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CORRESPONDENCE

Accredited genetic testing in the Arab Gulf region: reinventing the wheel

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The revolution of genetic testing technologies has helped in the advancement of diagnostic tools for genetic diseases, and proved very useful in preventing, managing or treating these diseases. It has been also proven useful in a timely intervention for treating the disease, when it is discovered at early stages of infancy through newborn screening. In addition to providing accurate estimates of the prevalence of genetic diseases, the establishment of molecular genetic testing for a disease has the potential to determine the molecular pathology of the genetic disease in order to improve its prognosis, and guide the development of customized molecular diagnostic approaches against the genetic diseases.

Consanguineous marriage is extremely common in the Gulf countries, ranging from 12 to 70%,[1] with high tendency towards first-cousin marriage (~35%),[2] which is responsible for the high frequency of genetic diseases;[3] these diseases include, but are not limited to, hemoglobinopathies, inborn errors of metabolism, cardiovascular diseases, neurodegenerative diseases, autistic spectrum disorders, gastro-intestinal disorders, and cancer. Genetic studies in consanguineous families will ultimately lead to the identification of novel inherited genetic aberrations related to rare recessive genetic diseases, and might lead to the appearance of new recessive disorders; this makes the Gulf region as a hot spot for medical and clinical genetic studies.[4]

Nonetheless, certain Gulf Cooperation Council (GCC) countries (Figure 1), including Saudi Arabia, Oman, Qatar and UAE, have realized that it is necessary to reinvent the wheel in molecular diagnostics. In these countries, the majority of molecular diagnostic samples are being sent to the West for testing and analysis, and the results from a significant amount of these samples come back negative or inconclusive. The question that these few countries have been debating was, why should significant investments be wasted while comparative studies have established differences in genetic profiles between the East and the West? Therefore, these countries have recently adopted local strategies to develop and establish their own accredited molecular diagnostic laboratories through research and development.[5]

Figure 1 Map of the Gulf Cooperation Council countries, including Saudi Arabia, Qatar, United Arab Emirates (UAE), Oman and Bahrain. A full color version of this figure is available at the *Journal of Human Genetics* journal online.

There is no doubt that these strategies will take a long time to attain fruition, but will ultimately lead to efficient genetic testing, especially with modern massive-throughput genetic technologies. The preliminary outcome from these efforts is indeed rewarding, because some of the recent studies have identified not only novel mutations, but also different common hotspot mutations than those found in the West.[6] These preliminary results propel an urgent call for a debate among Arab states on how to restructure the genetic testing programs in the Arabian Gulf region, and curtail on the huge spending on genetic testing, which in the majority of the cases turns out negative or inconclusive.

Nonetheless, the central issue here is to immediately develop and establish strategies for well-focused genetic research programs that will ultimately identify the culprit mutations underlying the genetic diseases in these Eastern countries, and further develop molecular diagnostic approaches that are unique for Arab patients. In fact Saudi Arabia started a 5-year project of sequencing 100 000 Saudi genomes from across the kingdom, shortly after Qatar launched the 'Qatar

Genome Project' that will ultimately guide the development and the establishment of future treatment strategies for genetic diseases through 'personalized medicine' approaches in the Gulf region. This is expected to help in novel gene discovery.[7] Interestingly, these findings will build an accurate molecular basis for local molecular genetic diagnostics.[8]

Despite these promising strategies, few challenging factors might slow down these megaprojects from achieving some of their anticipated goals. Religious affiliations might represent a real challenge for the application of genetic testing in the Arab world; this might be due to social stigma and the Islamic values, which might result in some individuals shying away from such testing, especially Arab women and children.

Given the prevalent consanguineous marriages in the Arab world, high prevalence of genetic disorders, the potential spread of genetic diseases among Arabs and the undeveloped infrastructure for accredited genetic services, the Arab world is in a 'not ready' status to cope with the requirements for achieving better health care. Therefore, the Arab countries should collaboratively recruit significant investment towards building the infrastructure needed for developing molecular diagnostic approaches tailored to the genetic profile of the most serious and common genetic diseases in the Arab world. In parallel, an active research program should be established to determine the proper molecular targets that are unique to Arab patients, to customize molecular genetic testing.

In conclusion, a number of recent studies have identified novel and distinctive hotspots for disease-causing mutations that are unique to the Gulf Arabian patients, explaining why the DNA samples tested in Western laboratories generate negative or inconclusive results. Here we send an urgent call to our GCC authorities to immediately adopt novel strategies to reinvent the wheel and establish our own accredited molecular diagnostics locally in order to customize the molecular genetic approaches. Interestingly, development and establishment of specialized clinical molecular geneticists, cytogeneticists and genetic counselors, as well as the needed facility infrastructure, will ultimately lead to long-term economic health-care benefits to the Arab nations.

CONFLICT OF INTEREST

The authors declare no conflict of interest.

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