The Centre for Arab Studies' Report on Rare Disease Day 2018

Rare diseases are defined according to their prevalence, whereby they affect less than 1in 2000 people. It is estimated that 1 out of 17 people suffer from a rare disease, this amounts to the 7% of the population. There are more than 6000 known rare diseases, of different prevalence rates, and this number is continuously growing. It is estimated that there are 25 million people living with a rare disease in the Arab World, 30 million in Europe and 300 million worldwide. As more rare diseases are identified, and as diagnostic tools become more effective, this number is expected to rise. In an effort to raise global awareness about rare diseases, Rare Disease Day 2018 is expected to rise. In an effort to raise global awareness about rare diseases, Rare Disease Day 2018 is happening in 53 countries around the world. The theme for this year is research. In this report, the Centre for Arab Genomic Studies, a division of the Sheikh Hamdan Award for Medical Science which organizes Rare Disease Day in the UAE, aims to stress the importance of research in identifying and managing rare diseases as well as the role of patients in driving research. We also provide here a list of resources and facilities for the care, management, treatment and study of rare diseases in the UAE.

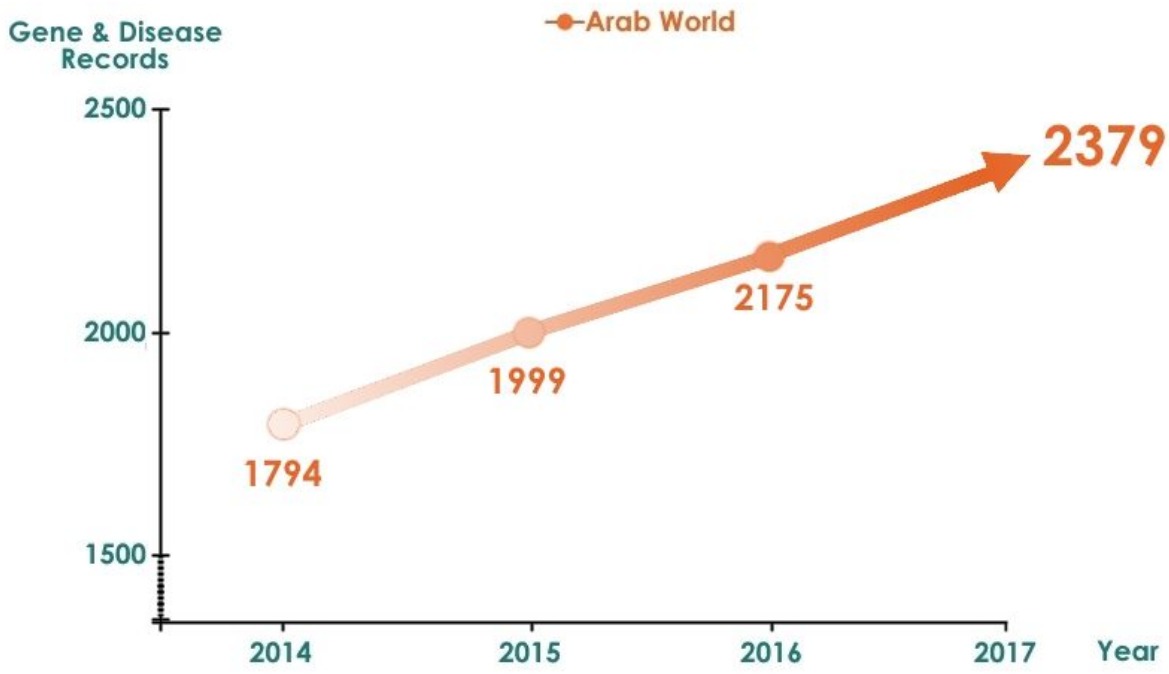
Scientific research enables the development of diagnostic tools, treatments and cures for diseases. The advancement of research requires the continuous effort of scientists, clinicians, and healthcare professionals, as well as the valuable involvement of funding agencies, policy makers, and the patients themselves. For rare diseases in particular, scientific research can be translated into real benefits for patients, as it has the potential of improving health and social care for them and their families.

As important as research is for rare disease patients, the reverse is also very true. In fact, patients are not only subjects, but also have a proactive role in research. Researchers need patients and rely upon their participation to ensure that research is meaningful. Patients drive research and help make sure it is truly relevant to their needs. Patients provide the data researchers need to discover rare diseases, essential for the development of diagnostic tools. Patients also allow researchers to learn about how the disease progresses by providing data regarding their health condition, which in turn helps researchers to determine how to stop the disease or how to develop a therapy. Moreover, by joining registries, families and patients with the same rare disease can form rare disease patient networks. These networks help create the critical mass of patients needed for research on their disease to take place.

