⮕ PROSTATE CANCER

Singled out: single-cell genomics for diagnosis

A pilot study exploring the applicability of single-nucleus sequencing (SNS) to prostate cancer diagnosis has shown that single-cell genomics could improve tumour grading and staging at the biopsy stage. Furthermore, the small amount of cellular material required for this technique means that less-invasive sampling procedures could be used to harvest tissue.

Single-cell genomic features were assessed and compared with histopathological measures of malignancy in samples from 11 patients. Clonality was strongly and significantly associated with Gleason score ≥6 in both biopsy and radical prostatectomy samples. Clonal heterogeneity at diagnosis (the number of subclones plus the number of clones without subclones), the proportion of cores containing clonal cells, the proportion of cells judged to be clonal, the genomic complexity (the number of clonal features), and the clonal spread (the average proportion of cells in an area from the clone affecting the most areas) were considered relative to histopathological findings. The five genome-derived descriptors significantly correlated with Gleason score in the biopsy sample. Notably, correlation between the genomic descriptors and Gleason score improved on regrading of cancer based on surgical specimens. Heterogeneity was the best performing of all genomic and histopathological parameters on regrading, followed by the other genomic characteristics, which better correlated with the revised Gleason score than the conventional pathological parameters.

To assist evaluation of SNS in context, alongside anatomy and histopathology, the researchers developed a new algorithm based on copy number variation profiles for inferring the clonal structure of a sample. The investigators also introduce a single-cell-genomics viewer, which is an integrated, interactive platform for viewing complementary genomic, histopathological, and anatomical data.

These results show the utility of single-cell genomics for diagnosis of prostate cancer, which could aid in primary treatment decision making for men with this disease.

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