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Personalized medicine for breast cancer

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As we all know that the incidence and prevalence of cancer is increasing day by day. There is an increased interest of researches medical professionals and even general public in the field of cancer.

There has been a change in the trend of treatment modalities and medications with newer guidelines in place due to an increased research in the cancer. These novel medications are aimed at causing lesser side effects. Better medications are available to manage these side effects efficiently.

What is precision medicine and why the need?

Cancer arises from one's own body cells. These cells, due to complex environmental and genetic and epigenetic interactions,

undergo genetic makeup changes (i.e mutations) and turn cancerous. As each individual is different from other individuals, hence each person's cancer is different from other. A study of each individual's genomic profile (known as a molecular profile) and clinical profile is the basis of precision medicine. Since it caters to a specific person, so it is also called personalized medicine (pertaining to that particular individual).

Thus, through this precision medicine, we aim to deliver treatment that is more specific and supposed to be more efficacious for that individual. For example in breast cancer patients, a drug called Trastuzumab is given to the patients with a specific gene profile called Her2neu gene amplification. Similarly, in patients with hormone receptor (estrogen and progesterone) positive, hormonal therapy is shown to help in decrease the chances of recurrence.

In cases where ER/PR and Her2neu are negative, they are referred to as Triple Negative breast cancer. In such cases, another gene known as BRCA 1 & 2 is tested. If BRCA 1 & 2 is positive (i.e mutated), in addition to starting targeted therapy with oral tablet Olaparib, family counseling for screening is done.

It is estimated that who has prior testing for pathogenic BRCA 1 & 2 mutations as negative, yet some still harbour and undiscovered pathogenic BRCA 1 & 2 mutations (due to limited sequencing) or pathogenic mutations if another cancer susceptibility gene. 3-4 % of high-risk individuals have germ-line pathogenic mutations in cancer risk gene other than BRCA 1 & 2 which include ATM, CHEK 2, PALB2, PTEN, TP53, CDH1, RAD51D, MSH6, NBN, RAD 51C etc.

The advent of next-generation sequencing (NGS) led in the last few years had led to a massive increase in cancer molecular profiling; allowing the characterization of DNA sequenced variants in tumour tissues to better understand cancer progression and to index cancer genomes ultimately aiming to inform therapeutic decision. NGS techniques are still expensive (although prices are decreasing considerably) for the sequencing of the entire human genome (3Gb) and are outside of the current reach of clinical diagnostic laboratories.

Targeted sequencing, including exom sequencing (coding regions {i.e, 1% of the human genome 28Mb}), or sequencing of a subset known genes or mutations hotspots (targeted NGS) are more routinely for clinical testing and research. Targeted NGS allows analyzing several hundreds of mutations located in oncogenes and in tumor suppressor gene (TSG) using dedicated cancer panel kits, thus expediting molecular diagnosis.

In conclusion, the cancer precision medicine will leverage advances in biotechnologies, such as next-generation sequencing, proteomics, transcriptome, epigenetic, pharmacology and bioinformatics, to identify precise causes for cancers and develop tailor- fit personalized therapies. One shoe fits for all approach is no longer valid.