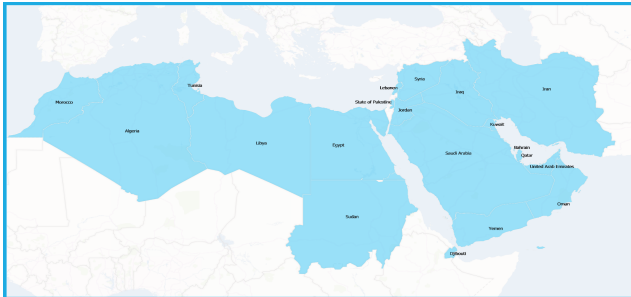


MENA DESCENDANTS & GENETIC ABNORMALITIES

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* ABSTRACT

This study aims to assess the impact of consanguinity in the Middle East and North Africa. The region, also referred to as the MENA region, spans from Morocco to Iran. These identified Arab countries (Figure 1) have an increased risk of genetic disorders that can result in sickle cell anemia and thalassemia ("Middle East & North Africa" 2024). Specifically, this study investigates the increased prevalence of these disorders in the MENA region, as well as the contributing factors behind the practice, such as education level and age at the time of marriage. An in-depth comparative analysis of previous research on this topic was undertaken to draw the correlation between the levels of consanguineous marriages in the MENA region and the levels of genetic variation disorders. The analysis shows that there is a correlation between the two variables while highlighting the practices to counteract such variations in the Middle East. These findings suggest a greater need for adaptive medical practices in the United States to adequately assist the growing number of Arab Americans nationwide.

1 INTRODUCTION

The MENA region is an encompassing term used to describe the Middle East and North Africa, consisting of 26 countries overall. Comparatively, the MENA region is also home to one of the highest rates of consanguinity, defined as the intermarriage of

either first or second cousins sharing the same bloodline. According to a study published in the Iranian Public Health Journal, the prevalence of consanguinity accounted for 20-50% of the overall population in both the Middle East and North Africa (El Goundali et al. 2022). In a similar study published in the European Journal of Public Health, researchers found that "consanguinity is common in North Africa (NA), and the estimates range from 40 to 49% of all marriages in Tunisia and 29-33% in Morocco" (Anwar et al. 2014). Furthermore, the detrimental effects of consanguinity have been proven to cause "Thalassemia and sickle cell disease/anemia, [as they] constitute the most common inherited recessive disorders" (Anwar et al. 2014). In order to understand the identification of these genetic variation disorders, one must further consider the social and economic impact on the affected individuals. The age of women at the time of marriage accounts for the most prominent factor within consanguinity, specifically in Morocco. As per the Iranian Public Health Journal, "almost all of the included studies reported that consanguineous marriage occurs in younger ages compared to non-consanguineous marriage" (El Goundali et al. 2022).

The results obtained in Morocco from 9,969 women ages 15-49 years showed that the highest proportion of consanguineous marriage was observed in the 15-19 age group, which not only highlights the disparities of age, but the level of education at the time of marriage as well. Geographical location is the second most important factor when connecting the elements associated with consanguinity in the MENA region. In that same study, researchers found that "in the latest demographic survey in Morocco of 9,969 women, consanguineous marriage is more common among rural resident women. The probability of entering into consanguineous unions among women is significantly higher for those who spent their childhood in the countryside; they are almost eighteen times more likely than their urban counterparts. [Yet women in] northwestern Morocco, [who reside in] rural [areas] before marriage have a highly significant effect on the choice of consanguineous marriage" (El Goundali et al., 2022). Lastly, while many MENA countries have made significant economic gains in recent years, economic status is the last piece of the consanguinity puzzle, allowing us to completely understand all the components behind consanguineous

marriages throughout Middle Eastern and North African countries.

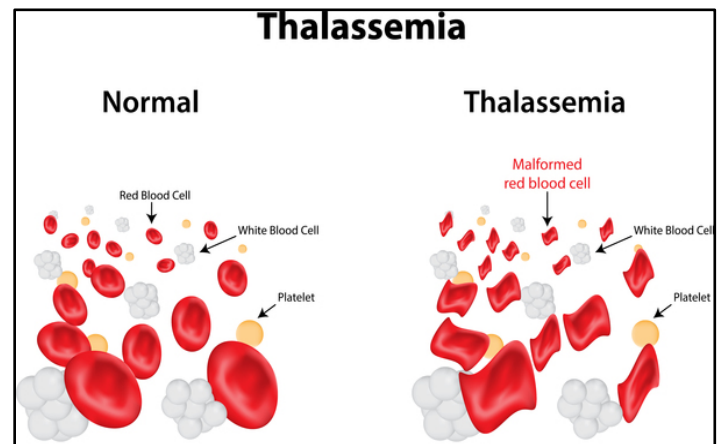
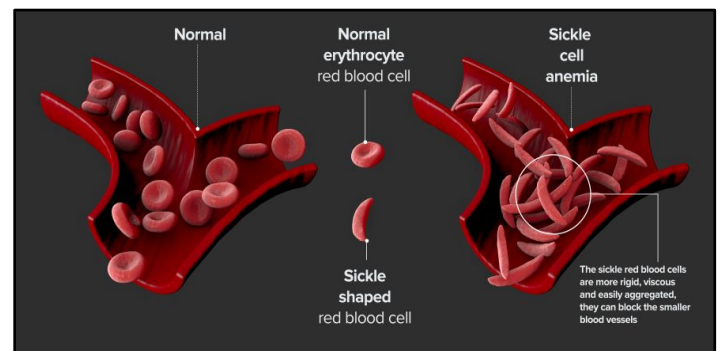
2 PREVENTATIVE MEASURES IN THE GULF REGION OF THE MIDDLE EAST

