

Double homeobox, 4 also known as DUX4 is a protein that in humans is encoded by the DUX4 gene. Its misexpression is the cause of facioscapulohumeral muscular dystrophy

CIC-rearranged [sarcoma](#) is a [high-grade sarcoma](#), most often harboring [CIC:: DUX4](#) fusion, and is characterized by a distinct round cell histology, co-expression of [ETV4](#) and [WT1](#), and a specific [DNA methylation](#) class. Satomi et al. reported a [brain tumor](#) with [ATXN1:: DUX4](#) that had an indistinguishable phenotype and DNA methylation profile from [CIC-rearranged sarcoma](#). A 40-year-old man presented with a 5 cm hemorrhagic mass in the right [frontal lobe](#) of the cerebrum. The tumor was resected and histologically showed a dense proliferation of relatively monomorphic round cells with multifocal [myxoid](#) changes. Immunohistochemically, the tumor was diffusely positive for ETV4, WT1, and DUX4. Through classic histomorphology and immune profile, the tumor was provisionally diagnosed as CIC-rearranged sarcoma. However, no CIC fusions or [mutations](#) were identified using CIC break-apart [fluorescence in situ hybridization](#) (FISH) or FoundationOne CDx. Despite multiple surgeries and adjuvant [chemoradiation](#) therapy, the patient succumbed 16 months after presentation. RNA exome sequencing detected an in-frame intraexonic ATXN1 (exon 9):: DUX4 ([exon 1](#)) fusion, which was validated by reverse transcription-polymerase chain reaction and ATXN1 FISH assay. Upon DNA methylation analysis, the tumor matched with CIC-rearranged sarcoma both by the Deutsche Krebsforschungszentrum classifier and t-distributed stochastic neighbor embedding. Along with a recent report of a similar [pediatric brain tumor](#), the present case suggests that ATXN1::DUX4 is a recurrent alternative molecular event in the sarcoma type that is presently defined by CIC rearrangement, which prompts an expansion of the tumor concept ¹⁾.

¹⁾

Satomi K, Ohno M, Kubo T, Honda-Kitahara M, Matsushita Y, Ichimura K, Narita Y, Ichikawa H, Yoshida A. Central nervous system sarcoma with ATXN1::DUX4 fusion expands the concept of CIC-rearranged sarcoma. *Genes Chromosomes Cancer*. 2022 Jun 17. doi: 10.1002/gcc.23080. Epub ahead of print. PMID: 35715887.

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