Around the world, there has been great progress in research for some rare diseases, in big part thanks to the advocacy work of the rare disease patient community. The majority of rare diseases however, remain massively under-studied. In the Arab World, most rare diseases have no cures and only few treatments are available. To help change this, patient involvement in research needs to improve. Through this report, CAGS joins the global call of Rare Disease Day 2018 on policy makers, researchers, and healthcare professionals to increasingly and more effectively involve patients in rare disease research. This will contribute to enhanced and faster diagnosis of rare diseases and therefore reduce the number of people around the world who face the daily challenge of living with an undiagnosed rare disease. In addition, it will help develop treatments that can greatly ameliorate the quality of life of rare disease patients.

Catalogue of Transmission Genetics in Arabs (CTGA)

The first step towards tackling the burden of rare diseases in the community goes through proper characterization of the current picture of these disorders on the local and regional levels. The Centre for Arab Genomic Studies (CAGS) carries out this complex task for all genetic disorders in Arab populations. Both clinical and molecular data are collected by CAGS after being reviewed meticulously. CAGS ensures high quality of its input data through the internationally recognized process of scientific peer-reviewing. Collected data is analyzed and organized to be fed into the Centre's Catalogue of Transmission Genetics in Arabs Database (also known as CTGA). The latter has developed qualitatively and quantitatively over the past few years to become the largest ethnic-based database worldwide. As of December 2017, the number of entries in has exceeded 2300, of which 970 are gene entries (see graph below). In addition to its horizontal expansion, whereby new disease/gene entries are made, CTGA has shown a continuous rapid vertical expansion (more comprehensive coverage of existing inputs). In fact, in contrast to past years, the growth of the database is now predominantly vertical, making CTGA's coverage deeper and richer. This may be attributed in part to the increasingly wide use of NGS techniques. Screening CTGA for rare diseases in the UAE and other GCC countries yields a great wealth of information that can provide proper guidance for healthcare providers and policymakers.



List of Resources and Facilities in the UAE for rare disease patients and their families

NOT FOR PROFIT CENTRES & ASSOCIATIONS	
Organization	**Contact**
Sheikh Hamdan bin Rashid Al Maktoum Award for Medical Sciences Dubai	http://hmaward.org.ae/ + 971 4 398 6777
H. H. Sheikh Sultan Bin Khalifa Al Nahyan Humanitarian & Scientific Foundation Dubai and Abu Dhabi	http://www.sbkf.ae/main/home.php +971 2 666 5144 (Abu Dhabi) +971 4 354 4412 (Dubai)
Manzil Centre Sharjah	http://www.manzil.ae/ +971 6 534 7663
Dubai Centre for Special Needs Dubai	http://dcsneeds.com/ +971 4 344 0966
GOVERNMENT INSTITUTIONS	
Ministry of Health and Prevention	http://www.mohap.gov.ae/en/Pages/default.aspx 800 111 11
Dubai Genetic Centre (DGC) – Dubai Health Authority **Latifa Women & Children Hospital** Dubai	Genetics@dha.gov.ae +971 4 219 3276
Health Authority Abu Dhabi (HAAD) Department of Health Abu Dhabi	https://www.haad.ae/ +971 2 449 3333
Tawam Hospital Al-Ain	https://www.seha.ae/tawam/english/Pages/default.aspx +971 3 767 7444
GENETIC TESTING CENTRES & CLINICS	
Imperial College London Diabetes Centre Abu Dhabi and Al-Ain	http://www.icldc.ae/ +971 2 404 0800 (Abu Dhabi) +971 3 746 4800 (Al-Ain)
Easternbiotech Dubai	http://www.easternbiotech.com/ +971 4 3255730
Fetal Medicine and Genetic Centre Dubai	www.my-baby.net +971 4 360 4040
Al Noor Training Centre for Children with Special Needs Dubai	http://alnoorspneeds.ae/ +971 4 340 4844
Viafet Genomics Dubai	info@viafet.ae +971 4 344 0238
Medeor Medical Centre – Paediatrics Dubai	http://www.medeormedicalcentre.com/pediatrics/ +971 4 363 8080
RESEARCH CENTRES	

Centre for Arab Genomic Studies Dubai	www.cags.org.ae + 971 4 398 6777
Zayed bin Sultan Al Nahyan Centre for Health Sciences Al-Ain	https://www.uaeu.ac.ae/en/dvcrgs/research/centers.shtml +971 3 7137 452
Research Institute of Medical and Health Sciences-University of Sharjah Sharjah	http://www.sharjah.ac.ae/en/Research/SIMHR/Pages/default.aspx +971 6 5057651