

# Consanguinity: Understanding the Health Risks and Guidance for Providers

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## Background

Consanguineous marriage, defined as a union between family members who are second cousins or closer, has a long and varied history throughout all major cultures of the world. It is currently estimated that approximately 10.4% of the global population are the offspring of consanguineous parents.<sup>1</sup> While the practice has been observed around the world for centuries, and up until the late 19<sup>th</sup> century was still commonly practiced in Western Europe and North America, in this current day and age the practice remains most firmly entrenched in communities across North Africa, the Middle East, and Western Asia. Most commonly, consanguineous marriages take place between first cousins, and can represent up to 50% of marriages within some communities.<sup>1,2</sup>

The practice of marrying within an extended family, for many communities, has deep cultural and even practical values. Consanguineous unions, particularly if the family is part of a small community or ethnic minority group, may ensure survival of a family lineage and cultural and religious beliefs and values, and reinforce the strong family structure.<sup>3-5</sup> Additionally, marriage within the family may ensure that wealth, such as land, is passed down to the next generation, and help to reduce the financial costs associated with marriage, such as the bride's dowry. Currently, the practice of consanguineous marriage is seen most frequently in rural communities associated with traditional lifestyles, though it may still be practiced in more urban hubs.<sup>6</sup> Contrary to what may be popular belief in the West, though widely practiced in several Muslim-majority countries, the practice is not directly associated with Islam and is observed within other religious groups within the regions.<sup>2,6</sup>

In refugee and other immigrant communities, these traditional marriage practices may shift in the new country of residence. In a survey of Syrian refugees in Jordan, a lower rate of consanguineous marriages was noted and attributed to the disruption of extended families when some leave to go abroad.<sup>7</sup> However, in other communities, the practice may remain prevalent or even increase, as was noted to be the case in a large longitudinal survey of the Pakistani community in Great Britain and the Turkish and Moroccan communities in Belgium.<sup>8,9</sup> The persistence of consanguineous marriage in such instances is attributed in part to the desire to keep marriage within the community and the smaller number of non-family options for marriage. Additionally, there could be an incentive to choose an extended family member from their home country as a potential husband or wife if family members of immigrants receive preference in obtaining a visa if planning to join family that is already abroad.<sup>2</sup>

## Health Implications

Consanguineous unions have long been known to be associated with increased health risks in offspring. In a marriage between first cousins, it is estimated that they would have 1/8<sup>th</sup> of their genes identical, so children born of the relationship would be homozygous at 1/16<sup>th</sup> of all loci; this may be increased in communities where the same families have been intermarrying for generations, or reduced in instances where marriage is taking place between more distant relations, such as second or third cousins.<sup>1,10</sup> The risk of congenital disease, particularly autosomal recessive conditions, has been estimated to be 1.7-2.8% higher in the offspring of consanguineous couples compared to the background population.<sup>1</sup> In the case of autosomal recessive conditions, if each parent is a carrier for the

disorder, each child has a 25% chance of inheriting the disease. The relationship between consanguinity and incidence of genetic disease has been well-characterized across populations where consanguineous marriage frequently occurs. Primarily, the focus has remained on the association of consanguinity with increased rates of rare autosomal recessive conditions, including endocrine and metabolic disorders, hemoglobinopathies such as sickle cell disease and thalassemia, and deafness.<sup>11,12</sup>

While recent literature has primarily examined the association between consanguinity and autosomal recessive disease, the potential health effects of consanguineous unions are not strictly limited to such conditions but also congenital malformations, such as structural heart defects. Studies of children in Saudi Arabia and Lebanon who were the offspring of consanguineous relationships found an increased incidence of atrial and ventral septal defects, emphasizing the potential role of inheritance in congenital heart disease and the connection between consanguineous relationships and this disease.<sup>13,14</sup>

Studies to date have not shown a strong association between consanguineous unions and miscarriage, though this may be undercounted as most spontaneous abortions secondary to severe genetic defects occur very early during pregnancy, possibly before the mother may be aware that she is pregnant.<sup>2,15</sup> However, data have indicated that there is an association between consanguinity and increased infant mortality. In Turkey, where it is estimated that approximately a quarter of the population is in a consanguineous relationship, an analysis attempted to understand why Turkey's infant mortality rate has remained higher than similarly developed nations and found a significant positive correlation between first-degree consanguineous marriage and infant mortality even when controlling for additional variables such as maternal age and education level, both of which tend to be lower in consanguineous marriages and are inversely correlated with infant mortality rates.<sup>16</sup>

### **Guidance for Healthcare Providers**

Primary care providers may be the first point of contact with the healthcare system for new patients who have just arrived in the United States. When working with new families, it is important to identify consanguineous couples that may be at increased risk of passing a genetic condition on to their offspring.<sup>17</sup> If the parents are known to be consanguineous, the provider should elicit further history to draw out whether or not there already exists a history of congenital disorders within an extended family that may signify that the parents are carriers of a genetic disease. Key questions to consider would be if there is a history of birth defects, intellectual disability, developmental delay, unexplained infant death, blood disorders, or early vision or hearing impairment, all of which may signify underlying genetic disease and may require further investigation.<sup>18</sup> In refugee populations, due to the nature of their exit from their home country, patients also may have had limited access to health services, particularly newborn screenings that may detect inherited conditions in infants. Children may present with undiagnosed conditions that necessitate further evaluation and treatment, and the onus lies with the provider to take a detailed history in order to elicit history and complaints that may signify an underlying congenital abnormality.

