An estimated of 495,000 infants are born each year in India with congenital malformations, 21,400 with Down syndrome, 9,000 with thalassemia, 5,200 with sickle cell disease, and 9,760 with amino acid disorders. Currently the diagnostic and rehabilitation facilities are scarce and focused in certain limited locations but the effects of these diseases are immense. Research shows that Genetic and congenital abnormality in India is the second most common cause of infant and childhood mortality and occurs with a prevalence of 25-60 per 1000 births. The growing burden of Genetic Disorders may be multifactorial in origin, comprising of climate changes, environmental pollution, later age for conceiving a baby, hectic lifestyles etc. But this can be curbed by making genetic testing centres easily accessible for expecting couples as and when needed. Currently, government has adopted universal screening for Congenital Hearing Deficit in the state of Kerala. Also, Goa Government had started the universal screening for all newborns for Congenital Metabolic Problems. But similar programmes are needed across the country to deal with the outset as early as possible.

India is facing an accelerating demographic switch to non-communicable diseases. Congenital malformations and genetic disorders are becoming important causes of morbidity and mortality. Due to the high birth rate in India, a very large number of infants with genetic disorders are born every year, almost half a million with malformations. Factors contributing to this high prevalence include consanguineous marriages, high birth rate, and a lack of expertise in genetic counselling.

"India currently has one of the highest birthrates in the world with about 27 million births annually. Unfortunately it also has a significant infant mortality rate with close to 9 million deaths per annum. Research shows that Genetic and congenital abnormality in India is the second most common cause of infant and childhood mortality and occurs with a prevalence of 25-60 per 1000 births. The number could be lowered significantly with making early screening of pregnancy a regular practice", points out Dr I C Verma, Genetic Medicine Specialist, Sir Ganga Ram Hospital. In India, every year, an estimated of 495,000 infants are born with congenital malformations.
infants are born with congenital malformations, 21,400 with Down syndrome, 9,000 with thalassemia, 5,200 with sickle cell disease, and 9,760 with amino-acid disorders. Currently, the diagnostic and rehabilitation facilities are scarce and focused in certain limited locations but the effects of these diseases are immense. The best defenses against genetic disorders is early diagnosis through genetic tests in a proactive strategy that lets an individual and family know of what to expect.

“Genetic disorders are a growing problem globally and the recent research conducted by the March of Dimes Birth Defect Foundation states 64.4 out of 1,000 birth defects suffer from genetic disorders. The most common genetic disorders in Indian ethnicity are Beta-Thalassemia, Cystic Fibrosis, Sickle Cell Anaemia, Spinal Muscular Atrophy and Haemophilia A. The best option is to get screened before marriage or before conception. The carriers are usually healthy people, but when both parents are carriers of a mutation in the same gene, the risk of having an affected child is high. This problem of genetic disorders is increasing due to the lack of awareness regarding genetic disorders and tests which can screen them. There is a need to spread awareness on advanced tests like Carrier Genetic Test (CGT), Preimplantation Genetic Diagnosis (PGD) and Preimplantation Genetic Screening (PGS) which are aimed at preventing genetic diseases. This will scale up adoption of high end genomic tests in the country”, points out Dr Rajni Khajuria, Lab Director, Igenomix.

Genetic Testing Scenario in India
Currently, traditional prenatal screening and diagnosis methods dominate the Indian market with Amniocentesis, and Chorionic Villi Sampling having a market size of over 60 per cent. However, both these techniques, being invasive pose some risk to the developing fetus which is why many conceiving mothers do not prefer these methods. This has led to a dire need for non-invasive techniques which can detect fetal problems in early stages of pregnancy.

Although a relatively new market, prenatal screening and diagnosis market is bound to be a thriving market in India by 2024. Non-invasive techniques include examinations of the woman’s womb through ultrasonography and maternal serum screens (i.e. Alpha-fetoprotein). Blood tests for select trisomy based on detecting cell-free placental DNA present in maternal blood, also known as non-invasive prenatal testing (NIPT), have already become available across major Tier I and Tier II cities in India. NIPT has also become a highly preferred technique by doctors who consider it to be a safer, faster, reliable, less tedious and much more superior method compared to traditional techniques like chorionic villi sampling and amniocentesis.

