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Sickle-cell Anemia and Consanguinity among the Saudi Arabian Population

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Abstract

Sickle Cell Disease (SCD) is one of the most common severe autosomal recessively inherited blood disorders. In Saudi Arabia, the prevalence of this disease is significantly varied in different regions of the country, and the highest prevalence in the Eastern province of the country. A consanguineous marriage has been linked to the high incidence and prevalence of Sickle Cell Anemia (SCA), which, accounts more than 50%, with the rate of marriage between first cousins ranging from 40% to 50%. However, the last few years showed no increase in the prevalence of sickle cell disease among Saudi's. This might be related to the remarkable scientific progress in the understanding of the complex pathophysiology of the disease, improving knowledge regarding SCA among community, better medical care, and the efforts of Saudi's government to provide genetic counselling services and implementing of mandatory premarital screening program. This review therefore is about the epidemiology, history of SCA among Saudi's, clinical complications, and consanguinity marriage and SCA, with a focus on its local premarital screening program.

Keywords: Sickle cell disease; Sickle cell anemia; Consanguinity marriage; Saudi Arabia

Introduction

Sickle cell disease (SCD) is one of the most common severe autosomal recessively inherited blood disorder, caused by a variant of the β-globin gene called sickle hemoglobin (Hb S). Expression of this disease requires either two copies of Hb S (Hb SS) leading to sickle cell anemia (SCA) or one copy of Hb S in combination with another β-globin variant (such as Hb C) leading to sickle cell disease [1]. This causes the Red Blood Cells (RBCs) to change from the normal biconcave disc shape to an irregular sickled shape [2]. In addition to change of cell's shape, sickled cells have a propensity to adhere to the walls of blood vessels. Thus, sickled RBCs can clog blood vessels, preventing normal blood flow and decreasing delivery of oxygen to organs and tissues. Individuals with Hb SS (sickle cell anemia) are most severely affected [2]. On the other hand, when individuals have one copy of the sickle variant and one copy of the normal β-globin gene (Hb AS), they considered carrier. The carrier state for sickle cell disease is often referred to as "sickle cell trait" and those individuals do not express the disease, however, this might be a risk factor for sudden death during physical training [2].

Epidemiology

Sickle cell disease SCD is the most predominant form of haemoglobinopathy worldwide [3]. The SCD is most common among people from Africa, India, the Caribbean, the Middle East, and the Mediterranean. However, statistical data about the prevalence of SCD in the Arab world is patchy [4]. The first documentation of abnormal HbS (HbS) in the Middle Eastern countries came from Egypt [5,6]. More than 200 000 infants are born with SCD in Africa every year, while in the United States, about 72 000 people affected with SCD [7]. Sickle cell disease is also associated with significant mortality. The highest mortality rate was observed among children between 1 and 3 years of age and adolescents younger than 20 year [8]. However, recently, the mortality rate has been decreased dramatically, primarily because of early diagnosis (via newborn screening), better medical care, and education of family members [2].

Brief History in Saudi Arabia

In 1963, sickle cell gene was first recognized in Saudi Arabia in the eastern province of the country by Lehmann and co-workers [9]. In Saudi Arabia the prevalence of sickle cell disease varies significantly in different parts of the country, the maximum prevalence was noted in the Eastern province, followed by the southwestern provinces [7]. In 2007, Al-Hamdan and colleagues reported the highest rates of sickling at Al-Ahsa region of the country (sickle cell trait 16.89% and sickle cell disease 1.20%), followed by Qunfudah and Jazan [4]. Other regions showed much lower prevalence rates [4]. Two major phenotypes for SCD are exists in Saudi Arabia with: a mild phenotype and a severe phenotype [7].

Complications in Sickle Cell Disease/ Anemia

SCA and SCD are responsible for a number of health problems. In most of the Middle Eastern Arab countries sickle cell anemia found to be associated with financial, social, and psychological cost for maintenance of patients with this disorder [4]. Sickle cell patients may be shorter or smaller than normal at their childhood. Puberty is often delayed but considerable growth takes place in late adolescence adults with sickle cell anemia are at least as tall as normal [10]. Adult patients with severe anemia, who suffered vertebral infarction and collapse may be shorter than normal. Another Physical effect of SCD is that the abnormal faces results from extension of the marrow into the cortical bone causing widening of the diploe spaces and thinning of the bone cortex [3]. Bone Pain Crisis (BPC) is another common complication associated with SCD that usually affect the long bones such as femur and humerus, vertebrae, pelvis, ribs and sternum [11]. High risk of infections were linked to impaired splenic synthesis of immunoglobulins as a result of loss of splenic function with about 30% occurs by first year of life and 90% by sixth year of life [12]. Hemolysis can be also generated from sickled cells, causing chronic anemia [13]. Symptoms usually appear within the first six months of life, but there is considerable variability in the severity of the disorder [3].

