the role of blood transfusion and hydr oxycarbamide for prevention of these complications is starting to be understood. Recurrent episodes of vasoocclusion and inflammation result in progressive damage to most organs, including the brain, kidneys, lungs, bones, and cardiovascular system, which becomes apparent with increasing age [2]. Clinical manifestations of SCD occur early in life, are variable and are modified by several genetic and environmental factors. Nearly 500 children with SCD continue to die prematurely every day, due to delayed diagnosis and /or lack of access to comprehensive care in sub -Saharan Africa, a trend that needs to be urgently reversed [3]. A simple representative and affordable approach to estimate SCD child mortality is to test blood specimens already collected through large population surveys targeting conditions such as HIV, malaria, malnutrition, and covering children of varying ages. Thus, although there is enough evidence to justify investments in screening, prophylaxis, and treatment for African children with SCD, better data are needed to estimate the numbers of children death preventable by such interventions, and their cost effectiveness [4].

2. Introduction

Sickle cell disease (SCD) is the world most frequent genetic disease caused by single mutation in beta-globin gene [5] (Glutamic acid-Valine) leading to the presence of abnormal hemoglobin (HB S). It is a recessive autologous disease, and reversible sickle-shape deformation of erythrocytes. This leads to increase hemolysis of erythrocytes and microvascular occlusion, ischemia perfusion injury, and tissue infarction, ultimately causing multisystem end- organ complication [5]. Sickle cell anemia (HBSS) is the most common and most sever genotype of SCD, followed by HbSC, HbSB0 thalassemia, HbSB+ thalassemia, rare and benign genotype [6]. In Africa Sickle cell disease (SCD) is reported to be associated with very high rate of childhood mortality, 50%-90% yet there is lack of reliable, up-to-date information. Quantifying the number of less than 5 child death from SCD in African countries is important to attract policy and support and resources for measures to reduce the burden of mortality [7]. In 2010, the WHO Regional office for Africa proposed a SCD strategy in official recognition of the fact that this disease is an important cause of child mortality in many African countries. for promoting widespread screening, health education, and treatment to prevent or manage SCD complication [7].

3. Classification

The most common subtype of SCD worldwide is homozygous SCD (SS). The distribution of SS allele (figure 1) has recently been mapped globally using detailed geo-referenced data and displays a close association with the historical distribution of plasmodium falciparum malarial endemicity [7]. With in Africa frequency of Bs, and accordingly SS, is highest in low-altitude equatorial region. The second sub type of SCD common in Africa is compound heterozygous for Bs and Bc (SC). The Bc allele is found almost exclusively among people of west Africa ancestry, being most common among those in Burkina Faso and Northern Ghana.Compound heterozygous with B+ thalassemia (SB-thalassemia) is a form of SCD that is believed to be rare in most of sub –Saharran Africa. In central, East, and Southern Africa, SCD is generally assumed to be synonymous with SS disease, although few studies have specifically looked for SB0-