Within different immigrant communities, there may be differing levels of awareness of the association between consanguineous marriage and disease as well as different attitudes towards the practice that are important for providers to consider. For example, in one survey in the Netherlands of Turkish and Moroccan refugee communities, researchers found that respondents from Morocco viewed consanguinity more favorably than those from Turkey. This was attributed in part to Turkey's campaign to reduce their current rate of infant mortality by educating communities on the health risks of consanguineous relationships

and creating programs targeted at offering genetic counselling and premarital screening, therefore resulting in increased awareness within the Turkish population of some of the risks of consanguineous relationships.<sup>3</sup> In contrast, in a survey of women in consanguineous relationships in a community in Iraq, there was a high preference towards intermarriage however little knowledge of the potential associated health consequences.<sup>19</sup> In other communities still, there appears to be a high awareness of the health implications of consanguinity but the practice continues to be viewed favorably as an important component of preserving cultural values and resources within a community and family. A study of adults in Oman found that there was both a high rate of consanguineous marriage and a positive attitude towards the practice despite two-thirds of respondents reporting awareness of the potential health implications.<sup>20</sup> Even in immigrant communities in the West, individuals may have been made aware that there are potential health implications of consanguineous marriage but not understand the genetic basis of disease and how this may be passed within a family.<sup>21</sup> Such studies indicate how awareness of health consequences does not necessarily shift long-ingrained cultural attitudes towards consanguineous relationships and illustrates how providers, when working with these patients, must take a multi-faceted approach when it comes to providing care. Guidance for these patients should be tailored to the patient's understanding while also taking care not to stigmatize consanguineous relationships or discourage pregnancies based on presumed risk.

Preconception and/or prenatal counseling remains the cornerstone of identifying couples that may be at risk of passing on an inherited disease to their offspring. The primary care provider may be turned to as a source of medical guidance for these patients when considering future pregnancies. Surveys of Syrian refugees in consanguineous relationships indicate that even in situations

where couples may be aware of the existence of preconception and prenatal screening to help navigate the process of assessing risk, there remains a deficit of knowledge when it comes to determining next steps as to how to access such services.<sup>22</sup> Offering prenatal genetic screening and counseling gives patients in consanguineous relationships the opportunity to make the choice themselves to identify if they carry genetic traits that may put offspring at risk for inheriting a recessive condition. With the rise of more affordable pan-genetic testing through expanded carrier screening (ECS) providers are able to offer genetic screening that tests for a wide range of autosomal and X-linked conditions to more patients planning to become pregnant.<sup>18,23</sup>

Just as providers must remember that different groups of patients engaged in consanguineous unions may differ in their knowledge and perception of the health implications associated with consanguineous marriage, it is also important to acknowledge that patients may differ in their decision whether or not to seek genetic screening in the preconception or prenatal stage. In one qualitative study of a number of patients who were receiving genetic counseling for consanguinity, participants reported that they did feel that increased knowledge about potential reproductive risks provided reassurance and helped them to make informed decisions regarding future pregnancies. However, barriers such as cost, fear of stigmatization, both by medical providers or other family members, or preconceptions that a provider will recommend termination of pregnancy or against pregnancy might dissuade others from pursuing counseling.<sup>23-25</sup> For some patients, there may be misconceptions as to the level of potential risk to future children, particularly if they have seen other extended family members intermarry and produce healthy children. Still other patients may feel that even if they were aware of a risk to a future baby it would not change the outcome and so may not wish to pursue screening.<sup>23</sup> In these instances, it remains

crucial for providers to offer up the opportunity to educate patients on potential risks and refer to genetic counseling if they desire, but be clear that they do not stigmatize the parents for their relationship and not dissuade the family from future pregnancies, particularly if there is not yet an established risk to any future offspring. An approach to genetic counseling, piloted within the Arab community in Israel, found that following a framework of identifying misconceptions that patients may have around genetic counseling and screening and allowing the patient to discuss their concerns about the process and their own perceptions of the basis of disease, or perceptions of risk to future children were important first steps in the counseling process. By giving patients the opportunity to express their own views initially, providers were better able to tailor their approach to the patient's own knowledge level and concerns, providing a basis for providers and patients to engage in shared decision-making when it came to determining next steps to take in the process.<sup>24</sup>

## Conclusion

Consanguineous relationships continue to be an integral part of life in many communities across the world to this day. While no longer widely practiced in North America, and in fact expressly prohibited in many states, providers that serve refugee and other immigrant communities may continue to see patients in consanguineous relationships<sup>15</sup>. For the children of these marriages, there is an increased risk of inheritance of rare autosomal recessive conditions and other congenital malformations. It is important for primary care providers to be able to understand what may be the potential health implications for the children of their patients and be able to provide proper counseling and refer to the proper subspecialty services, such as a genetic counselor, going forward. Patients may differ widely in their knowledge and understanding about potential health risks and their perceptions of genetic counseling, so it remains key that providers tailor their approach to help when guiding

patients through this delicate process and consistently engage in collaborative decision making with patients and their families for the best outcomes.

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