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Germline prognostic markers for urinary bladder cancer

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ABSTRACT

The majority of urinary bladder cancer (UBC) cases are diagnosed as non-muscle invasive malignancies, having a favourable prognosis in terms of overall 5-year survival. However, non-muscle invasive bladder cancer (NMIBC) cases show high recurrence and progression rates and inconsistencies within the NMIBC risk group, resulting in a substantial burden on patients and health systems.

The evidence for genetic risk factors having a role in NMIBC susceptibility and prognosis make NMIBC a good candidate for personalised medicine approaches; however, multiple genome-wide association studies (GWAS) have primarily focused on NMIBC risk alone, even though investigating prognostic factors would arguably yield more benefit.

It is recognised that genetic variation contributes to complex traits in the form of multiple effects of low-penetrance, as well as interacting not only with each other, but with various environmental factors as well, resulting in a complex problem to resolve.

In a setting of a bladder cancer patient cohort, our project aims to identify genetic variants of genome-wide significance that might be associated with certain NMIBC characteristics at diagnosis, and potential gene-environment interaction effects with smoking. Furthermore, we aim to address the importance of replication in genetic association studies by utilising a resource of UK Biobank, whilst introducing a novel approach for identifying prognostic events from routinely collected data.

In conclusion, the current thesis provides additional evidence to the field of bladder cancer genetics and suggests further research topics of interest that could lead to optimising NMIBC patients' care.