ABSTRACT

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Coexisting lipomatous meningioma and glioblastoma in Cowden syndrome: A unique tumor association.

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Cowden syndrome (CS) is a rare hereditary hamartoma-cancer disorder related to germline mutations in the tumor suppressor phosphatase and tensin homolog (PTEN) gene. Association of CS with intracranial tumors, apart from Lhermitte-Duclos disease (LDD), is not well recognized. We present an exceptional instance of concomitant meningioma and glioblastoma in CS, the first case ever reported. Following a new-onset seizure, a 62-year-old male harboring the PTEN gene germline mutation c.334C > G was diagnosed with multiple brain tumors, which were erroneously thought to correspond to metastases. Because no primary cancer was found, an operation was proposed for histopathological diagnosis. Examination of surgical specimens obtained from the two lesions removed, one extra-axial and the other intracerebral, demonstrated a metaplastic meningioma with a lipomatous appearance and an isocitrate dehydrogenase wild-type glioblastoma, respectively. Loss of the PTEN gene expression was demonstrated immunohistochemically in both lesions, a finding that supports their relation to CS. A thorough literature review revealed only 25 additional CS patients with intracranial tumors other than LDD. All of them corresponded to primary lesions, with meningiomas accounting for 76% of the cases (19 patients), followed by pituitary tumors (three cases) and glioblastomas (two patients from the same family). Our report and literature review highlight the association between CS and primary brain tumors rather than metastasis. For judicious management of a CS patient with multiple intracranial tumors, different primary brain pathological entities should also be suspected first before considering metastasis. Close neurological monitoring and brain magnetic resonance imaging are advocated as part of the cancer screening in CS patients, particularly in cases with a family history of intracranial tumors.

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