Experts call for more genetic research support for those living with rare diseases in MidEast

DUBAI: This Rare Disease Day, experts are calling for increased research to unlock the answer to rare diseases which will, in turn, help the 25 million patients from the Middle East living with a rare disease. There are more than 6000 known rare diseases, of different prevalence rates, and this number is continuously growing. Most rare diseases are caused by a genetic defect, which means that children are born with the condition and will not get better by themselves. The symptoms of rare disease can often be very serious, making patients very sick or causing disabilities that impact on how long they will live and their quality of life. It is reported that rare diseases affect 7% of the population in the Arab world.

A 2018 report on Rare Diseases by the Centre for Arab Genomic Studies (CAGS) highlights that in the Arab World, most rare diseases have no cure and only few treatments are available and that, to help change this, patient involvement in research needs to improve. Although there is a need for more research in the Middle East, CAGS have successfully increased the number of gene and disease entries for the Arab population so it now exceeds 2,300 entries.

Dr Mahmoud Taleb Al-Ali, Director of the Centre for Arab Genomic Studies said, "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."

New research and techniques

International centers such as Great Ormond Street Hospital for Children (GOSH) based in London, who treat 1,500 children from the Middle East every year, also aim to help children from the Middle East through using their specialist expertise to pioneer new research and techniques into rare, genetic diseases. GOSH is home to 17 nationally commissioned services for rare diseases and has helped nearly 100 children from the Middle East with immunology problems last year.

Dr Matthew Buckland, who specializes in primary immune deficiency at GOSH says, "The Immunology services at GOSH have been at the forefront of novel therapies in rare and inherited disorders for decades. The first gene therapy in the UK for gamma chain SCID was developed here and trialed over 15 years ago. Since that time the team has worked with others in the field. The first licensed gene therapy for an immune deficiency (ADA-SCID) is now available in Europe, there are further developments in Wiskott-Aldrich, Chronic Granulomatous disease and novel approaches for other disorders to follow. Genetic discovery is incredibly important, but so is translating this into treatments that work for affected children."

Gene therapy is one of the revolutionary treatment areas GOSH specializes in. Gene therapy is the introduction of normal genes into cells in place of missing or defective ones to correct genetic disorders. It tackles the root cause of genetic disease, offering the prospect of effective and lasting treatment for children with conditions that were previously difficult or even impossible to manage.

GOSH runs more gene therapy trials for immune deficiency in children than any other centre in the world. Fifty children have now been treated with gene therapy at GOSH, including two patients from the Middle East. With ongoing support, the research team hopes to roll out the hospital's gene therapy program to a wider range of life-threatening and life-limiting genetic diseases allowing many more patients to be offered this ground-breaking new therapy as a front-line approach.

How the blind use clicks to 'see': Study

PARIS: Just as bats bounce sound waves off objects to find their way in the dark, some blind humans spontaneously make clicking sounds with their mouths to navigate the world, scientists said yesterday. Not only that, but they adjust the speed and volume of the clicks when they need to zoom in on a hard-to-place object.

"Even though people have not been 'designed' to echolocate, they have adapted their brains extraordinarily well to detect faint echoes and to instinctively adjust emissions as the task changes," said Lore Thaler of Durham University in the UK, co-author of a study published in the journal Proceedings of the Royal Society B. The research team tracked down eight blind people with experience in echolocation for the study.

They placed the "expert echolocators" in a noise-insulated room, then conducted experiments by placing a wooden disk at different angles from the person-straight ahead, at 45, 90, or 135-degree angles to their left, or directly behind them-always at a distance of a meter. Standing in one place and without moving their heads, the subjects' task was to make clicking sounds with their mouths to determine whether the disk was present in the room or not.

At zero, 45 and 90 degree angles, the participants were correct 100 percent of the time, the researchers found. Their success rate dropped to about 80 percent when the disc was placed at an 135-degree angle, just over their left shoulder. At 180 degrees, directly behind them, the participants were correct half the time.

For the larger, more difficult angles, the echolocators "increased the number of clicks they made and the intensity of the clicks," the team discovered. "Our results clearly demonstrate that people, just like bats, adjust their emissions to situational demands." The team also measured the echoes, and found they were "very faint". "Indeed, based on previous research, people would have said that it should have been impossible for them to perceive these echoes," Thaler told AFP.

The fact that blind people can teach themselves to echolocate had been documented before, he said, but never that they can adjust their sound emissions in sonar-like fashion. The clicks are made by pressing the tongue against the roof of the mouth, then quickly pulling the tongue down, creating a vacuum that "pops". The sound travels, and bounces from surfaces and objects around the blind person, returning as echoes. "What we have found is very useful when teaching people echolocation," said Thaler. "Now we can tell them, for example, that if they find a task or situation difficult or they find it hard to hear an echo they should consider making the click louder and make some more clicks and this will help them hear the echoes better." —AFP