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GENE HUNTERS ON THE AUTISM TRAIL

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Will creating a genomic data bank on autistic children and their families hold the key to early diagnosis and treatment of Autism Spectrum Disorder (ASD)? An increasing number of global researchers think so as they invest time and effort into genetics to learn more about the condition.

ASD is a group of lifelong neuro-developmental disorders that emerge during early childhood and interfere with a child's ability to socially relate to and interact with others. While some children can grow up to live independently, others have severe disabilities and require lifelong care and support. As of 2010, an estimated 52 million cases have been reported globally. The World Health Organisation (WHO) says that one in 160 children has ASD. In India, the figure is estimated to be 1 in 65.

"Autism is not one disease but a collection of rare diseases," says Dr. Khalid Fakhro, Director of Human Genetics at Sidra Medicine, a paediatric hospital, in Doha, Qatar. He adds, "Some of them have a known therapy. For example, the child may be missing an enzyme that helps metabolise fat in the brain. Supplementing this specific fatty acid could help improve the condition. There are many emerging stories like this but the fact remains that autism is a tough nut to crack. It is one of the most difficult conditions to understand. Thus the more we know, the better." The hospital has recently started a gene bank on children with autism. Its researchers will collect blood samples, fractionate them into various components such as plasma and serum and freeze them for long-term investigation.

The project aims to enrol, bank, sequence and characterise genes from 1,500 families with autistic children. Says Dr. Fakhro, "This means that we are not only taking blood samples of children with autism but also their parents and siblings. We have created epidemiological-grade questionnaires, and sought their compliance for clinical and psychological evaluation. Besides a characterisation at the molecular level, we will also assess the family history of the children. For example, pre-natal history, exposure to environmental factors such as pollution, lifestyle, eating habits at home, complications and medications in the past, history of schooling, physical activity and every other aspect. We have a 25-page questionnaire that covers all of this."

The hospital has enrolled three families since October. Dr. Fakhro adds, "We believe that the future is in therapeutics. The ultimate goal is to treat autism. The resources that we are creating will not only help us in therapeutics but also aid international researchers to understand the disorder better."

The study at Sidra Medicine is being conducted in collaboration with the Hospital for Sick Children, Toronto, Canada, which has been studying the genetics of autism for several years. A study in 2015 by the Canadian hospital found that ASD is clinically and genetically diverse. It stated: "The total number of genetic and genomic variations that confer ASD susceptibility is in the hundreds. Significant progress has been made over the past five years in identifying genetic changes that predispose a person to the development of ASD."

Another genome sequencing study carried out at the hospital revealed that contrary to what was commonly believed, siblings with ASD carry very different genetic mutations.

"Studies like these are very important. India needs to undertake such studies because at present we know nothing about the genetics of our ethnic population," says Tatyana Dias, a

neurobiologist, who runs Veruschka Foundation, a non-profit organisation in Mumbai that works for autism. “At present, the drugs that we use are general anti-epileptic, anti-depressant medications which are prescribed depending on the symptoms and patterns that the child presents with. Genetic studies will lead us to a pathway where we could possibly have a proper drug to target autism.” she says.

However, Mumbai-based developmental paediatrician, Dr. Samir Dalwai, has some reservations. “The point is that there is no single gene or a set of genes that are particularly known to cause autism. But there are over 200 genes which are associated with a higher incidence of autism. In a country as diverse as India, a much larger gene pool will be needed to thoroughly understand the disorder,” he says. Dr. Dalwai is the author of the National Consensus Guidelines for ASD by the Indian Academy of Paediatrics. He adds, “The genetic-data banking is more feasible in smaller countries such as Qatar where there is limited variability of genes. Doha’s genetic research may offer us some cues. It is definitely a positive step.”

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Reduced govt. attention has made life difficult for 2 lakh people living there

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