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Case reports

Cytochrome c oxidase assembly factor COX14 is a protein that in humans is encoded by the COX14 gene. This gene encodes a small single-pass transmembrane protein that localizes to mitochondria. This protein may play a role in coordinating the early steps of cytochrome c oxidase (COX; also known as complex IV) subunit assembly and, in particular, the synthesis and assembly of the COX I subunit of the holoenzyme. Mutations in this gene have been associated with mitochondrial complex IV deficiency. Alternative splicing results in multiple transcript variants.

A 43-year-old man who presented with an intracranial mesenchymal tumor. Histopathological examination showed a wide spectrum of peculiar morphological features and a non-specific immunohistochemical profile. Whole transcriptome sequencing revealed the presence of a novel genetic rearrangement involving COX14 and PTEN genes, which has never been reported before in any other neoplasm. The tumor did not cluster in any defined methylation class of the brain tumor classifier, but resulted in a calibrated score of 0.89 for the methylation class "Sarcoma, MPNST-like", when analyzed by the sarcoma classifier. Our study is the first to report about this tumor with unique pathological and molecular features, characterized by a novel rearrangement between COX14 and PTEN genes. Other studies are necessary in order to define it as a new entity or as a novel rearrangement involving recently described and incompletely characterized CNS mesenchymal tumors ¹⁾.

1)

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