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ARTICLE **SHORT COMMUNICATION**

Premarital Blood Tests to Decrease the Burden of Consanguinity-Related Diseases in the Arab World: A Call for Action

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Consanguinity or inbreeding is defined as a married couple with at least one common ancestor. Autosomal recessive gene mutations are inherited from these ancestors and expressed, increasing their offspring's risk of genetic disorders. Children of consanguineous parents could develop a variety of diseases including protein-C or protein-S deficiency, beta-thalassemia, phenylketonuria (PKU), hypertension in children, low birth weight, Down syndrome, and immunodeficiency disorders. The incidence of multifactorial disorders such as cardiovascular diseases, diabetes, certain types of cancer, and obesity is also increased in the offspring of consanguineous parents.[1] Moreover, percentages of recurrent abortions, child deaths, and stillbirths were significantly less among non-consanguineous parents when compared to consanguineous ones (67%, 80%, and 80.6% respectively).[1]

Consanguinity is currently practiced by more than a billion people around the world, with significant prevalence in the Arab world stretching from the Gulf States and Iraq in the east to Mauritania on the Atlantic coast of North Africa and Morocco in the west. Rates have reached around 20%-50% in North Africa and the Middle East; 1%–5% in Japan, South America, and Southern Europe; and less than 1% in Oceania, Western Europe, and North America. Various underlying factors promote this type of marriage, including: (1) lower cost and easier premarital and marital arrangements and financial benefits of dowry (2) social and cultural beliefs (3) lower educational levels (4) marriage of women at younger ages (5) residing in rural areas (stronger family and tribal bonds in the tight-knit communities) [2], Figure 1.

The likelihood of inheriting identical copies of detrimental recessive genes increases as the blood relationship between parents becomes closer. About 86% of consanguineous unions involve first cousins who are expected to share approximately 1/8 of their genes. Consequently, their offspring will be homozygous at approximately 1/16 of their gene loci, meaning they will inherit identical gene copies from each parent at these specific parts of their genome. [1, 2]. In Jordan, among families affected by autosomal recessive conditions, marriages between first cousins accounted for 69% of the total. In the case of dominant, X-linked, and chromosomal conditions, first-cousin marriages constituted 22% of the total marriages. Additionally, among families with sporadic undiagnosed conditions, 41.7% of marriages were between first cousins.[3]

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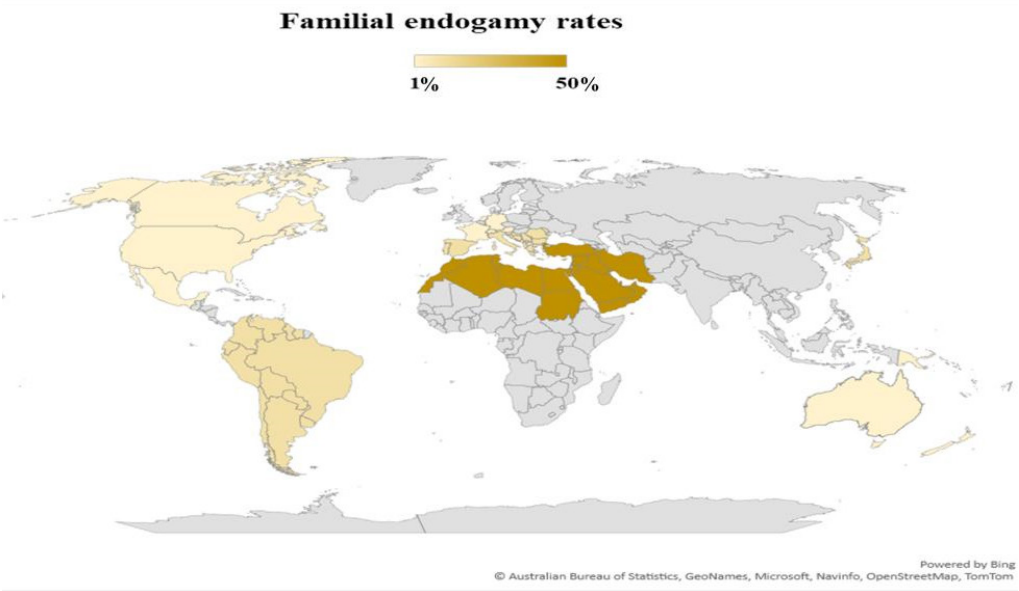
Figure 1. Demonstrates familial endogamy rates throughout the world



In Saudi Arabia, an increased risk of congenital heart disease was observed in children of consanguineous parents. 40% of children in Saudi Arabia who had heart defects were the outcome of first-cousin marriages. In addition, incidences of epilepsy, cleft lip or palate, spina bifida, congenital deafness, and hydrocephalus were all higher in consanguineous couples. Some other non-organic consequences were hyperactivity, intellectual disability, and reading and learning disorders. These are all multifactorial in nature, but consanguinity might be a contributing factor [4].

Premarital Screening (PMS) is a recommended screening program designed to point out if any of the individuals planning to get married are genetic disease carriers. In Qatar, a mandatory test program for sickle cell disease, thalassemia, cystic fibrosis, sexually transmitted diseases (STDs), and classic homocystinuria was initiated. The increasing consanguinity rate in Qatar (around 54%) was the drive behind mandating this program since they have the highest frequency of classic homocystinuria (~1:1800) worldwide in addition to an increased rate of hemoglobinopathies (30.4%). Evidence has shown that PMS was effective in reducing the number of marriages of high-risk couples along with reducing the incidence of genetic diseases like β-thalassemia. This supports the idea that there should be more health education programs increasing awareness about PMS in countries with high rates of hereditary diseases and consanguineous marriages to reduce the burden of genetic diseases among future couples. According to recent studies, the percentage of people who had satisfactory knowledge about PMS in Qatar and Saudi Arabia was 56% and 9.2% respectively. In addition, half of the participants in a study in Oman were unaware of premarital testing. [5]

Newborn Screening (NS) programs include multiple tests that can detect various debilitating and lethal disorders including cystic fibrosis, hemoglobinopathies, congenital adrenal hyperplasia, inborn errors of metabolism, congenital hypothyroidism, severe combined immunodeficiency, and genetic syndromes. NS programs are one of the solutions that emerged to improve the outcomes of consanguineous marriages. The heel prick test is usually done during the first couple of hours or within a few days after birth to screen for any diseases. It would be very beneficial to intensify efforts in educating mothers about neonatal screening programs, their significance, and the optimal timing for conducting these tests. Such educational initiatives should be carried out by healthcare professionals to ensure the effective dissemination of knowledge. [6]

Other options like prenatal diagnosis and preimplantation genetic testing used to avoid the risk of having children with certain disorders are more widely available now. Therefore, there is technically no legal obligation to ban high-risk marriages as doing so will focus the spotlight on an important ethical side of this argument which is the liberty to choose your partner. Another

important limitation of PMS that should be overcome, especially in Islamic countries, is the idea that PMS interferes with God's will and what we are destined to live with. That's why 87.6% of the participants in a study conducted in Saudi Arabia believed that a crucial part of PMS campaigns should be involving religious scholars. In conclusion, encouraging PMS is achievable with some efforts to resolve the aforementioned issues [5].

AUTHOR CONTRIBUTIONS

HU Writing the original draft

AM Conceptualization and writing of the original draft

HH Visualization

OM Update the draft and revision

KA Final editing and revision

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