

within epilepsy syndromes. For GEFS+, the *SCN1A* gene mutation penetrance is approximately 70%, but phenotypic severity may also be affected by extrinsic factors [6,7]. Mutations in the pore area of the sodium channel are associated with the most disabling epilepsies [5]. Increased disease severity is linked with spontaneous mutation, whereas inherited cases are more likely to have milder phenotypes [5]. This probably occurs because people with severe epilepsy are less likely to reproduce.

5. Conclusion

This case report describes a novel inherited *SCN1A* gene mutation that caused encephalopathic epilepsy in a child but only benign epilepsy in the father. Whether specific localization of these mutations and/or variable expressivity confers seizure severity and phenotype is not clear. Modifying alleles may explain why identical mutations sometimes lead to vastly different phenotypes. Thus far, no specific mutation has been correlated to a specific phenotype. This novel mutation adds to a growing list of mutations, which may eventually be used for effective therapeutic development and prognostic purposes.

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Conflict of interest

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Low grade astrocytoma causing dural and calvarial destruction



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ABSTRACT

This case report describes destruction of overlying dura and calvaria by a low grade glioma in the absence of prior surgery or radiation. Bone and dural involving is known to occur with some malignant tumors, but due to low grade glioma is very rare.

The initial radiologic examinations demonstrated a heterogeneous mass in the right parietal region with both extra- and intra-axial components. No inward displacement of the adjacent dura was observed. Initial consideration for extra-axial lesions includes metastatic lesions, lymphoma, or an aggressive meningioma. The pathologic findings demonstrated a glial cell origin.

To our knowledge, destruction of the dura and calvaria from a low-grade glioma, without prior surgery or radiation, has not been well documented previously. Calvarial destruction with associated intra-axial lesions on imaging may prompt the diagnosis of extra-axial tumors such as aggressive meningiomas, metastasis and lymphoma.

We report an unusual case of parietal low-grade glioma with destruction of the dura and calvaria in the absence of prior surgery or radiation. The erosion probably is due to pressure atrophy of the dura and inner table and thinning of the diploe.

The mechanism of skull erosion in these superficial gliomas relates to their chronic mass effect (8). The mass displaces the CSF, which normally cushions and diffuses brain pulsations over a wide area. Once the CSF space is effaced, the brain may directly transmit these pulsations to the inner table. Over time, this localized elevated pressure may erode the cortical bone of the inner table.

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1. Case report

1.1. History, examination

A 64-year-old male with a history of congestive heart failure, hypertension and stroke presented to the neurosurgical service

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with increasing headaches, clumsiness and weakness of the right hand and leg lasting a few days. On physical examination, he had right hemianopsia with right hemiparesis. Palpation of the scalp revealed a firm tissue mass, with bony consistence, over the left parietal tuber. Further work-up included chest radiography, without evidence of malignancy and a prostate specific antigen level within normal limits.

1.2. Imaging

First, a nonenhanced CT study was performed (Fig. 1). The images showed an ill-defined, heterogeneous left parietal mass causing destruction of the calvaria (Fig. 2).

Intra- and extra-axial localization was difficult secondary to lack of intravenous contrast enhancement. Overlying calvarial

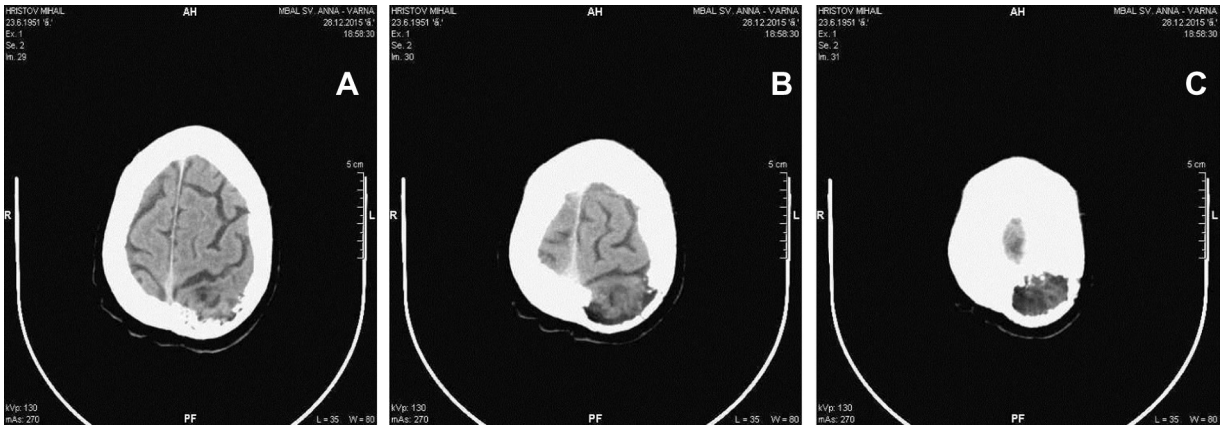


Fig. 1. (A–C) Nonenhanced scans obtained at slightly different heights show a heterogeneous mass in the left parietal region with erosion of the calvaria.