Due to proactively countering consanguineous marriages and the detrimental effects on unborn children, countries such as Saudi Arabia (KSA), Bahrain, Iraq, Qatar, and The United Arab Emirates have mandated a premarital screening for hemoglobinopathies. The increased prevalence in the region has resulted in screenings for thalassemia and sickle cell anemia. This change has decreased the number of sickle cell anemia cases in just Bahrain alone (Abou Tayoun et al. 2021). However, the medical accessibility and acceptance has caused a stalemate in medical screenings for MENA residents. According to the *Genomic Medicine in the Middle East* study, researchers state that “population genetic screening, in the form of limited or expanded gene panels, is still lacking in the Middle East. Cytogenetic services, mostly based on traditional karyotyping and FISH, have become widely available in major hospitals within the region. The most prominent gap in the region is the lack of adequate clinical genomic-sequencing facilities”, which highlights the growing need for resources based on medical necessities due to high levels of consanguinity (Abou Tayoun et al. 2021).

3 DISORDERS OF GENETIC VARIATION

To further understand the severity of these genetic mutations, one must understand the cause and symptoms associated with genetically varied disorders like with sickle cell anemia. According to the Mayo Clinic, those who inherit this disease have red blood cells shaped like crescent moons or sickles. This particular shape causes these cells to be rigid and sticky, slowing or blocking blood flow. On the other hand, regular red blood cells that are typically round and flexible while moving throughout the bloodstream. The onset of sickle cell anemia symptoms can begin around the age of six months, with many patients experiencing episodes of pain,

swelling of the hands and feet, anemia, habitual infections, vision problems, delayed growth or even puberty (Mayo Clinic 2022). Common complications associated with this disease include, but are not limited to, organ damage, pulmonary hypertension, leg ulcers, blindness, and pregnancy complications. As recommended by medical professionals, the easiest way to prevent this genetic abnormality and reduce the risk of passing such mutations is through genetic testing. Figure 2 below shows the breakdown of sickle cell anemia and the difference in blood cells passing through the bloodstream between regular (left) and crescent-shaped cells (right) (“Sickle Cell Disease” 2021).

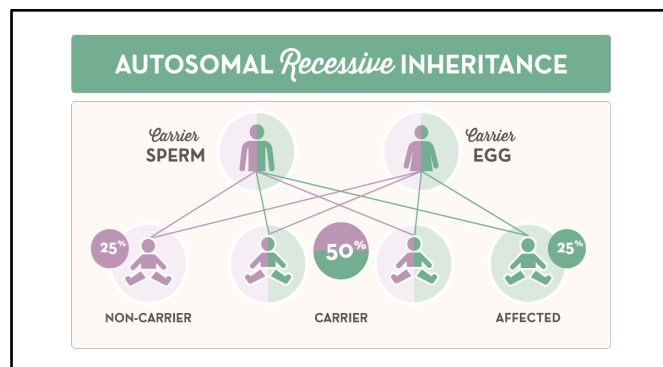


Moreover, one of the more prevalent genetic abnormalities when examining descendants from the MENA region is β -thalassemia. This specific demographic is genetically predisposed to an increased probability of inheriting this blood disorder. This issue arises once the body does not make enough beta globin as it should. Beta globin and alpha globin are building blocks of hemoglobin, which is a part of the red blood cell (RBC). The red blood cell is a carrier of

oxygen across the body and decreases in beta-globin can cause anemia (Schultz 2022). One of the main problems with this blood disorder is primarily due to the lack of hemoglobin. According to the American Family Physician Journal, beta globin chain production can range from near normal to completely absent. This leads to varying degrees of excess alpha globin to beta globin chain production, where the beta thalassemia trait (minor) is asymptomatic and results in microcytosis and mild anemia. While many do not experience iron deficiency, the typical path to treatment is contingent on low iron levels, resulting in blood transfusions, chelation, and even bone marrow transplants for severe cases (Muncie & Campbell 2009). Given by the previous research conducted by the Mayo Clinic and the National Library of Medicine, it is critical to understand how consanguinity affects the DNA's makeup. Through genetic testing and medical advancements, these congenital abnormalities become more transparent and manageable for medical professionals and researchers to prevent such mutations from arising. Figure 3 above shows a side-by-side comparison of normal red blood cells to that of malformed red blood cells in a person with beta thalassemia (MedlinePlus 2020).

