Differential frequencies in TERT promoter and \textit{FGFR3} mutations between Chinese and western patients with urothelial bladder cancer

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The TERT promoter and \textit{FGFR3} gene mutations are two most common genetic events in urothelial bladder cancer (UBC) and these mutation assays in patient urine have been shown promising biomarkers for UBC diagnosis and surveillance based on analyses of UBC patients from western countries. However, we observed TERT promoter and \textit{FGFR3} mutations in 87 of 182 (47.8\%) and 7 of 102 (6.7\%) Han Chinese patients with UBC respectively, which are lower than those in UBC patients from western countries. In 46 urine samples from patients with TERT-promoter mutation-carrying tumours, the mutant promoter was detected in 24 (52\%) prior to operation and disappeared in 80\% urine samples one week post-operation. TERT mRNA was detected in urine derived from 46 of 49 (94\%) of patients determined before operation independently of TERT promoter mutations. Collectively, \textit{FGFR3} mutations occur at a very low rate in Han Chinese UBC patients compared with western patients indicating a different aetiology between Chinese and western UBCs. Han Chinese UBC patients have a relatively lower TERT promoter mutation frequency and simultaneous detection of both mutant TERT promoter and TERT mRNA improves sensitivity and specificity of urine-based diagnosis.

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