Detection of IDH1 mutation in the plasma of patients with glioma.

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**Abstract**

**OBJECTIVE:** The IDH1(R132H) mutation is both a strong prognostic predictor and a diagnostic hallmark of gliomas and therefore has major clinical relevance. Here, we developed a new technique to detect the IDH1(R132H) mutation in the plasma of patients with glioma.

**METHODS:** Small-size DNA (150-250 base pairs) was extracted from the plasma of 31 controls and 80 patients with glioma with known IDH1(R132H) status and correlated with MRI data. The IDH1(R132H) mutation was detected by a combination of coamplification at lower denaturation temperature and digital PCR.

**RESULTS:** The small size DNA concentration was 1.2 ng/mL (range 0.1-6.6) in controls vs 1.2 ng/mL (range 0.1-50.3) in patients with glioma (p = not significant) and 0.9 ng/mL (0.0-3.0) in low-grade gliomas vs 1.5 ng/mL in high-grade gliomas (p < 0.01). The small size DNA concentration correlated with enhancing tumor volume (1.6 ng/mL [0.4-24.9] when <10 cm(3) and 14.0 ng/mL [0.6-50.3] when ≥10 cm(3)). The IDH1(R132H) mutation was detected in 15 out of 25 plasma DNA mixtures (60%) from patients with mutated tumors and in none of the 14 patients with a nonmutated tumor. The sensitivity increased with enhancing tumor volume (3/9 in nonenhancing tumors, 6/10 for enhancing volume <10 cm(3), and 6/6 for enhancing volume ≥10 cm(3)).

**CONCLUSION:** With a specificity of 100% and a sensitivity related to the tumor volume and contrast enhancement, IDH1(R132H) identification has a valuable diagnostic accuracy in patients not amenable to biopsy.

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