Clinical evaluation of a CE-IVD liquid biopsy pan cancer genomic profiling test

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INTRODUCTION

- Essential to the successful delivery of personalized cancer therapy is non-invasive diagnostic tests that comprehensively characterize the genomic signatures occurring within individual tumours
- In this study, we report clinical evaluation of a circulating cell-free total nucleic acid (cfTNA)-based pan-cancer NGS (Next Generation Sequencing) genomic profiling liquid biopsy (LB) test that identified genomic signatures across solid organ cancers

METHODS

- 3630 samples belonging to 44 solid organ cancers from Asian and Caucasian origin were used in this clinical evaluation.

- Total cell-free total nucleic acid (cfTNA) was isolated from blood plasma. ~15-25 ng cfTNA underwent Pan Cancer Library preparation using Oncomine Pan-Cancer Cell-Free Assay (Thermofisher Scientific, Waltham, MA, USA) followed by template preparation and sequencing using Ion Proton / Gene Studio S5 semiconductor sequencer (Thermofisher Scientific, Waltham, MA, USA).

- Data was analysed with in-house proprietary bioinformatics pipeline to detect SNV, CNV and gene-fusions.

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RESULTS

- A total of 5960 SNVs were detected. The gene harbouring the highest number of SNVs was TP53 (38%), followed by KRAS (11%) and PIK3CA (10%).

- The highest number of SNVs were detected in Breast, Colorectal, Lung, Pancreas, Prostate.

SNV

CNV

- Majority of the CNVs were found in MYC, FGFR1, EPBB2, CDK4, CCND1, FGFR3 genes, mainly belonging to the Breast, Colorectal, Lung and Prostate cancer patients.

The graph above demonstrates the detection of CNV in the recurrently mutated genes across cancer types.

CONCLUSIONS

This proof-of-concept clinical evaluation of a liquid biopsy test successfully identified low frequency genomic variations via SNV, CNV, gene fusion signatures from cfTNA, across different solid organ cancers, belonging to varying ethnicity. This study demonstrates potential for liquid biopsy genomic profiling of solid organ cancers at diagnosis or for patient monitoring/therapy management, post treatment.