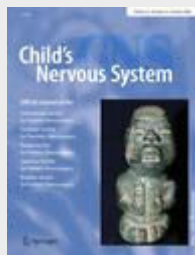



Journal Article



Fatal glioblastoma multiforme in a patient with neurofibromatosis type I: the dilemma of systematic medical follow-up

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Abstract

Introduction Neurofibromatosis type I (NF1) is one of the most prevalent genetic diseases of the nervous system. Although the majority of NF1 patients are only mildly affected, the risk of developing malignancies is significantly increased in this population. **Case report** Here, we present a 9-year-old girl with clinical stigmata of NF1 and a rapidly evolving glioblastoma multiforme. Molecular genetic analysis uncovered a novel missense mutation in Exon 32 of the *NF1* gene [c.6032C>A(p.Ala2011Glu)]. **Discussion** The girl's death 3 days after diagnosis of the brain tumor exemplifies that NF1 still is a life-threatening disease despite its generally benign course in most patients. However, it remains questionable if a fatal course as reported here can be prevented by routine MRI screening.

Keywords Children - Follow-up MRI - Glioblastoma multiforme - Neurofibromatosis type I - Novel *NF1* mutation



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