

Tracing the trends in genetic diagnostics

By **Neelam M Kachhap** on June 15, 2015



Clinical diagnostics took a big leap with the advent of medical genetics but has it found its footing in India? Amid the euphoria surrounding this diagnostic revolution, can India overcome major analytical and interpretative challenges that have emerged in recent years? **By M Neelam Kachhap**

Cure of an ailment is no longer magical or mystical in today's time. In fact we know that accurate diagnostic is the key to right therapy. And genetic diagnostics has been able to take accuracy to a new level of truth. "With a population of 1.26 billion and 26 million births every year the burden of genetic diseases is very high. Many diseases can be predicted with great accuracy, prevented if genetic testing is done at the appropriate stage and some of the diseases can be cured or managed better if we know the precise genetic defect," shares VL Ramprasad, COO, MedGenome, Bengaluru.

The widespread awareness and recognition of rising incidence of congenital and hereditary genetic diseases in urban India has led people to seek genetic testing and counselling services. "In the urban population, congenital malformations and neonatal disorders are the third most common cause of mortality in newborns," reflects Saamant Jauhari, CFO – and genetic disorders are the third most common cause of mortality in newborns," reflects Seemant Jauhari, CEO – Research and Innovations, Apollo Hospitals.

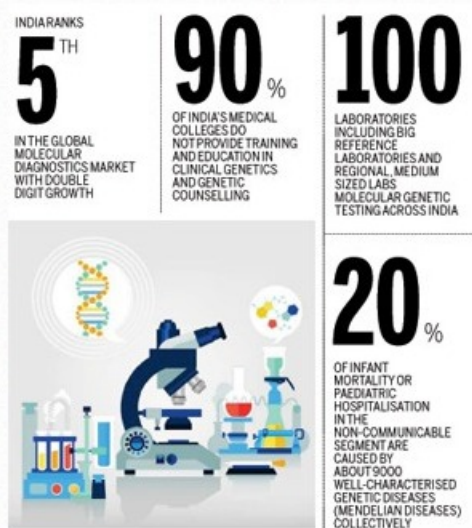
Market for genetic testing

Due to explicit benefits related to rapidity, high sensitivity and specificity and scope for detecting diseases and disorders in early stage, today molecular/ genetic testing scores over the traditional methods. "Globally and in India, genetic testing and molecular diagnostics have made notable inroads into medical practice, particularly for infectious disease testing. However, more complex and advanced testing approaches (for cancer and other complex genetic diseases) may only be in the early stages," says Dr BR Das, President – Research and Innovation, Mentor – Molecular Pathology and Clinical Research Service of SRL Diagnostics. "India holds the 5th rank after US, Europe, China and Japan in global molecular diagnostics market with double digit growth rate," he adds. Quoting a WHO case study on genomics, Jauhari says, "Genetic diagnostics market is based on physician referral, although there is about 10 per cent self-referral by the patients.

Today, after 15-20 years of frantic pace of development, molecular diagnostics is universally acknowledged as the modern face of pathology and more poignantly, as the pathology of the future. And with the arrival of new technology genetic testing has become more viable option for patients today. "With the new DNA sequencing technologies it has become practically feasible in terms of cost and time to do genetic testing," says Ramprasad. "There are about 9000 well-characterised genetic diseases (Mendelian diseases) and even though individually they are rare, collectively they account for 20 per cent of infant mortality or paediatric hospitalisation in the non-communicable disease segment," he adds.

How many centres and what tests?

A decade ago there were about a handful of centres offering cytogenetic services, with only a few clinics providing advanced cytogenetic, biochemical, prenatal diagnostic services and counselling. The Department of Genetic Medicine at Sir Ganga Ram Hospital, New Delhi established in 1997 is one of the pioneer genetics centre in India. The Genetics Unit, Department of Paediatrics, AIIMS, New Delhi; Department of Medical Genetics, Sanjay Gandhi Postgraduate Institute of Medical Sciences (SGPGI); Centre for DNA Fingerprinting and Diagnostics, Hyderabad; Indian Council of Medical Research, Mumbai; Department of Medical Genetics, Kasturba Medical College, Manipal are some of the notable institutes with good clinical medical genetics practice.



