GENOME SEQUENCING

CHANGING THE PARADIGM OF CANCER DIAGNOSIS & TREATMENT

Over the past few years, cancer has become the second leading cause of death in India. Indian Council of Medical Research (ICMR), through its National Cancer Registry Programme reports that more than 1300 Indians die every day due to the disease, attributed largely to poor prevention, diagnosis, and treatment. Various kinds of cancer are reported every year, most common of them being breast, oral, cervical, lung, stomach and colorectal cancers, accounting for nearly 50% of all new cases. Out of these, breast cancer is among the other cancers with higher mortality rates. The incidence rates of cancers are caused by both internal (genetic, mutational, hormonal, poor immune conditions etc) and external or environmental factors (food habits, social reasons, industrialization, overgrowing population, etc.) and associated mortality rates has led to a lot of efforts being channelized to study the disease, by researchers and clinicians across the globe. The past five years has brought about many significant achievements in the field of cancer genetics, driven by rapidly evolving technologies and plummeting costs of next-generation sequencing (NGS).In particular, The Cancer Genome Atlas Project, a pan institution effort across different countries, has led to mining of genomic data which is being channelized to drive targeted therapies for different kinds of cancers.

Though challenges remain, the increased affordability and accessibility, of sequencing platforms and bioinformatics analysis capabilities, and multigene panels has brought NGS to the clinics. The speed of sequencing technology has progressed to such an extent that sequencing of even entire human genomes can be carried out within 24 hours.

Illumina Inc. is the global leader in genomics, and creates sequencing and microarray tools to fuel ground breaking advancements in lifesciences research and molecular diagnostics. Premas Life Sciences (PLS) represent Illumina's cutting-edge tools in India and works closely with researchers and clinicans to evangelize this technology for better research and clinical outcomes.

What experts have to say

Dr Ramprasad V.L., COO, MedGenome Labs Ltd

The presence of a BRCA1/2 mutations leads to an increased risk of developing breast and ovarian cancer. Due to the rising incidence of breast cancer in India, it is important to understand the risk of developing breast cancer as well as possible prevention or screening options before the onset of cancer. Over and above risk assessment, the presence of BRCA1/2 mutations help in targeted treatment for cancer patients. BRCA1/2 mutated breast and ovarian cancer have shown good response to targeted therapy using PARP inhibitors. Recently, in 2018, FDA had approved PARP inhibitor for breast cancer patients, based on the diagnosis result, which showed, significant improvement in progression free survival (PFS) and overall survival (OS) in patients.

Over the years the diagnostics industry has grown tremendously with newer technologies. The Next Generation Sequencing (NGS) is one such technology that is enabling faster processing of samples which is reducing the turnaround time while giving accurate results. This has helped aid precise diagnosis of cancer and makes clinicians make an informed treatment decision."

Dr Sunita Polampalli, MD Path, Kokilaben Dhirubhai Ambani Hospital

Breast cancer is the most frequent malignancy in women worldwide and is curable in ~70–80% of patients with early- stage, non- metastatic disease. Approximately 10% of breast cancers are inherited and associated with a family history. Women with BRCA mutations also have a higher risk of developing ovarian cancer and some other cancers. Men with BRCA mutations also have a higher risk of breast cancer, as well as prostate and some other cancers. A contributory factor to the demand for BRCA testing has been heightened public awareness of the consequences, costs and prophylactic options surrounding BRCA testing. Against this backdrop of rising demand, more diagnostic laboratories are adopting next-generation sequencing (NGS) technology for BRCA testing, which offers the potential of fast, scalable, cost-efficient and comprehensive sequencing. The rapid evolution of NGS technology is revolutionizing the management of inherited diseases, where traditionally molecular diagnostics have been underused due to the issues of cost, time, labour and availability of services.

Dr Rajiv Sarin, In-Charge, Cancer Genetics Unit, Tata Memorial Hospital & ACTREC

Normal cells of our body could become cancerous when certain genes critical for their functioning are altered or mutated. The Genetics Clinic at the Tata Memorial Hospital has enrolled almost 9000 such cancer prone families who are offered genetic counselling, genetic testing and cancer prevention advice based on genetic test results. In the last 10 years, NGS has revolutionized the ways we look at 10-100 or sometimes all the 22000 human genes. The cost, time and accuracy of NGS based genetic testing continues to improve year after year and now the entire genetic makeup; called whole genome of an individual or a single cell can be studied very accurately and quickly. In addition to predicting hereditary cancer risk, NGS based analysis of tumour cells in many blood cancers, childhood cancers and some other cancers helps in making the molecular diagnosis and prediction as how a person's cancer is likely to behave or respond to a particular type of targeted therapy.

Dr Phil Febbo, Chief Medical Officer, Illumina

Tumor tissue diagnostic biopsies are a critical component in cancer care. These valuable samples confirm whether a patient has cancer, provide a snapshot of the tumor-host microenvironment, and help determine a malignancy's potential aggressiveness. Add in next generation sequencing (NGS), which provides comprehensive molecular insights, and biopsies can further improve risk stratification, identifying specific mutations and genomic markers that can predict therapeutic benefit. As we make progress towards using liquid biopsies to better manage patients known to have cancer, there is growing interest in developing tests that detect tumors during their earliest stages, when they are most treatable. In this vision, liquid biopsies could be part of a standard annual physical, like cholesterol and blood glucose tests. In the near future, we will move from single-sample analyses that provide only one, transitory snapshot of a tumor's progression, to longitudinal studies that track cancer over time. This technology, combined with targeted therapies, immunotherapies, and other emerging approaches, will ultimately help us control or cure cancer in many patients.

Dr Ashutosh Upadhyay, Product Manager-Clinical Genomics, Premas Life Sciences

Cancer care is certainly one of the biggest challenges of the 21st century. We have greatly improved our knowledge for cancer in the last two decades. Information gathered through various studies have revealed a huge variability that can be found not only between different types of cancer but also between patients with the same type of cancer. Now it is evident that we need diverse therapies to cover the gamut of cancer. In recent years, there has been a surge of new technologies which made a big difference in the way we treat cancer. With the increasing number of biomarkers in cancer research, NGS technology has provided options for simpler usage with functional advantage. NGS is one of those technologies which has provided a holistic approach to look at the issues. This approach has allowed oncologists to reorganize the way they think about cancer and make treatment recommendations based upon genomic drivers of tumorigenesis.