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Spotlight on Comprehensive Genomic Profiling

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Over 1.2 million mutations in 350 genes in a human body causing cancer make every cancer case a unique one

From the understanding of 250 types of cancers by the conventional diagnostic methods the medical field is moving fast forward to identification of mutated cell. “The medical field has moved from histological diagnosis in 2000 to molecular diagnosis in 2018,” said Dr Christian Rommel, Global Head, Oncology Pharma Research, Roche.

The new generation technology now knows the uniqueness of each cancer case by understanding what mutations in which of the 350 genes that contribute to cancer development cause it. Dr Rommel informed that scientists are targeting to find more genes that may contribute to cancer.

“Advances in cancer biology are driving future and cancer care is becoming personalised,” said Josh Jordon, Lifecycle Leader, Foundation Medicine, Roche. Advances in genomic testing are driving the personalisation of cancer care and helping physicians understand patients’ unique cancer types.

This uniqueness of each cancer case leads to precision and personalised medicine which is “changing the treatment paradigm and offering a big promise to cancer patients,” said Oliver Bleck, GM, Roche Pharma, Switzerland. He was addressing the media from different countries at a media event on “The future of personalised healthcare: Advances in precision medicine and genomics” held at the Roche Innovation Centre in Zurich.

The two-day event by Roche and its partner Foundation Medicine, a molecular insight company from the US, introduced to the international media the new emerging world of Comprehensive Genomic Profiling (CGP) leading to precision medicine and targeted & personalised healthcare. Several executives and researchers of Roche and Foundation Medicine, as well as academicians from universities made presentations on various aspects of genomic testing and its importance in understanding cancer and treating it.

Charlotte Colthorpe, Group Global Scientific Director, Medical Affairs Team Lead, Skin and Rare Cancers, Roche, said, “Transition to precision medicine leads to paradigm shift in cancer care.” But the main requirement of precision medicine is a massive data of patients. Researchers need large database so that even for the rarest type of cancer they have large sample size to understand the specific cancer.

“Foundation medicine has been building such a database for the past nine years and has now one of the largest database

containing genomic profiles of over 300,000 cancer patients," said Dr Prasanth Reddy, VP, Medical Affairs, Foundation Medicine. The Foundation Medicine is providing services like FoundationOne® CDx which is first FDA approved broad companion diagnostic designed to provide physicians with clinically actionable information based on the individual genomic profile of each patient's cancer. Its FoundationOne Heme is a comprehensive genomic profiling test for hematologic malignancies and sarcomas and FoundationOne Liquid is next-generation liquid biopsy test for solid tumors utilizing circulating tumor DNA (ctDNA). All these services are available in India too.

Josh Lauer, Global Head, Personalised Healthcare Market Development, Roche, said that for driving value from data and analytics what was needed was meaningful data at scale and advanced analytics. With high resolution view of each patient "we have increasing level of information and the challenge was that how we use it perfectly," he added.

Benjamin Szilagyi, Business Lead, Enhanced Data & Insights Sharing, Roche, said that meaningful medical data gives exquisite insights in the disease in the last 30 years. When asked about the requirement of facilities like artificial intelligence (AI) for analysis of such a huge data, he said, "before analysis even data management is a problem that will have to be looked into. Even in some developed countries the data is stored on paper, which needs to be changed."

"The data of Swiss patients is kept in the lab here only and the patients have the choice to give data for research or not", clarified Prof Holger Moch, Medical Director, Department of Pathology and Molecular Pathology, University Hospital Zurich (USZ). He explained how the role of pathologist has changed in molecular pathology. "It is two-fold and evolving," he said.

He explained physicians are now able to personalise treatment to the unique molecular profile of the patient and thus precision medicine was driving personalised healthcare. He informed that Roche, Foundation Medicine and the USZ have joined forces in an academic-industry collaboration to improve personalised cancer therapy using comprehensive tumor profiling and to enhance personalised and targeted therapy options.

USZ runs the lab to do genome profiling and generate reports of the cancer patients in Switzerland on the basis of the Foundation Medicine system. "When a patient is diagnosed with cancer, his genome profiling is done here in the lab", said Dr Martin Zoche, Director, Molecular Tumor Profiling, USZ while showing the media persons functioning of the lab. The profiling costs $ 4000 in the US and the process takes 14 to 20 days.

Dr Abdullah Kahraman, Head, Clinical Bioinformatics in Molecular Tumor Profiling, USZ, explained about the report generation. Besides the gene alternations, the report also carries potential therapy options and relevant clinical trials. "It is a simplified clear report," said Dr Merlene Thomas, Global Medical Lead for Integrated Molecular Medicine, Roche.

The report is then matched with the existing database to find out matching profiles of earlier patients and then put before the Molecular Tumor Board. The board facilitates its interpretation, assign precise treatment and provide education, she said. "How to integrate ever-increasing amount of information into clinical decision is a challenge", she added.

Prof Alwin Kramer, Head, Clinical Cooperation Unit, University of Heidelberg, who discussed the cancer of unknown primary (CUP), said that CUP incidences have decreased, probably due to improved diagnostics, but mortality has not improved. He added that CGP has the potential to transform how we care for even CUP patients as it opens up the possibility to bring personalised therapies to patients with CUP. He described the details of CUPISCO, a Roche-initiated, multi-cohort trial in CUP patients based on genomic profiling for comparing the efficacy and safety of molecularly-guided therapy versus platinum-containing chemotherapy in patients with newly diagnosed, previously untreated CUP. "It is planned at 128 sites in 32 countries over 790 patients", he said adding 193 patients have been enrolled till now. Every patient in the CUPISCO study will be discussed in molecular tumor board, said Prof Moch.

Prof David Thomas, Head, Cancer Division, Garvan Institute, gave details of WINTHER, I-PREDICT, TARGET clinical trials launched to study various aspects of CGP. He predicted that molecular guided therapy will be routine in clinical practice.

Later in a panel discussion he said, "We are measurably improving patient survival." He pointed out that molecular testing can be wrong some time and we are re-analysing the data. Kramar said that over 80 molecular track treatments were approved. It was extremely difficult to do randomised studies, he added.

Bogi Eliasen, Special Advisor on Future of Health, Copenhagen Institute of future studies, gave details of Faroe genome project, a holistic genomic health approach in Faroe in Denmark. He said, "Genomics is going to be the most important baseline to be worked with." Prof Roger Moos, President, Swiss Group for Clinical Research, said that 32 per cent of cancer patients in Switzerland were treated outside of treatment guidelines leading to lack of data collection. He gave details of the Swiss Group of Clinical Cancer Research (SKSS).

The event also included narrat ions of experiences by the cancer survivors. Prostate cancer survivor Bryce Olson, Global Strategist, Health and Life sciences Group, Intel Corporation; breast cancer survivor journalist Susan McClure, Founder and CEO, Genome Creative; and sister of a cancer victim Ruth Knott, Co-founder and Head of Governance, the SJK Foundation, presented their experiences of passing through cancer and cancer care respectively.

“The one-size-fits-all approach to healthcare no longer cuts it, and it is time for people to understand that in order to get targeted treatments tailored uniquely to them, they have to do some form of genomic testing,” said McClure, who has devoted her life to better educating the public about genomics.

Milind Kokje

(Milind Kokje was invited to participate in the Media Event at Zurich and his trip was sponsored by Roche)