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ASIA EDITION

Genetic diagnostics usher in a new era in healthcare

18 December 2012 | Analysis | By BioSpectrum Bureau

Thanks to the Human Genome Project, scientists now have a set of tools that can be readily used to understand the complexity of diseases and their variability in human beings. These tools assist scientists in refining risk prediction and also in evaluating the response to therapies with greater precision. The project has in turn fuelled the diagnostic industry, which was not perceived to be as exciting as therapeutics but is now one-of-the-fastest growing segments of healthcare.

From being a method to identify whether a patient has a specific disease, diagnostics are now being used to support clinical development of drugs, predict diseases before appearance of symptoms, forecast the progress of a disorder and identify patients who are most likely to respond (or not respond) to specific treatments.

Solutions from the APAC

Currently, healthcare applies a trial-and-error method where patients and their physicians are faced with uncertainty of outcome. In the patient's case, it often entails greater physical and financial burden. Advances in genomics enable scientists to examine the DNA of an individual and predict if the person is susceptible to a particular disease or whether a particular medicine will suit him or her.

As a result, companies have started investing in pharmacogenomics, which offers advantages such as eliminates the unpredictable nature of drug development, to bring new products to the market.

Indian start-up Xcode Lifesciences has come up with the In DNA technology to provide solutions to lifestyle-related diseases, such as coronary diseases, diabetes and obesity. A person can order the test online. Following which a saliva kit is shipped to the customer. This kit, which is non-invasive and safe to use, is used by the customer to ship a smaple of his saliva back to the testing lab. DNA extracted from the saliva is used to determine the allelic information of the individual using high-throughput genotyping techniques.

Talking about the new kit that the company launched this year, Dr Saleem Mohammad, CEO and co-founder of Xcode, said, "Xcode's preventive program, Lifelong Wellness, is a unique offering that combines DNA and healthy living. Lifelong Wellness aims to empower individuals with knowledge about their genes and pointers to modify their lifestyle, so they can lead a

healthy life." The genetic assessment provided by Xcode gives insights into two things: health risk, especially to diabetes, obesity, CVDs and stroke, of a person and his or her metabolic profile or how the person metabolizes fats and carbohydrates. "Using this information, our team prepares nutrition and fitness plan that is personalized because we provide recommendations that are best suited to your genetic makeup," he said.

Molecular Diagnostics tests are part of a new wave of cutting-edge technology that analyse genetic information. Australia's Universal Bionsensors is conducting research into molecular diagnostics at the point-of-care. The market for molecular diagnostics is valued at more than $5 billion worldwide and is expected to grow at over 15 percent annually. The company is looking to combine some locally licensed technology with its own proven biosensor technologies to detect DNA or RNA electrochemically. This would imply that the tests would take minutes, rather than hours, and the system would be simple to use and cost-effective.

In April this year, NutraGene launched India's first commercial genetic test for type 2 diabetes. NutraGene's Type 2 Diabetes Genetic Scan is a DNA testing service that screens DNA variations that have been widely replicated as risk factors for type 2 diabetes. It is based on a buccal (cheek) swab sample and the methodology of targeted mutation screening (genotyping). The tests cost around $136 (Rs 7,500) and also include complimentary genetic counselling to help customers and their physicians understand patients' genetic results.

Such preventive healthcare programs are important for the patients because it allows them to take proactive measures to prevent or, at worst, postpone the onset of what would otherwise have been an inevitable consequence. For example in diabetes, a disease that has got familial inheritance pattern, it has been established that a pre-diabetic prognosis could prevent or postpone the onset of diabetes significantly through a program of dietary changes, regular exercise and preventive doses of Metformin.

UAE-based start-up Eastern Biotech and Life Sciences provides preventive diagnosis for diabetes cancer (breast cancer, hereditary non-polposis colon cancer, malignant melanoma) and cardiovascular disease. The disease-specific test costs around $700 whereas the cancer tests cost over $1000. The company also has several other solutions in the genetic diagnostics space such as pre-marital genetic screening, prenatal diagnosis, including non-invasive test for trisomy, new-born screening, lifestyle disorder screening and whole genome scan.

Metropolis, an India-based lab that processes over 10 million tests a year, too has pharmacogenomics-based solutions for cardiovascular diseases, cancer, KRAS mutation analysis and EGFR mutation analysis. The prices start from $36 (Rs 2,000) for the tests.

Another start-up based in Singapore, VolitionRx, is developing epigenetic cancer diagnostics with broad applicability. The company's initial focus is on colorectal, breast, lung and pancreatic cancers. Dr Mark Eccleston, external collaborations manager and chief scientific officer, HyperGenomics division, VolitionRx, explains the company's technology. "VolitionRx is developing a range of epigenetic cancer diagnostics based on our proprietary Nucleosomics technology platform. This technology allows epigenetic profiling through a liquid biopsy (blood, urine etc.) in a standard ELISA format."

"Specific cancers will be identified through their unique profiles based on a combination of histone modifications, histone variants, DNA modifications in nucleosomes (DNA or protein complexes released by dying cells) and adducts between nucleosomes and other nuclear proteins such as steroid receptors. Volition is developing this technology for a range of diagnostic and prognostic applications as well as well as therapeutic efficacy monitoring," he added.

