Integration of Next Generation Sequencing, Artificial Intelligence and Machine Learning in Cancer Diagnostics: A Major Leap Forward

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Cutting-edge recent advances in the field of diagnostic and preventive medicine are bringing about a paradigm shift in the ways physicians used to manage their patients. The development of the latest diagnostic equipment is being further powered by the availability of various analytical tools not only enhancing diagnostic and preventive precision but also facilitating earlier diagnosis as well as effective disease monitoring.

Cancer is a major healthcare burden across the globe. In the year 2020, 19.3 million new cancer patients and a total of 10 million cancer deaths were registered across the globe.[1] Cancer progression involves complex molecular circuitry involving various molecular players of cellular as well as stromal origin. Moreover, genetic variations (both somatic and germline) drive the tumour-specific cellular transformation to a challenging battlefield where the already exhausted (and sometimes altered) immune players struggle to maintain a balance between the pro-and-anti tumour responses in cancer patients.[2,3] It is therefore highly relevant to identify molecular players in cancer progression, not only to better understand the pathogenesis but also to discover novel molecules of diagnostic, therapeutic and prognostic significance.

In this context, the integration of the next-generation sequencing (NGS) methods in cancer diagnostics is a major step forward. It has not only revolutionised our capacity to analyse the genomic profile of cancers but has also enabled us to monitor therapeutic responses to various drugs available.[4] This high-throughput massively parallel-sequencing technique offers unprecedented accuracy and highly valuable insights to driver (and other) mutations bringing about cancer progression and relapse. The groundbreaking advancements in the NGS technology also offer minimally invasive diagnostic procedures, such as liquid biopsy, which have significant potential to improvise patient care, diagnostic accuracy, and to devise strategies for personalised medicine. Moreover, NGS can be exploited to identify novel molecular/genetic biomarkers to identify cancers at early stage and to formulate effective diagnostic, screening as well as treatment protocols.

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The NGS technology generates huge and complex data sets of genomic information which are beyond the manual human capacity to decipher meaningful insights from these data. Coupling of such complicated, and difficult to analyse, data analytics to the powers of artificial intelligence (AI) and self training machine learning (ML) algorithms considerably improves our understanding of disease processes and identification of actionable variants. Smart diagnostic capacity of AI and ML algorithms are already making their impact in cancer diagnostics, suggesting the powers of AI and ML in the realm of NGS-based cancer diagnostics.[5]

While we cherish these advancements in cancer diagnostics, there are serious challenges associated particularly in a resource-limited setup. The cost of the kits/reagents required for performing NGS poses a major challenge.[6] Trained and skilled human resources involved in technical aspects as well as to undertake meaningful clinical informatics are also required. Moreover, the identification of actionable genetic variants is meaningful only if relevant/appropriate therapeutic modalities are available to practice personalised medicine.

It is therefore very important that relevant policy-makers/organisations devise appropriate strategies to support and invest in research and development in this domain, so that such interdisciplinary approaches may benefit cancer patients. Taken together, the integration of NGS in cancer diagnostics holds a promising future and is a major leap forward in cancer care and management of cancer patients.

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