University of BRISTOL

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NHS Foundation Trust

headandneck 5000

Genome Wide Association Study of Head and Neck Cancer progression

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Scientific Outline

Summary

There are over 0.5 million head and neck cancers (HNC), here considered to be oral and pharyngeal cancers, a year and incidence is increasing. Furthermore, only 40–50% of patients with HNC will survive beyond 5 years and those that do often have facial disfigurement, loss of function and pain as result of surgery or radiotherapy. The well-established risk factors for developing HNC are tobacco use, alcohol consumption and oral human papilloma virus (HPV) infection, yet factors that affect progression and survival are less well understood.

Understanding the factors that affect the prognosis of HNC patients is crucial to improving survival and quality of life. A recent Genome-wide association study (GWAS) of HNC risk implicated genetic variants in genes closely related to alcohol (rs1229984, *ADH1B)* and HPV infection (rs3828805, *HLA-DQB1*), both known risk factors. A GWAS of HNC progression may identify genetic variants in genes involved in cancer progression. This would increase our understanding of these cancers and provide clues for therapeutic targeting but larger sample sizes are required to provide adequate power for this study.

At present, 1,079 participants have been genotyped in HN5000. Further genotyping of samples will improve power for this GWAS and a range of genetic epidemiological methods including Mendelian randomization (MR) and recall-by-genotype (RbG).