

headandneck 5000

Genome Wide Association Study of oral and oropharyngeal cancer using the oncochip

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Scientific outline

Head and neck cancers are neoplasms of the oral cavity, pharynx and larynx. The majority are squamous cell carcinoma. The main risk factors are tobacco and alcohol use. Another risk factor, predominantly restricted to the oropharynx and base of the tongue, is exposure to human papilloma virus type 16. Only one GWAS has been published. This study, led by IARC included Illumina GWAS 317k data on 2000 cases and 3,500 controls from two multicenter European studies and incorporated an additional 13 head and neck cancer studies in the replication phase comprising a further 6500 cases and 7900 controls. Multiple susceptibility loci were identified and confirmed at positions 4q21, 4q23 and 12q24. Focused IARC led candidate gene studies have also confirmed additional genetic variants with genome-wide significance at positions 4q23, 15q25 and 22q12.

The Genetic Association and Mechanisms in Oncology consortium has recently designed a customized Illumina genotyping array, the 'Oncochip' of approximately 565,000 variants. The Oncochip custom array content includes a GWAS backbone of approximately 260,660 variants, a common content covering approximately 39,000 variants (comprising variants related to a wide range of phenotypes and fine-mapping of common loci that are associated with more than one cancer), and site specific variants from five major cancers

The studies contributing to this genome wide association study and the numbers they are contributing are: Chance (1,463), Gencapo (1,064), IARC Arcage (1,464), IARC_CE (202), IARC_EPIC (1,676), IARC_LA (1,057), IARC_ORC (343), Maastricht (555), Pittsburgh (1,606), Rome (180), Toronto (1,975), Head and neck 5000 (1,034).