ABSTRACT

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Cerebellar liponeurocytoma: clinical, histopathological and molecular features of a series of three cases, including one recurrent tumor.

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Cerebellar liponeurocytoma (CL) is an unusual tumor, histologically composed of a mixture of small to medium-sized, rounded neurocytic cells and a variable lipomatous component. Although CL was originally considered as a subtype of medulloblastoma, subsequent molecular studies demonstrated that this tumor was a distinct entity, exhibiting the tumor protein p53 gene (TP53) missense mutations in 20% of cases, chromosome 17 deletion, and the absence of mutations in the adenomatous polyposis coli gene (APC), the protein patched homolog gene (PTCH), the kinase insert domain receptor gene (KDR), and the β-catenin gene (CTNNB). Apart from these molecular features, little is known about the pathogenesis and the genetic landscape of CL to date. In order to characterize the mutational landscape of CL and identify alterations that are driving tumorigenesis, we report a series of three cases, including one recurrent tumor, analysed by next-generation sequencing (NGS), which identified a total of 22 variants, of which four were missense mutations, nine were synonymous variants, and nine were located on intronic regions. In particular, DNA sequencing identified missense mutations in APC, KDR, and TP53 that could be implicated in promoting tumor progression and angiogenesis of CL. Furthermore, the NGS analysis revealed that recurrent CL did not have additional genetic changes compared with the primary tumor. Moreover, the high frequencies of detected mutations suggested that the identified alterations are germline variants. Indeed, an additional NGS on the genomic DNA obtained from one of the three patients confirmed the presence of the variants in the germline DNA. In conclusion, the obtained data support the hypothesis that CL is a distinct pathological entity that does not show specific somatic alterations driving tumorigenesis.

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