Tumour Mutational Burden using the TSO-500: How does it help in patient management

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The TSO-500 is a large panel of 523 genes plus 44 fusions that are associated with cancer development or treatment. The aim of Tumour Mutational Burden (TMB) testing is to reveal what actionable changes occur within a tumour and the number of mutations associated with a given tumour.

The TMB is a measure of the number of mutations per megabase, with more than 10 per megabase being deemed high TMB. The TMB score correlates highly with the likelihood of success in treating patients with immune checkpoint inhibitors (ICIs). Included in the assessment of TMB is microsatellite instability, which contributes to the overall TMB status.

The TSO-500 will detect some gene fusions, somatic variants, novel transcripts, single nucleotide variants (SNVs), insertion deletion variants (indels) and copy number variants. Together, this data provides a comprehensive view of the molecular events underpinning tumour growth that can reveal which targeted therapy is most likely to be effective in treatment of malignancy. Furthermore, the TSO-500 provides information about the variant allele frequency which can be used as a guide to determine if the patient is most likely to carry a germline change. When the variant allele frequency is ~50% confirmatory germline testing is requested.

Finally, TMB assessment can aid in the identification of cancers of unknown primary disease and provide limited information about environmental exposures that result in malignancy. How the TSO-500 is performed, and what requirements are needed to provide useful information from this assay will be presented.