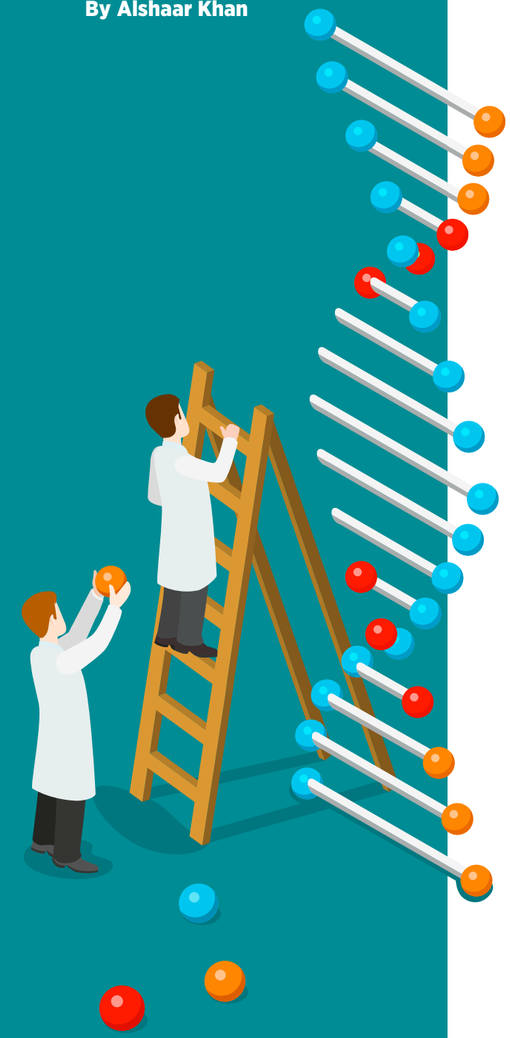
It's all in the genes

Bengaluru-based MedGenome is spearheading the genetic testing field in India.

By Alshaar Khan



IT WAS DURING A workshop at the turn of the previous decade, with Professor Jeremy Lipschultz at the University of Nebraska in Omaha, that I first chanced upon outfits like 23AndMe. Among other services, these personal genomics and biotechnology companies allow you to take an ancestry test — which essentially lets you find out what per cent of your DNA comes from populations around the world, if you have relatives across continents, and also how much of your ancestry can be traced down to the Neanderthal species that mated with Homo Sapiens around 50,000 years ago.

Though obviously excited by this information, I unfortunately didn't have the time (or money — it cost $999 back then) in America to take up the test, and no such service existed in India for the longest time. Thus my eyes lit up when I learned about MedGenome, which set up shop in India not too long ago. Unfortunately, they don't offer ancestry tracing in the country at the moment, as I was informed when I contacted the genome testing company.

But though the continuously rolling wheel of the Bengaluru-based firm might currently not comprise that cool cog, it certainly does impress with the critical and significant ones, which lay the foundation for genome research and diagnostics for ailments like cancer, metabolic diseases, neurological disorders, prenatal disorders and eye diseases. My conversations with the scientists at MedGenome in the recent weeks have only strengthened my belief in this technology, especially when it comes to the best interests of parents-to-be and their coming generations.

What exactly does MedGenome offer? The company's chief operations officer, Dr VL Ramprasad, said "Our range of genetic testing across various disease areas provides useful insights to clinicians for better diagnosis, treatment and management of diseases. It presents Next-Generation Sequencing based genomic solutions in cancer immunotherapy. Its unique access to genomics data with clinical and phenotypic data provides insights into

complex diseases at the genetic and at molecular level, and facilitates research in personalised health care."

How are the masses to make the most of this? "The genetic testing is suggested by a doctor if either he or the patient feels the need for it, on the basis of a lot of factors, like family history, consanguineous marriages (marriage within family), birth defects, genetic disorders, chromosomal abnormality etc. Also, since we have a network of more than 400 hospitals for diagnostics and 10-plus research collaborators across the country, we get samples from all over. And based on the patient location, we suggest to them the nearest sample collection centre," said Dr Ram.