Novel NIPT methods like pre-implant genetic diagnosis (PGD) have become a highly useful technique across many IVF centers in India which involves the sampling of cells from human embryos before implantation. Nuchal scan, also known as Ultrasound detection has gained excellent popularity as a common NIPT method in first and second trimester which is used to identify higher risks of Down’s syndrome. But genetic testing is extremely expensive ranging from Rs 12,000 to Rs 1.5 lakh and not all diagnostic labs offer standardized results.

“A nationwide study across 10 hospitals has found that NIPT is much more effective, accurate and safe for detecting chromosomal abnormalities in the unborn baby, compared to conventional
biochemical tests. This is India’s first-ever systematic study on NIPT. While the efficacy of NIPT has been well proven in the more developed markets, the results of this unique study in India clearly indicate that even for Indian women, NIPT is highly accurate when compared to conventional screening tests”, shares Dr Priya Kadam, Programme Director, NIPT, MedGenome.

Being a novel segment to enter the Indian healthcare sector, NIPT currently faces few major challenges that limit its growth to few urban regions of India such as high cost of noninvasive prenatal screening, ethical and legal hurdles regarding prenatal diagnosis and dominance of traditional prenatal tests.

“Since the 1970s, blood tests have been conducted routinely during pregnancy in India to rule out genetic diseases such as Down’s Syndrome. Today, we’re able to catch rare genetic disorders earlier. Diseases like thalassemia, dwarfism and Gaucher’s are now being identified early”, says Dr Seema Thakur, senior consultant, genetics and foetal diagnosis, at Indraprastha Apollo Hospitals.

Industry players are currently focusing on strategic expansion through acquisitions, as mergers and collaborations help the players to strengthen and enhance the product portfolio. Recently, Hyderabad based personal genomics company MapMyGenome teamed up with ThinkGenetic to help consumers, patients, and physicians with knowledge about genetics and genomics through genetic testing and genetic counselling services. Also back in 2015, MedGenome and Natera reached an exclusive agreement to provide Natera’s Panorama non-invasive prenatal test in India. Under the terms of the agreement, MedGenome received the license to develop the capacity of and to perform the test, which screens for trisomies, 21, 18, and 13, and certain sex chromosome abnormalities, such as monosomy X.

The genetic disease diagnosis market is mainly driven by the increasing global prevalence of various chronic diseases, such as different cancer types, asthma, and diabetes. The soaring popularity of personalized medicine in various developing and developed regions is a key factor propelling the market. Increasing awareness about prenatal diagnostic testing, ceaseless efforts of researchers and clinicians to prevent the development of genetic diseases, and significant advances in gene therapy are expected to fuel the market over the forecast period. In addition, the increasing role of predictive or predispositional genetic testing techniques in preventing the onset of certain cancer types has stimulated the demand for the diagnosis.

“The major disease profiles or conditions for which Genetic Testing is used globally include Alzheimer’s Disease, Cystic Fibrosis, Cancer, Down Syndrome amongst others. Cancer accounts for the largest share amongst the diseases for which genetic testing is done. This large share can be attributed to the rising prevalence of cancer, which is expected to increase as the global population ages. Around 5-10 per cent of all the cancers are caused by inheriting genetic mutation. Notable improvements can be made in the lives of cancer patients by diagnosing cancer early and avoiding delays in care resulting in greater probability of surviving, less expensive treatment and reduced morbidity. Rising prevalence of cancer worldwide will significantly accelerate the cancer diagnosis segment in the coming years”, says Arunima Patel, MD, iGenetic Diagnostics.

