Trinity College Dublin
Coláiste na Tríonóide, Baile Átha Cliath
The University of Dublin

Trinity Translational Medicine Institute
Trinity Centre for Health Sciences

Translational Cancer

Representative Case Study — Development of a semi-conductor sequencing-based panel for genotyping of colon and lung cancer by the Onconetwork consortium.

Prof. Orla Sheils

Clinical and Market Need

Personalised Medicine relates to the broader concept of patient centred care, which takes into account that, in general, healthcare systems need to better respond to patient needs. It refers to a medical model using characterisation of individuals' phenotypes and genotypes (e.g. molecular profiling , medical imaging , lifestyle data) for tailoring the right therapeutic strategy for the right person at the right time, and/or to determine the predisposition to disease and/or to deliver timely and targeted prevention. The ability to select patients for treatment with targeted agents on the basis of specific molecular alterations within their cancer cells has led to novel drugs for molecularly selected patient populations, with a view to improving treatment outcome, while minimising side effects.

Partnership

Prof. Orla Sheils has been working as a founder member of the OncoNetwork Consortium in collaboration with ThermoFisher to develop and validate the CE-IVD Oncomine Solid Tumor DNA and Solid Tumour Fusion Transcript kits which enable highly accurate and reliable multiplexed sequencing of formalin-fixed, paraffin embedded (FFPE) tumour samples with the quality and performance needed for the clinical laboratory.

Approach

Current approaches to detect numerous mutations in a clinical sample run the risk of the tumor sample being consumed before an actionable variant is uncovered. However, next-generation sequencing (NGS) can assess a broad range of genes in a single test and the consortium designed and validated informative NGS panels based on the Ion Torrent AmpliSeq Technology. The resulting kits allow detection of various types of mutations using as little as 10ng of FFPE DNA. This allows laboratory clinicians to analyze samples that may contain partially degraded or limited tumor material, and generate reportable results from more samples than was previously possible. The panels were verified by leading clinicians from the OncoNetwork Consortium with a focus on colon and lung cancers. Disease driving DNA and RNA mutations and genetic rearrangements that cause fusion transcripts are an important and expanding class of actionable biomarkers in cancer. These new kits enable detection of these aberrations even from samples with low DNA/RNA quantity, quality or integrity, ultimately providing actionable information to a greater number of patients and beneficial insight that may help guide treatment.

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