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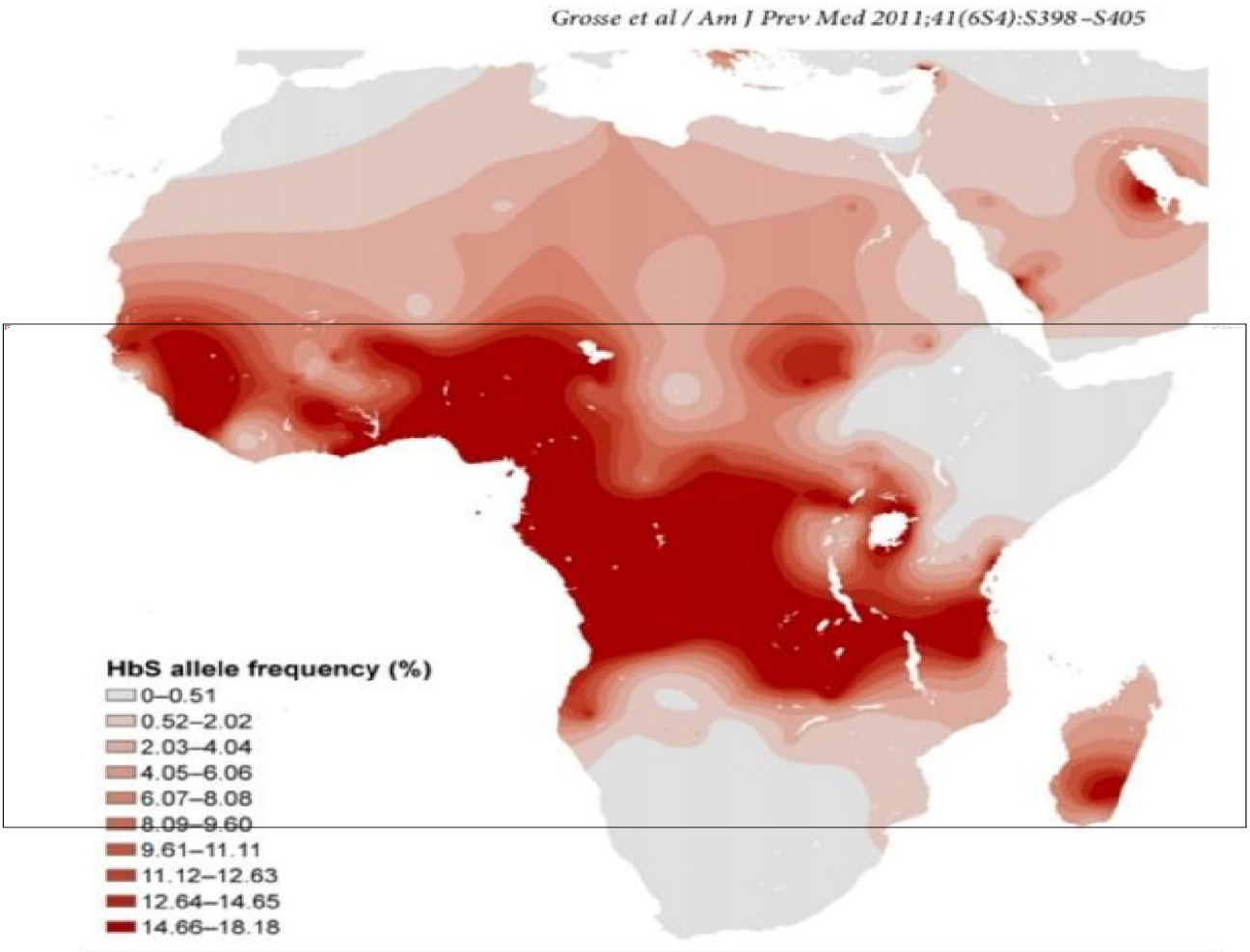

Figure 1. Map of the distribution of the β^S gene in Africa

Note: The map is based on representative indigenous population samples and is adapted from Figure 1b in Piel et al.[24] The figure shows the global distribution of the sickle cell gene and geographic confirmation of the malaria hypothesis.

thalassemia.

4. Discussion

This is the first study that present the rate and risk factors for mortality in SCA in Africa. In Africa, sickle cell disease (SCD) is reported to be associated with a very high rate of childhood mortality, 50 % - 90 %, yet there is a lack of reliable, up - to- date information. [1-7] The most frequently cited study was conducted in the Garki district in rural northern Nigeria in the early 1970s. Sickle Cell Disease in Children and Adolescents: A Review of the Historical, Clinical, and Public Health Perspective of Sub - Saharan Africa and Beyond. According to the systematic analysis of the Global Burden of Disease Study, 176,000 people die every year from complications linked to sickle cell disease [8]. Currently, life expectancy is estimated at over 50 years in the United States but less than 10 years in sub - Saharan Africa. Sickle cell trait (SCT), not categorized as a SCD, refers to a state where individuals are heterozygous for the sickle allele (HbAS, also known as carriers). It results from inheritance of one abnormal (HbS) allele from one parent and one normal (HbA) allele from another parent [9]. One in every five people has SCT in high - burden countries like Nigeria [10]. Earlier studies characterizing the DNA structure of the B – glob in locus of Hb S suggest that the mutation arose on at least three independent occasions in the African continent, referred to as ß - globin haplotypes. These haplotypes are determined by restriction fragment length polymorphism (RFLP) and were named after the ethnic group or geographic area where they were first described , namely, Benin (BEN), Senegal (SEN), Came roon (CAM), Central African Republic (CAR) or Bantu, and Arab - Indian (Al) haplotypes [11]. Atypical haplotypes have been described in an ethnically diverse Sudanese population with SCA [12]. Results of whole - genome sequencing reveal that the sickle allele had a single origin 259 generations back (7,300 years ago) in West Africa during the Holocene Wet Phase [11]. The distribution of the sickle allele correlates with the distribution of malaria [13]. World wide, nearly 5.5 million neonates are born with SCT annually, 65 % of them in the WHO African region, where malaria is highly prevalent. A 2014 multicenter study of 11,890 cases of severe Plasmodium falciparum malaria and 17,441 controls recruited from 12 locations in Africa (The Gambia, Mali,

Burkina Faso, Ghana Navrongo and Kumasi, Nigeria, Cameroon, Kenya, Tanzania, and Malawi) , Asia (Vietnam Oceania (Papua New Guinea) found that people with Hb AS have an 86 % lower risk of developing severe malaria [14] . Individuals with SCT are usually clinically asymptomatic [15] and have a normal life expectancy similar unaffected people, but are not necessarily free of adverse outcomes.