Types of Genetic Tests available

- Newborn Screening
- Diagnostic Testing
- Carrier Testing
- Prenatal Testing
- Preimplantation Testing
- Predictive and presymptomatic testing

Industry players are currently focusing on strategic expansion through acquisitions, as mergers and collaborations help the players to strengthen and enhance the product portfolio. Recently, Hyderabad based personal genomics company MapMyGenome teamed up with ThinkGenetic to help consumers, patients, and physicians with knowledge about genetics and genomics through genetic testing and genetic counselling services. Also back in 2015, MedGenome and Natera reached an exclusive agreement to provide Natera’s Panorama non-invasive prenatal test in India. Under the terms of the agreement, MedGenome received the license to develop the capacity of and to perform the test, which screens for trisomies, 21, 18, and 13, and certain sex chromosome abnormalities, such as monosomy X.

The genetic disease diagnosis market is mainly driven by the increasing global prevalence of various chronic diseases, such as different cancer types, asthma, and diabetes. The soaring popularity of personalized medicine in various developing and developed regions is a key factor propelling the market. Increasing awareness about prenatal diagnostic testing, ceaseless efforts of researchers and clinicians to prevent the development of genetic diseases, and significant advances in gene therapy are expected to fuel the market over the forecast period. In addition, the increasing role of predictive or predispositional genetic testing techniques in preventing the onset of certain cancer types has stimulated the demand for the diagnosis.

“The major disease profiles or conditions for which Genetic Testing is used globally include Alzheimer’s Disease, Cystic Fibrosis, Cancer, Down Syndrome amongst others. Cancer accounts for the largest share amongst the diseases for which genetic testing is done. This large share can be attributed to the rising prevalence of cancer, which is expected to increase as the global population ages. Around 5-10 per cent of all the cancers are caused by inheriting genetic mutation. Notable improvements can be made in the lives of cancer patients by diagnosing cancer early and avoiding delays in care resulting in greater probability of surviving, less expensive treatment and reduced morbidity. Rising prevalence of cancer worldwide will significantly accelerate the cancer diagnosis segment in the coming years”, says Arunima Patel, MD, iGenetic Diagnostics.
“As the advanced technologies are evolving very fast and the new genetic technologies are being introduced, this brings the need to validate all these new techniques based on preclinical and clinical research evidence for their efficacy, cost effectiveness and long term benefits before implementing them in clinical setting. There is a need of having a multidisciplinary approach including government, policy makers, regulatory committees, clinicians, geneticists, counselors and the patients, so that a proper diagnosis and counseling can be given to the family to make better decisions on choosing the right test after proper understanding”, elaborates Ramesh Hariharan, CEO, Strand Life Sciences.

Currently, there is no provision for genetic evaluation for most of the population, and no national public health programme is functional for carrier or newborn screening for genetic disorders. Although population based carrier screening for Beta Thalassemia, which was initiated as part of scientific projects, is currently being started as government supported programme in some states and has been recommended for countrywide implementation. Acceptance and performance of these programmes, and improved budget allocations is required for the possible expansion to nationwide screening in the near future.

“The number of geneticists and laboratories are too scarce to cater to the huge population of India. Improved health budget allocations, sensitization of the health authorities and policy makers towards the burden of Genetic Diseases, inclusion of genetics in medical curriculum and fostering awareness amongst medical practitioners from other fields, are important steps required for this purpose. Also, screening programmes like newborn screening, Thalassemia Prevention and Down Syndrome screening need to be implemented by the government, rather than current pattern of private providers. Data regarding prevalence of rare diseases, especially treatable ones and their registries, is essential for empowerment of patient advocacy group and proactive involvement of policy makers in implementation of appropriate screening and management programs for such patients”, highlights Dr Asmita Mahajan, Consultant Neonatologist & Paediatrician, SL Raheja Hospital.