Jai Health, a start-up in India, is a personalized healthcare solution provider that has launched its product Jai Heart, which is India's first genetic test for total cardiovascular risk. Heart disease arises as a result of a combination of genetic, environmental and lifestyle risk factors. With the discovery of major genetic risk factors over the past few years, the company has developed Jai Heart test to detect this risk and to bring that information together with the assessment of conventional risk factors.

Dr Sanjay Kakkar, founder and chairman, Jai Health, said, "Jai Heart is the first genomics-based risk estimation solution for heart disease developed specifically for Indian, other Asian, Middle Eastern and African populations. Jai Heart provides an overall estimate of an individual's susceptibility to developing heart disease facilitating more targeted screening, earlier intervention and prevention."

Jai Heart analyses DNA from saliva and cheek cells to look for genetic variations associated with heart disease. This is integrated with the client's medical history and conventional risk factors provided online at the time of registration. A detailed

personalized report is produced that estimates the risk of developing heart disease, highlights individual risk factors and outlines what the client can do to prevent heart disease. Importantly, the incorporation of genomics into this test can give a signal of risk long before heart disease becomes symptomatic. This will help clients and their doctors initiate the necessary lifestyle changes, and initiate investigations and medical interventions to reduce the risk of a heart attack.

The client is sent a kit containing a saliva or cheek cell collection tube, which is simple, non-invasive and easy-to-use, through courier. The kit can be couriered back for analysis and the client is informed electronically when the report is ready to be viewed. Jai Heart is priced below $90 (Rs 5,000).
Xcyton Diagnostics, a medical biotech company which develops products for diagnosing, managing and containing critical infectious diseases, launched its Syndrome Evaluation System (SES) platform earlier this year.

The patented platform is capable of identifying 26 different pathogens in a single sample in a single test. Using the SES platform, Xcyton has developed kits, trade marked as Xcyto Screen, for critical infections such as brain infection, eye infection, septicaemia, infection of Febrile neutropenia, acute pyrexia and HPV genoptyping (for cervical cancer). The tests cost between $271-$325 (Rs 15,000-Rs 18,000) per patient sample. One of the key players in the genetic diagnostics segment is Genetic Technologies of Australia. The company offers a range of diagnostic tests that are primarily focused on cancer, in particular women's health.

Dr David Sparling, vice president, legal and corporate development, Genetic Technologies, said, "Our suite of women's health diagnostic tests include tests for breast, ovarian and endometrial cancer, and we are constantly looking to expand our test menu. We also offer diagnostic tests for bowel cancer, cancer of unknown primary and epilepsy."

The company launched its product BREVAGen, a first-in-class predictive risk test for sporadic breast cancer, in 2011. The company is now looking to expand BREVAGen sales into new regions and is on an acquisition footing to acquire late-stage development or on-market diagnostic assets that fit the company's objectives.

Pitfalls for the players
The genetic diagnostic industry is still in its infant stages. There are several hurdles for the companies operating in this segment. For instance, there is lack of awareness among people and medical professionals about the available diagnostic products. Also, combining genetic markers with conventional risk factors is not a common practise. The genetic markers, when combined with conventional risk factors using intelligent algorithms, have the ability to estimate the total risk of an individual developing a disease, thereby enabling better prevention and treatment through personalization.

"For the effective prediction of risk and subsequent prevention or treatment of complex multi-factorial disease, such as cardiovascular disease and diabetes, genetic diagnosis alone is of little use. Genetic markers need to be combined with conventional risk factors to improve the estimation of total risk of disease," said Dr Sanjay Kakkar of Jai Health.

The regulatory landscape is another hurdle. "There are numerous organizations offering direct to consumer (DTC) as well as un-validated or poorly validated genetic tests," adds Dr David Sparling of Genetic Technologies.

Dr Faruq Badiuddin, chairman, Eastern Biotech and Life Sciences, agrees. "Regulatory environment in this segment is still immature. This industry, as far as I know, is poorly regulated, especially in Asia. It is not big enough for the regulators to allocate significant resources to this industry," he said. He also highlights another major challenge which is the shortage of trained professionals to offer genetic counseling.

"Current medical professionals are not well-equipped to handle pre-and post-test counseling and there isn't enough qualified genetic professionals to offer the services on a wider scale. Although the US and the UK are heading towards making it mandatory for a genetic testing provider to offer genetic counseling services, but this is not yet the law," he said.

Dr Nilesh Shah, COO, head technical services, Metropolis group, also notes that "there is shortage of qualified personnel with required competencies to carry out genetic testing."

There are other hurdles such as high costs of genetic diagnosis and the data specific to a population is scarce.

The future looks bright
Genetic testing is likely to increase and will be driven by the demand for tailored approaches. A lot of innovation and exciting science is happening in the genetic diagnostics segment. One interesting point to note is that in this relatively new segment of smart healthcare, start-ups are the ones who have taken the lead. One of the reasons behind this could be that a start-up

needs to bring novel solutions to the market rather than adding to the exisiting product line.

The companies need to keep in mind that the solutions they offer need to be cost-effective to have a wider reach. The day is not far when a doctor would look at the patient's genome data to prescribe a medicine and start a treatment.