A total of 400 tests are presently offered by MedGenome, covering various disease areas like neurology, oncology, cardiology, autoimmune, rare inherited diseases, endocrinology, haematology, ENT, metabolic disorders, prenatal and population genomics. But what catches the eye is the exclusive set of non-invasive prenatal tests (NIPT), which were introduced by the company to India for the first time, in 2015.

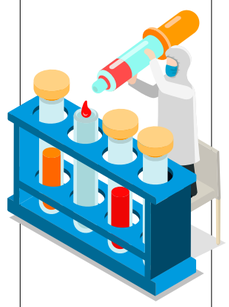
NIPT examines foetal DNA in the mother's blood, to assess whether a developing baby is at high risk for having an abnormality in its chromosomes. The liquid biopsy-based Oncotrack, launched in March this year, can identify genetic alterations and interpret, assess and treat various forms of cancer. Another such examination, the Claria Carrier Screening Test, provides vital information of 'Carrier' status to couples, and their risks of passing down recessive diseases (a condition where a person has two copies of an abnormal gene) to their child. In addition, the Sequoia Capital-backed firm also offers PGS (pre-implantation genetic screening) to check for chromosomal disorders in an IVF embryo, by checking the number of chromosomes.

"With awareness increasing and more people opting for such tests, the costs are expected to come down further. This will help even the marginalised sections of society to avail of such tests. Currently, the cost is such that not everyone can opt for it, as insurance doesn't cover them. With the government's help, it can be covered under insurance and certain tests can be made compulsory, which will benefit all sections," Dr Ram added.

In current times, when medical science has progressed by leaps and bounds, genomic analysis could add another dimension to research and prevention of critical ailments. Prenatal testing has advanced to a stage where it's possible to differentiate and check the cell-free DNA of a foetus from the mother's blood. Similarly, by testing for circulating tumor-DNA in the blood stream of cancer patients, we can better understand the present status, expected prognosis and even the response to different forms of therapy. These tests can not only detect certain abnormalities, but also provide the opportunity to choose a healthy embryo for implantation (in the case of IVFs), which can help a couple have a healthy baby.

In this air of excitement, however, Dr Ram cautions that through genetic testing, some might be trying to infer too much from genetic data, and extrapolating correlation to causation. "There may be certain gene variants that may confer a higher risk, but not necessarily enough to cause a disorder. This is especially true for polygenic disorders, such as diabetes and hypertension. And though numerous strides have been taken in identifying the underlying genes for such traits, currently a one-is-to-one correlation between the gene and the disorder is not possible," he concluded, but not before giving me the hope that ancestral testing will soon be available in the country, to settle the butterflies in the stomachs of curious souls like yours truly.

Dr VL Ramprasad



ASIAN DATABASE

MedGenome has joined hands with South Korean biotech firm Macrogen Inc and Singapore-based Nanyang Technological University (NTU) to sequence the whole genome of 1,00,000 people in Asia. The aim is to build an open genome database, accessible to researchers of public institutions and pharmaceutical companies.

LIVES MATTER

CASE STUDY 1

Just two months old, a male baby was brought to a hospital in Guntur, Andhra Pradesh, with complaints of a coloboma in the right upper eyelid, ocular hypertelorism, a bifid nose, a small nasal ala, omphalocele, renal agenesis, anorectal anomalies and bilateral undescended testis. In order to investigate the aetiology of this disorder and thereby console the distraught consanguineously married parents with at least an answer, genetic testing at MedGenome was advised. Testing confirmed a genetic diagnosis of Manitoba-oculo-tricho-anal (MOTA) syndrome, an extremely rare disorder that is predominantly confined to the Oji–cree community of northern Manitoba, Canada. This is the first case of MOTA syndrome to be reported from India. The report of this extremely rare disorder within India highlights the genetic heterogeneity of the Indian population and the enormous potential for leveraging this diversity for genetic research.

CASE STUDY 2

A seven-year old girl presented with severe fatigue and malaise and was referred to a leading cancer centre in Kolkata. Clinicians, after examinations, suspected the disease to be Myelodysplastic Syndrome (MDS) (myelo- = bone marrow; dysplastic = abnormality), which is a result of bone marrow failure to produce healthy blood cells. At MedGenome, further testing led doctors to pinpoint the exact disease - Diamond-Blackfan Anemia.
The diagnosis helped in her clinical care, and today she is on steroid treatment and is now transfusion independent.