A dual-platform approach to DNA methylation profiling in blood and tissue for non-invasive diagnosis of urinary bladder cancer

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The present study evaluated DNA methylation as a non-invasive biomarker for urinary bladder cancer (UBC) by comparing blood and matched tumour tissues from 243 patients and 200 controls (110 matched tissues). Methylation of P16, CALCA, CCNA1, TMEM106A and AL021918.2 was profiled using whole-genome bisulphite sequencing (WGBS) and validated by methylation-specific PCR. In blood, the five-gene panel achieved 92.3% sensitivity and 88.1% specificity; in tissues, sensitivity was 100% with 92.7% specificity. High blood–tissue concordance supports blood-based methylation assays as viable, cost-effective alternatives to cystoscopy for early detection and monitoring of UBC.

Keywords: Biomarkers, epigenetics, methylation, sequencing, urothelial.

CpG islands of gene promoters, thereby regulating gene expression. Alterations in DNA methylation patterns, especially hypermethylation of tumour suppressor genes in promoter regions, have been extensively documented in various malignancies, including bladder cancer, indicating their potential role in tumourigenesis and progression[4,5]. Epigenetic modifications offer advantages as biomarkers due to their stability, reversibility, and detectability in samples such as blood and tissue. Genes including P16, CALCA and CCNA1 have been implicated in bladder cancer through altered methylation status, correlating strongly with disease severity and patient outcomes[6,7]. Despite many findings, significant gaps remain in the comparative analysis of methylation profiles between blood and matched tumour tissues, particularly when using advanced sequencing technologies.

Whole-genome bisulfite sequencing (WGBS) provides complete methylation profiling at single-base resolution, enabling the identification and quantification of methyla-