**Key players in global genetic testing market**

- 23andMe
- Abbott Molecular
- Bayer Diagnostics
- Biocartis
- BioHelix
- BioMerieux
- BGI
- Cepher
- Counsyl
- deCODEme
- Genentech
- Genomictree
- Genomic Health
- HTG Molecular Diagnostics
- IntegraGen
- LabCorp Diagnostics
- Luminex
- MolecularMD
- Myriad
- Natera
- PacBio
- Pathway Genomics
- Qiagen
- Roche Diagnostics
- Sequenom
- Siemens

Currently, there is no provision for genetic evaluation for most of the population, and no national public health programme is functional for carrier or newborn screening for genetic disorders. Although population based carrier screening for Beta Thalassemia, which was initiated as part of scientific projects, is currently being started as government supported programme in some states and has been recommended for countrywide implementation. Acceptance and performance of these programmes, and improved budget allocations is required for the possible expansion to nationwide screening in the near future.
The growing burden of Genetic Disorders may be multifactorial in origin, comprising of climate changes, environmental pollution, later age for conceiving a baby, hectic lifestyles etc. But this can be curbed by making genetic testing centres easily accessible for expecting couples as and when needed. Currently, government has adopted universal screening for Congenital Hearing Deficit in the state of Kerala. Also, Goa Government had started the universal screening for all newborns for Congenital Metabolic Problems. But similar programs are needed across the country to deal with the outset as early as possible.

Genetic Counselling
Another area that needs to be highlighted is genetic counselling. It is an important clinical practice since it assists patients in choosing preventive screening test, adopting healthy behaviours, and making decisions about having genetic tests and preventive measures. Communicating accurate genetic risk levels effectively is the core process of genetic counselling. However, due to the complicated nature of the genetic risk, conducting such communication is...
often challenging in clinical settings. For this reason, genetic counsellors are required as experts to carry out such tasks effortlessly.

“The number of genetic counsellors are gradually increasing in India. Specialized courses are being implemented in this direction. In 2014, VIT University, Vellore started a post graduate program in Biomedical Genetics with a specialisation in Genetic Counseling. Up until then, there were a couple of genetic counseling programs for MBBS students as a specialty and a couple of diploma programs for non-medical students. Very recently, Osmania University has also started a course on genetic counselling emphasizing on prenatal and postnatal diagnostics”, points out Pooja Rayasam, Genetic Counsellor, Dhiti Omics Technologies.

Genomics based Research
On the other hand, the excellent science that is going on in India is only a fraction of what can go on. There are only a few institutes that are working in the field of medical genetics. “In view of the huge ethnic-diversity of humans in India, complexities of mixing of genetic pools leading to desirable or undesirable manifestations thereof, there is a huge need to increase focus on human genetics research. Currently there are just a few focused R&D institutions carrying out work and this needs to go up. R&D funding has to increase and teams need to work in a well-coordinated manner. Indian Genetic History is a fascinating topic owing to its incredible diversity carried over several centuries and there is lot to understand and discover. A lot can be done through innovative research in this field such as convergence of new technologies to enable earlier, faster and precise diagnosis of genetic predisposition followed by counseling of couples, and to develop replacement therapies if it is clear that the disorder is due to a mutation leading to deficiency of a particular metabolite(s)”, explains Dr Jagadish Mittur, Principal Consultant, Biotechnology, Department of IT, BT, Government of Karanataka.

Very recently, the Council of Scientific and Industrial Research’s Delhi-based Institute of Genomics & Integrative Biology (CSIR-IGIB) announced the commercialisation of a set of 27 genetics tests it has developed over the years. The institute has entered into an agreement with Dr Lal PathLabs, which has a large network of diagnostic centres across the country, for licensing the genetic tests. “The genetic tests licensed would provide a much needed support system to doctors in diagnosing identified genetic disorders and would, thus, help the patients eventually”, shares Dr Girish Sahni, Director-General, CSIR. IGIB has also embarked on a unique outreach programme called GOMED (Genomics and other Omics technologies for Enabling Medical Decision). The programme provides a platform for clinicians to tap into the rich and varied expertise of CSIR-IGIB in disease genomics to solve clinical problems.

Control and management of the genetic disorders depend on identification of the variants in the genome that are causally linked with the disease. The need of the hour to improvise the situation is integration of genetic services into primary health medical services, appropriate genetic counselling by trained physicians, increasing the number of departments of medical genetics in medical schools and to give an insight to communities largely affected by this.  

Dr Manbeena Chawla
manbeena.chawla@mmactiv.com