Sickle Cell Anemia and Consanguinity Marriage

Most of the previous researches demonstrated the association of SCA with parental consanguinity concluded that the populations with high consanguinity marriages rates have a significantly higher incidence of inherited blood disorders such as sickle cell anemia [14-17]. It has been shown that the first cousin marriages in Saudi Arabia and other Arab countries are significantly higher than in the South and North Americans, Europeans, South Africans, eastern Asians, and the populations in the Oceanic countries. In Saudi Arabia, the high rate for consanguinity marriages, which, accounts more than 50%, with the rate of marriage between first cousins ranging from 40% to 50%, are related to traditional and social factors to keep property within families [18-21]. The main harmful genetic effects of the cousin marriages are a higher frequency and incidence of autosomal recessive disorders and higher morbidity and mortality rates among the offspring [18,22].

Sickle Cell Disease/Anemia Control Program

For a successful control programme community screening, carrier detection and genetic counseling to prevent further transmission of the trait are considered important steps for controlling SCA. Carrier detection should be offered through neonatal and prenatal investigation programs for hemoglobin abnormalities, which, have been recognized cost-effective in populations with a high prevalence. Beside the need for care and rehabilitation for the affected patients, effective strategies for control and prevention were considered as an essential measure toward decreasing the birth of affected children (primary prevention). Additionally, for reducing mortality and mortality associated with SCDs, an early detection and diagnosis of sickle cell disease is crucial [3].

In addition, education and increasing the awareness about SCA among general public and the health care providers are essential approaches. These can be achieved through; inclusion of relevant information about the disease in the school curricula, arranging for workshops, symposia and special days for SCA as well as publishing in newspapers and talk shows on the radio and TV may also help.

Sickle Cell Disease Control and Current Challenges in Saudi Arabia

Saudi Arabia was one of the several countries, which adopted effective steps directed toward prevention. In 2003, the government of Saudi Arabia decided to implement a premarital screening program to decrease the incidence of the common hemoglobinopathies in Saudi Arabia, including sickle cell disease. In the next year, the screening test was made mandatory for all couples planning to marry and applying for a marriage license [4]. The programme was complemented by genetic counselling services for the carriers and the diseased and offered by trained counsellors and wedding authorities. These services were effective in improving awareness and provide equitable access to health services, improve quality of life of those affected and help achieve primary, secondary and tertiary prevention. A network of more than 1800 primary health care centers, 200 hospitals and a number of governmental and private organizations were involved in the screening and providing of health care services to the whole Saudi population [4]. At these health care centers, trained laboratory staff recorded the basic demographic information for each couples and collected the blood samples in EDTA anticoagulant tube [4]. At the laboratory, a number of hematological investigations were performed on the sample including; complete blood count (hemoglobin, red cell count, hematocrit, mean corpuscular volume, red cell distribution width), peripheral blood film for red blood cell morphology, reticulocyte count and sickling test (addition of 2 mL of sodium metabisulphite to 20 mL of blood; considered positive if the solution becomes turbid, whereas a clear solution is considered negative). Main hemoglobinopathy diagnosis was depended on hemoglobin electrophoresis, and results were interpreted in accordance with standard laboratory diagnostic protocols [23].

Marriage certificate cann't be given to couple with high risk who, had positive sickle cell trait/disease and were referred to a regional genetic counseling clinic. However, couples had the right to marry regardless of the screening test results. Followed up of all the high-risk couples were provided with genetic counseling physicians working in the different clinics [4].

Unfortunately, during the few first years after program implementation at Saudi Arabia the main goal for decreasing the incidence of this disorder was not successful. Since, almost 90% of the high-risk couples married each other despite being aware of the risk to have children with inherited haemoglobin disorders. Cultural pressure was reported as the main reason to proceed with marriage in the majority of cases [24].

However, in 2006, downward trend in the marriage of high-risk couples was observed. Between 2004 and 2009 it has been found that there was no increase in the prevalence of sickle cell disease (among 1000 examined persons) [17]. More recent study performed at Marriage Center of Northern Border Region in Arar showed that more than 60% of the participants at-high risk marriages were cancelled their marriage proposals, which result in reducing the incidence of SCA and other genetic disease across the area [25].

Conclusion

Sickle cell trait/disease, is one of the major autosomal recessively inherited blood disorder in Saudi Arabia. This disorder is associated with consanguinity and significantly related to high rate of prevalence and mortality among Saudi's. The implementation of mandatory premarital screening program for all couples planning to marry and the Saudi government efforts to provide genetic counselling services at different health care centers and a number of governmental and private hospitals have a significant impact in decreasing the incidence of this diseases. This indicates that the premarital screening program to detect hemoglobinopathy at Saudi Arabia was successful in diagnosing the vast majority of eligible individuals, identifying the couples at high risk of having children with sickle cell trait/disease, and providing them with health education.

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