Private labs like SRL Diagnostics are also doing exemplary work. A number of new companies like Bengaluru-based MedGenome, NutraGene and Eurofins Genomics, Genotypic Technology, Strand Life Sciences; Hyderabad-based Mapmygenome, DNA Labs India; Mumbai-based Datar Genetics, Positive Biosciences; Chennai-based X-code LifeSciences are bringing genetic diagnostics closer to people. "Over 100 laboratories, including big reference laboratories as well as regional medium-sized labs across India, offer molecular genetic testing," informs Dr Das.

These centres receive samples not only from different parts of India, but also from Bangladesh, Pakistan, Sri Lanka, and Middle East and Africa.

Range of tests

India is at par with any other country in the world in terms of availability of genetic tests. "Currently, worldwide estimated 1000–1300 genetic tests are available for approximately 2500 conditions, both for rare and common. Considering the health priorities, disease prevalence and affordability, molecular/ genetic diagnostic tests pertaining to infectious diseases and cancer are more commonly available," informs Dr Das. A number of genetic tests are available in India to look at prenatal and postnatal anomalies. "The key focus areas of genetic tests are: diagnostic tests; prenatal, familial and risk assessment," says Jauhari.

Regulations

One of the gray areas in genetic testing is the absence of adequate regulations and framework. There is no official framework for assessing new genetic tests that become available in India, nor any formal system for approving which tests may be used in a clinical setting. "Presently there are no specific regulations pertaining to molecular/ genetic diagnostics in India," says Dr Das.

"PCPNDT Act regulates the practice of invasive and non-invasive prenatal tests so that they are used only to detect the genetic defects but not revealing the gender of the foetus," informs Ramprasad. Talking about prenatal genetic testing, Jauhari says, "There is a mandatory registration system for all the laboratories that provide prenatal diagnostic services in India. As per the Act, all the genetic laboratories, clinics and counselling centres offering prenatal diagnosis should be registered under the Pre-conception and Prenatal Diagnostic Techniques (Prohibition of Sex Selection) Act, 1994. The government passed this Act to prohibit the use of prenatal diagnostic techniques for sex selective abortion. Registration requires that these organisations comply with a range of quality assurance requirements."

New diagnostic methods and interventional strategies for genetic conditions that are prevalent in India are developed and validated under the guidance of Indian Council of Medical Research (ICMR) through a task force on human genetics and by the Genetic Research Centre (GRC), a permanent national centre under the umbrella of ICMR.

Accreditation is one of the ways to preserve quality and safety of the process in genetic testing. "National Accreditation Board for Testing and Calibration Laboratories (NABL) has recently modified its scope for inclusion of specific guidelines related to molecular testing. Besides few major laboratories in India voluntarily follow guidelines mandated by College of American Pathologists (CAP), US, thereby ensuring services of global standards," informs Das.

Training and education

Healthcare in India is plagued by the short supply of adequately trained personnel. Clinical genetics is no different. According to a report appearing in the Indian Journal of Human Genetics over 90 per cent of India's medical colleges do not provide training and education in clinical genetics and genetic counselling, contributing to lack of professionals specialising in these fields. "Genetics literacy in India is sorely lacking even in the current medical training curriculum. There is an urgent need for awareness and education regarding the use and interpretation of these advanced molecular tests," says Dr Das. "SRL Mumbai is the only Maharashtra University of Health Sciences recognised centre in India which offers Fellowship Course in Molecular Pathology for post graduate doctors," he adds.

Dr Shubha R Phadke, Professor and Head, Department of Medical Genetics, Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow; and President, Indian Academy of Medical Genetics opines, "We need to impart an understanding of the clinical applications of medical genetics to undergraduate and postgraduate students. Training of committed medical college teachers from various specialties like paediatrics, obstetrics, pathology, anatomy, biochemistry, internal medicine, oncology, etc. and helping them to collaboratively set up genetic units in each medical college is the only way to take clinical genetics to all clinics. Training of teachers will have rapid, long term and multiplying effects. We need to take the fruits of research in genetics to the masses through clinicians from all specialties."

"The Indian Council of Medical Research has initiated plans for creating a network of genetic centres in medical colleges and I hope this initiative proves to be successful in achieving its goal of disseminating the knowledge of genetics to all medical practitioners," she adds.

Today, genetic testing has grown from a niche speciality for rare disorders to a broad scope of applications for routine, complex diseases and personal use. However, there is an urgent need for all stakeholders to step-up and uplift genetic diagnostics, making it available, accessible and safe.

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