Virchows Arch. 2023 May 22. doi: 10.1007/s00428-023-03560-3. Online ahead of print.

Case report of a pediatric medulloblastoma with concurrent MYC and MYCN subclonal amplification in distinct populations of neoplastic cells

Simone Minasi¹, Francesca Gianno^{# 2}, Lavinia Bargiacchi², Valeria Barresi³, Evelina Miele⁴, Manila Antonelli², Francesca Romana Buttarelli²

Affiliations PMID: 37212894 DOI: 10.1007/s00428-023-03560-3

Abstract

Medulloblastomas (MDBs) are classified into molecular groups showing peculiar immunohistochemical and genetic features and distinct DNA methylation profile. Group 3 and group 4 MDBs have the worst prognosis; the former is treated with high-risk protocols and features MYC amplification, whereas the latter receives standard-risk protocols and harbors MYCN amplification. Herein, we report a unique case of MDB showing histological and immunohistochemical features consistent with non-SHH/non-WNT classic MDB, with both MYCN (30% of tumor cells) and MYC (5-10% tumor cells) amplification in distinct subclones of neoplastic cells at fluorescence in situ hybridization (FISH), characterized by specific patterns. In spite of MYC amplification in only a small percentage of tumor cells, this case had DNA methylation profile consistent with group 3, emphasizing the importance to test both MYC and MYCN amplifications at a single cell level using highly sensitive methods, such as FISH, for diagnostic and therapeutic purposes.

Keywords: FISH; MYC; MYCN; Medulloblastoma; Methylation profile.

© 2023. The Author(s), under exclusive licence to Springer-Verlag GmbH Germany, part of Springer Nature.