Weill Cornell Medicine-Qatar

Mapping the Qatari genome heralds new era of precision medicine

Doha – June 29, 2016: Qatar now has its own population-specific genome resource after researchers at Weill Cornell Medicine-Qatar (WCM-Q) mapped the genomes of more than 1,000 Qatari nationals.

This resource gives scientists a powerful reference tool that will facilitate efforts to identify genetic variations that cause serious and distressing conditions such as cystic fibrosis, sickle cell anemia and muscular dystrophy among the local population.

The new resource will also help doctors treating Qatari nationals to more effectively practice 'precision medicine,' which involves analyzing a patient's genome in order to more effectively predict, diagnose and treat disease. Furthermore, better understanding of the subtle variations in Qatari genomes will help researchers discover how certain ancestral genetic traits interact with environmental factors such as poor diet, lack of exercise and smoking to cause disease.

The completion of the project is an important milestone in a new phase of genetic research, which has progressed from mapping the entire human genome - first achieved in 2003 after 13 years of painstaking investigation - to focusing on specific populations to identify correlations between shared heritage and susceptibility to particular diseases. This project is considered the most significant resource of genetic variants in any Arab population to date.

The project is part of a series of research studies investigating the Qatari population led by Dr. Ronald Crystal, Chairman of Genetic Medicine at Weill Cornell Medicine in New York and one of the world's leading genetic researchers.

Dr. Crystal said: "This study is the first step in the development of precision medicine in Qatar. Our genes decide how we respond to our environment and our risk for disease, and the variations in our genes are different for each population. With this initial description of the Qatari genome as a basis, and with future refinements to be made by the Qatari Genome Project, we now have the basis for defining the genetic risk of the Qatari population for disease, and how Qataris will respond to medical therapies."

The project, entitled *The Qatar Genome: A Population-Specific Tool for Precision Medicine in the Middle East,* is the work of a research team comprising scientists from WCM-Q, Weill Cornell Medicine in New York, Sidra Medical and Research Center, the Laboratory of Medicine and Pathology at Hamad Medical Corporation and the Department of Biological Statistics and Computational Biology at Cornell University at Ithaca in the United States.

The research team gathered samples anonymously from a total of more than 1,000 Qatari nationals at Hamad Medical Corporation clinics and then used the advanced computing technology in the lab of the WCM-Q Genomics Core to analyze and map the genomes of each individual.

Earlier projects in the series identified three broad genomic groups within the Qatari population, termed Q1, Q2 and Q3. Q1 are largely Bedouins, while Q2 are a Persian or South Asian mixture, and Q3

are Qataris with sub-Saharan African heritage. In the latest study, higher resolution of ancestry was achieved; for example, individuals of Persian ancestry can now be distinguished from individuals of South Asian ancestry. This genetic diversity is important to recognize and understand as each group is likely to be susceptible to different conditions and react to environmental hazards in different ways.

Until now, researchers have usually attempted to identify disease-causing genetic variations by using powerful computers to compare the genomes of affected individuals with a global genome resource and searching for telltale differences between the two. Unfortunately, comparing the genomes of Qatari nationals with the global genome is problematic because the computer identifies many thousands of 'false positives' - tiny variations that may appear rare relative to the global genome average but are in fact very common among individuals of similar ancestries to the affected patient. Sorting the harmless variations from the harmful ones is a monumental task involving careful checking of vast amounts of complex genetic code.

The new population-specific resource mitigates this problem for Qatari nationals by providing a Qatari-specific genome resource, compiled from more than 1,000 Qataris whose families have enjoyed good health for at least three generations, that is a far more effective basis for comparison than the global resource. This is particularly true of one of the largest global resources, the 1000 Genomes Project, which contains no representation of people from the Arab World. Given the shared heritage of Qatar's population with people in other parts of the Middle East and North Africa (MENA), the new Qatar Genome reference could also benefit patients right across the region.

Qatar-based human genetics expert Dr. Khalid Fakhro, an Investigator at Sidra Medical and Research Center and an Assistant Professor in the Department of Genetic Medicine at WCM-Q, is the lead author of this study. He explained: "One of the reasons genetics is complicated is that in the genomes of every population we look at we find there are millions of mutations, yet most of these do not actually cause disease - they appear to be harmless. So when a person with disease shows up in the clinic, it is more difficult than people imagine to identify the few potentially harmful mutations in a sea of mostly harmless variation.

"One way around this is the type of population-specific genome resource that we have created for the Qatari population. Because many of the same harmless mutations are shared by members of the same population, using a population-specific resource makes it easier to identify abnormal mutations in the genome that do cause disease. Specifically, if we find a mutation shared by patients but it has never been observed in more than 1,000 ethnically-matched controls, we have higher confidence in its possible pathogenicity. "

The group of WCM-Q research projects investigating the Qatar Genome has benefited from support from Qatar Foundation and Qatar National Research Fund (QNRF).

Dr. Crystal added: "This research has proven to be extremely exciting and worthwhile, not only for the new discoveries we have made but because there is great potential for clinical applications that will be of benefit to patients in Qatar and the wider region. We are very grateful for the support provided by Qatar National Research Fund and Qatar Foundation, without which this research could not have been undertaken."

Photos Captions:
Photo1: Dr. Ronald Crystal and Dr. Khalid Fakhro
Photo2: Dr. Khalid Fakhro
Photo3: Dr. Ronald Crystal

About Weill Cornell Medicine - Qatar
Weill Cornell Medicine - Qatar is a partnership between Cornell University and Qatar Foundation. It offers a comprehensive six-year medical program leading to the Cornell University M.D. degree with teaching by Cornell and Weill Cornell faculty and by physicians at Hamad Medical Corporation (HMC) and Aspetar Orthopedic and Sports Medicine Hospital who hold Weill Cornell appointments. Through its biomedical research program, WCM-Q is building a sustainable research community in Qatar while advancing basic science and clinical research. Through its medical college, WCM-Q seeks to provide the finest education possible for medical students, to improve health care both now and for future generations, and to provide high quality health care to the Qatari population.

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