4 MEDICAL TESTING IN THE UNITED STATES

To effectively combat this issue head-on, implementing a program which focuses on the current issue in Saudi Arabia and Bahrain will create an equal opportunity for Arab Americans to receive access to healthcare. In addition, this program will also allow medical practitioners to promote the educational aspect to Arab Americans in order to stay informed about the risks. Through the use of testing in CLIA-certified testing laboratories and in conjunction with prenatal screening and gene carrier testing, the United States healthcare system will better accommodate the relevant healthcare solutions most suitable to the needs of the growing population of Arab Americans.



CARRIER TESTING

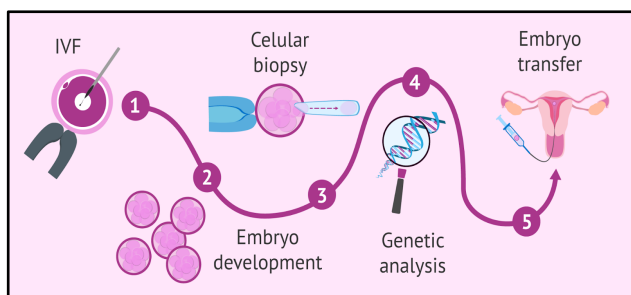
To counteract, the specific disorders of genetic variations disproportionately affect the Arab Americans. Carrier testing can be utilized if one parent has a "family history of congenital abnormalities or mutations that are passed on in an autosomal recessive way," according to the Cleveland Clinic. According to ORM Genomics, 30-50% of all patients screened at ORM using carrier testing are found to be carriers of at least one genetic condition ("Genetic Carrier Screening"). However, carrier testing can range anywhere from \$100-2,000, depending on the types of testing being performed and may be covered by insurance if recommended by a specialist (Mannarino & Ghazal 2023). Figure 4 above shows how carrier testing is conducted, what is evaluated, and when it is used for predictive prenatal testing ("Genetic Carrier Screening").

PREIMPLANTATION TESTING

Another way to mitigate the growing number of genetic abnormalities present in MENA descendants is preimplantation screening. This medical technique is one of many ways used to detect and determine the possibility of passing disorders with genetic variation to future generations. Figure 5 below shows the process of preimplantation screening to evaluate genetic risks from beginning to end ("The PGD Process Step by Step" 2020). This test is conducted by specialists who "search for genetic mutations in the [female's] embryos that were made using assisted reproductive techniques (ART), [such as] in-vitro fertilization (IVF), [where] a small number of cells are taken from your embryos and tested for [specific] mutations. [Once completed], only [the] embryos without these mutations are implanted in your uterus [attempting] to start a pregnancy" (Cleveland

Clinic 2022). Those who are “over age 37, couples who are at risk of having a child affected with an inherited genetic disease [and those who have a] history of recurrent miscarriages due to chromosomal abnormalities” qualify for this type of genetic screening (PFCLA 2022). However as per the Pacific Fertility Center in Los Angeles, this procedure can vary anywhere from \$4,000-10,000 for PGT-A/PGS and is not covered by most private or public health insurance within the United States (Drazba et al. 2014).

5 CONCLUSION



This paper highlights the stark differences between preventative and curative practices currently available in the United States compared to those of Middle Eastern and North African countries. Considering that there are over 3.7 million Arab Americans (Arab American Institute 2023) currently living in the U.S., this research identifies the flaw in American healthcare for MENA descendants. Receiving proper medical resources can determine if genetic variations are prevalent and can also offer preventative care to MENA descendants. Medicare, which currently provides medical coverage to roughly 65 million Americans, offers little to no coverage for genetic testing or inherited genetic disorders (“Medicare & Medicaid Coverage of Genetic Services”). According to the U.S. Department of Health & Human Services for the Office of the Inspector General, genetic testing under Medicare Part B will not be covered for predictive purposes as it is considered a screening exam (Washmuth 2023).

