

Allelic imbalances and chromosome 9p microdeletion demonstrated in cutaneous squamous cell carcinomas using single nucleotide polymorphism microarray analysis

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Cutaneous squamous cell carcinomas (SCC) are the second most commonly diagnosed cancers in white-skinned populations yet the genetic mechanisms involved in SCC tumorigenesis remain poorly understood. We have employed single nucleotide polymorphism (SNP) microarray analysis to examine genome-wide allelic imbalance in 16 primary and 2 metastatic SCC against corresponding non-tumour samples. The most frequent genetic change was loss of heterozygosity (LOH) on chromosome 9p, observed in 13 of 16 primary SCC. Other recurrent events include LOH on chromosomes 3p, 8p and 13 (observed in 10, 8 and 7 primary tumours respectively) as well as allelic gain on chromosomes 3q and 8q (each in 6 primary SCC). Copy number analysis demonstrated that most LOH was due to deletion. However, 3 of 7 samples with chromosome 13 LOH and 3 of 13 samples with 9p LOH did not show copy number loss, implying that LOH was due to somatic recombination leading to acquired uniparental disomy (UPD), an event not previously demonstrated in SCC. As well as identifying recurrent patterns of gross chromosomal changes, SNP microarray analysis revealed a microdeletion on chromosome 9p23 within the protein tyrosine phosphatase receptor type D (*PTPRD*) locus. This event was observed in 3 primary SCC, 2 of which had demonstrated metastatic potential. Our data validates SNP microarray analysis as an accurate and sensitive method for evaluating the global genetic changes underlying SCC tumorigenesis.