Accumulated evidence shows that they have a high isk of vaso - occlusive pain, hyposthenuria, proteinuria, hematuria renal medullary carcinoma, venous and pulmonary thromboem bolism, and rhab domyolysis following extreme physical exertion [16, 17-19] . Nonetheless, these Discussion: History of pain, depression, and sleep quality were longitudinal predictors of pain over 1 year in youth with SCD. Identifying longitudinal predictors of pain may lead to earlier identification of patients with a high - risk SCD pain phenotype and earlier medical, psychological, and behavioural interventions.

5. Conclusion

Sickle cell cisease is one of the most common monogenic diceacees world wide and is prevalent in SPANISH. It has been over a hundred years since the first SCD formal diagnosis from a dental student from Granada was made by Dr. James Herrick in Chicago (sergent, 2010). SCD has since ravaged the world in particular amongst people of Africa and mediterranean heritage [20] . Given the enormous current and future burden of SCD in SSA, the contribution that it makes to total child mortality, the condition has been historically neglected. In recent years some progress has been made in reversing this situation; however, substantial investment will be required to move SCD up the agenda to its right ful place as a priority disease for African government going forward. Some key areas for future work include better and more up to data description of burden of SCD of local scale and studies of the natural history of SCD among patients in SSA who are on routine treatment, with particularly emphasis on describing the incidence of the acute and chronic complications of disease in African context. While wide spread programs the early diagnosis and treatment of SCD in SSA could yield substantial benefits for the patient and their families a great deal more needs to be known about the local anthropological and social attitudes to SCD, and steps need to be taken to mitigate against any adverse consequences for affected population. Health care planner will need to be convinced of the benefits of greater focus on SCD through cost economic analysis of various approaches to diagnosis and treatment. [21] SCD it had been largely neglected as global health priority until 2006 when the world health organization recognized it as a global health priority. More than 500 children die every day because of delayed diagnosis and lack of comprehensive ca re for SCD [22]. Implementation of newborn screening in sub-Sahara Africa is still in infancy, antyet, early diagnosis facilitates early enrollment into comprehensive care program and family health education, hence reducing the morbidity and mortality associated with SCD in newborn.

Their use of Pocts, which are significantly cost - effective and accurate Screening of SCD in newborn their use should be integrated into existing preventive care programs such as immunization and HIV early infant diagnosis in high burden countries in SSA. [22]. Additionally, there is an urgent need to implement the wide-scale use of hydroxyurea in combination with approved disease modifying the Aries (L-glut amine, crizanlizumab, voxel otor) for children with SCD in SSA. Since curative therapies (HSCT and gene therapy) are distant options for the majority of African children with SCD, Countries should improve the access and quality of comprehensive care, to further enhance the survival and quality of life of patients with SCD, countries in SSA should invest in health systems research to stream the transition from. adolescent to adult care. [22].

6. Abbreviations:-

- SCD: Sickle Cell Disease
- DALY: Disability adjusted life year
- DHS: Demographic and Health Survey
- RFLP: Restriction Fragment Length Polymorphism
- HbF: Fetalhemoglobin (hemoglobin F)
- HbSS: Homozygous hemoglobin S genotype
- HIV: Human immunodeficiency virus
- SCT: Sickle Cell Trait
- PCV: Pneumococcal conjugate vaccine
- POCT: Point - of - care test
- SCA: Sickle cell anemia
- DNA: Deoxyribonucleic Acid
- SPARCO: Sickle Pan - African Research Consortium
- SPIN: Stroke Prevention in children with sickle cell anemia in Nigeria
- Hb: Hemoglobin
- HPFH: Hereditary persistence of fetalhemoglobin
- HU: Hydroxyurea
- NBS: Newborn screening
- RBC: Red blood cell
- SSA: Sub-saharran Africa
- USA: United States of America
- WHO: World Health Organization.

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