6 REFERENCES

- [1] Abou Tayoun, A. N., Fakhro, K. A., Alsheikh-Ali, A., & Alkuraya, F. S. (2021). Genomic medicine in the Middle East. *Genome Medicine*, 13(1). <https://doi.org/10.1186/s13073-021-01003-9>
- [2] Anwar, W. A., Khyatti, M., & Hemminki, K. (2014). Consanguinity and genetic diseases in North Africa and immigrants to Europe. *The European Journal of Public Health*, 24(suppl 1), 57–63. <https://doi.org/10.1093/eurpub/cku104>
- [3] Arab American Institute. (2019). Demographics - Arab American Institute. Arab American Institute. <https://www.aaiusa.org/demographics>
- [4] Cleveland Clinic. (2022, May 21). DNA Test - Genetic Testing Overview. Cleveland Clinic. <https://my.clevelandclinic.org/health/diagnostics/23065-dna-test--genetic-testing>
- [5] Cleveland Clinic. (2022, May 21). DNA Test - Genetic Testing Overview. Cleveland Clinic. <https://my.clevelandclinic.org/health/diagnostics/23065-dna-test--genetic-testing>
- [6] Drazba, K. T., Kelley, M. A., & Hershberger, P. E. (2014). A Qualitative Inquiry of the Financial Concerns of Couples Opting to Use Preimplantation Genetic Diagnosis to Prevent the Transmission of Known Genetic Disorders. *Journal of Genetic Counseling*, 23(2), 202–211. <https://doi.org/10.1007/s10897-013-9638-7>
- [7] El Goundali, K., Chebabe, M., Zahra Laamiri, F., & Hlali, A. (2022). The Determinants of Consanguineous Marriages among the Arab Population: A Systematic Review. *Iranian Journal of Public Health*. <https://doi.org/10.18502/ijph.v51i2.8679>
- [8] Genetic Carrier Screening. (n.d.). ORM Genomics; ORM Genomics. Retrieved April 16, 2024, from <https://ormgenomics.com/genetic-carrier-screening/>
- [9] Herbert L. Muncie, J., & Campbell, J. S. (2009). Alpha and Beta Thalassemia. *American Family Physician*, 80(4), 339–344. <https://www.aafp.org/pubs/afp/issues/2009/0815/p339.html#:~:text=Persons%20with%20thalassemia%20trait%20have>
- [10] Mayo Clinic. (2022, March 9). Sickle cell anemia - symptoms and causes. Mayo Clinic; Mayo Clinic Staff.
- [11] Medicare & Medicaid Coverage of Genetic Services. (n.d.). Medicare & Medicaid Coverage of Genetic

Services. Retrieved February 19, 2024, from <https://www.facingourrisk.org/support/insurance-paying-for-care/genetic-services/medicare-medicaid>

- [12] Middle East & North Africa. (2024). Unicef.org. <https://www.unicef.org/mena/sites/unicef.org.mena/files/MENA-map-update.gif>
- [13] PFCLA. (2022, August 9). PGS/PGT-A testing costs and candidates. Wwww.pfcla.com. <https://www.pfcla.com/blog/pgs-pgta-testing-costs#:~:text=Depending%20on%20the%20fertility%20clinic>
- [14] Schultz, C. (2022, March). Beta Thalassemia (for Parents) - Nemours KidsHealth. Kidshealth.org. <https://kidshealth.org/en/parents/beta-thalassemia.html#:~:text=Beta%20thalassemia%20is%20an%20inherited>
- [15] Sickle Cell Disease. (2021). Sc.gov; South Carolina Department of Public Health. https://dph.sc.gov/sites/scdph/files/2024-06/Sickle_Cell_Example.jpg
- [16] The PGD Process Step by Step. (2020, December 30). InviTRA. <https://www.invitra.com/en/about-the-pgd-process/>
- [17] Washmuth, D. (2023, November 12). Non-Physician Practitioner | Importance, Types & Examples. Study.com. <https://study.com/academy/lesson/non-physician-practitioners-in-healthcare-systems.html>



Ms. Guennouni is a rising senior in the School of Arts and Sciences at Rutgers University New Brunswick, where she is pursuing her B.A. in Political Science with minors in Spanish, Business & Technical Writing. The inspiration for this research paper came from the personal experiences of being an Arab American, where she has seen such practices take place. Yet, the development of her research began in her Writing in the Business and Professions class with Professor Rasheeda Young, where she was tasked with formulating a research topic for a business proposal, accompanied by a presentation and examination of preliminary findings in detail. Throughout her extensive research and guidance under Dr. Amrik Sahota from the Department of Genetics, she gained a more profound understanding of the cultural and historical factors that come into play when looking at the formation of different disorders of genetic variation. This included a high percentage of consanguineous marriages that date back several centuries, through which practices have served as a common practice. Since the paper's original formulation, she has presented her findings as a panelist at the 13th Annual Undergraduate Research Writing Conference (URWC) in April 2025. She was published in the Spring 2024 issue of the Rutgers University Undergraduate Law Review for her paper on "MENA-Related Genetic Abnormalities & Insurance Fraud".

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