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Analysis of the IDH1 codon 132 mutation in brain tumors.

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A recent study reported on mutations in the active site of the isocitrate dehydrogenase (IDH1) gene in 12% of glioblastomas. All mutations detected resulted in an amino acid exchange in position 132. We analyzed the genomic region spanning wild type R132 of IDH1 by direct sequencing in 685 brain tumors including 41 pilocytic astrocytomas, 12 subependymal giant cell astrocytomas, 7 pleomorphic xanthoastrocytomas, 93 diffuse astrocytomas, 120 adult glioblastomas, 14 pediatric glioblastomas, 105 oligodendrogliomas, 83 oligoastrocytomas, 31 ependymomas, 58 medulloblastomas, 9 supratentorial primitive neuroectodermal tumors, 17 schwannomas, 72 meningiomas and 23 pituitary adenomas. A total of 221 somatic IDH1 mutations were detected and the highest frequencies occurred in diffuse astrocytomas (68%), oligodendrogliomas (69%), oligoastrocytomas (78%) and secondary glioblastomas (88%). Primary glioblastomas and other entities were characterized by a low frequency or absence of mutations in amino acid position 132 of IDH1. The very high frequency of IDH1 mutations in WHO grade II astrocytic and oligodendroglial gliomas suggests a role in early tumor development.

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