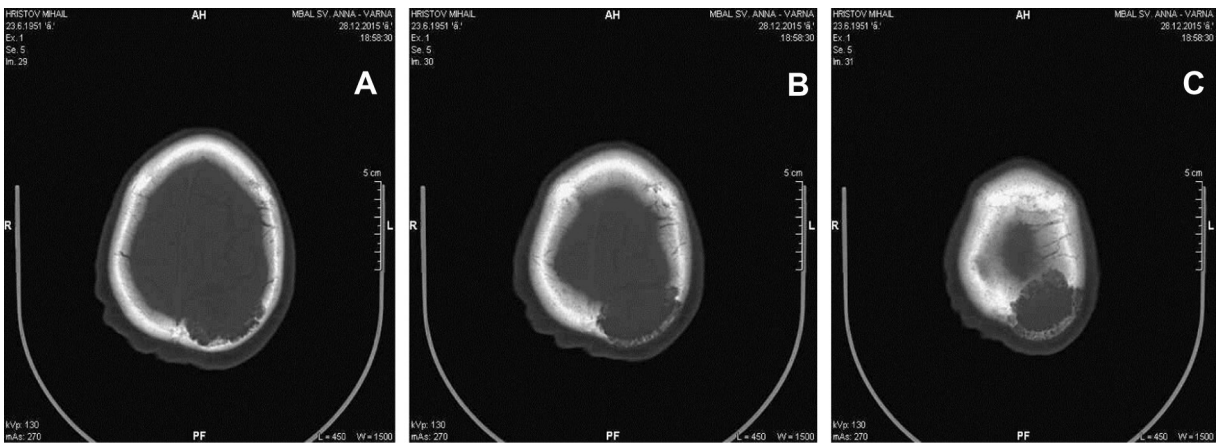


Fig. 2. (A–C) Bone window settings from the same nonenhanced CT study as in Fig. 1 shows calvarial destruction.

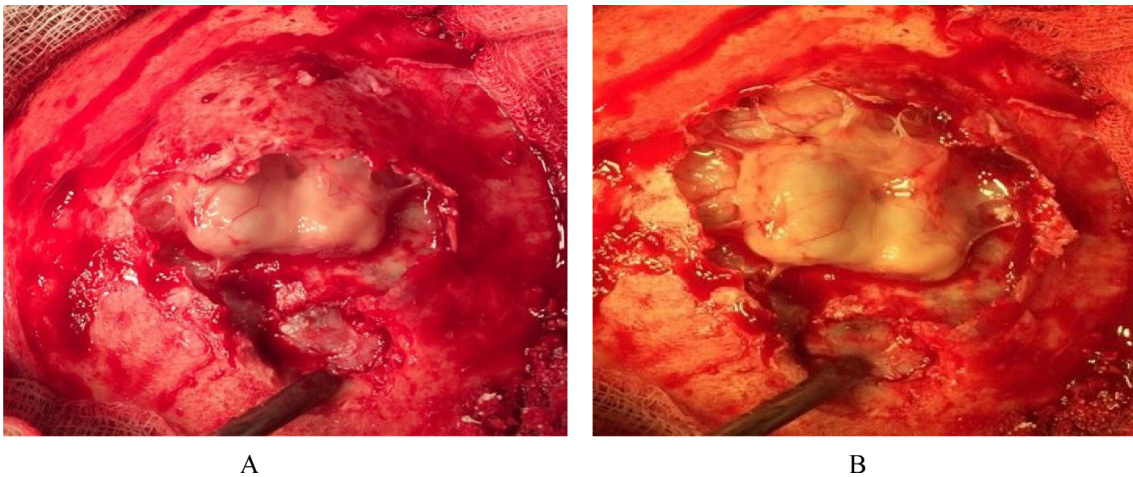


Fig. 3. (A) Removing of the external table. (B) The tumor mass in the diploe.

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