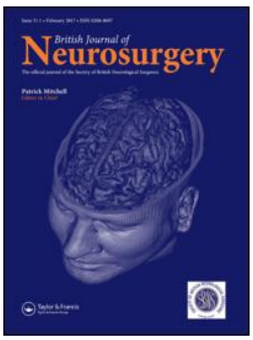

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SHORT REPORT

Fahr's disease associated with anaplastic ependymoma: a case report and review of the literature

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ABSTRACT

Fahr's disease, also known as familial idiopathic basal ganglia calcification or bilateral strio-pallido-dentate calcinosis, is a rare entity characterized by abnormal vascular calcium depositionin the thalamus, basal ganglia, cerebral cortex and the dentate nuclei of the cerebellum. Intracranial ependymomas comprise approximately 2% to 9% of all neuroepithelial tumors. It is reported that supratentorial ependymoma constitute 30% to 50% of all intracranial ependymal tumors. Among supratentorial ependymomas, approximately 50% of them are located extraventricular and demonstrate no relationship with the ventricularsystem.The association of brain tumor with Fahr's disease is a rare entity and has been reported several times before. Whereas, to best our knowledge, the association of Fahr's disease and supratentorial anaplastic ependymoma is described in the present study for the first time.

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Fahr's disease; brain tumor; supratentorial; ependymoma

Introduction

Fahr's disease is a rare entity characterized by abnormal vascular calcium depositionin the thalamus, basal ganglia, cerebral cortex and the dentate nuclei of the cerebellum.[1,2] The etiology of Fahr's disease remains largely unknown.[3] Clinical findings are often in the form of dysfunction of the regions where calcification is found.[4]

Intracranial ependymomas comprise approximately 2–9% of all neuroepithelial tumors and 8–10% of brain tumors in children younger than 20 years.[5,6] It is reported that supratentorial ependymoma constitute 30% to 50% of all intracranial ependymal tumors.[5,7] Among supratentorial ependymomas, approximately 50% of them are located extraventricular and demonstrate no relationship with the ventricular system.[6] World Health Organization (WHO) classify ependymal tumors such as Grade I (subependymoma and myxopapillary ependymoma), Grade II (ependymoma), and Grade III (anaplastic ependymoma).[5]

The association of brain tumor with Fahr's disease is a rare entity and has been reported several times before. Whereas, to best our knowledge, the association of Fahr's disease and supratentorial anaplastic ependymoma is described in the present study for the first time.

Case report

A 33-year-old female patient was admitted to our clinic with severe headache. Physical examination revealed no abnormality except blurred consciousness.There was no finding in the patient anamnesis except congenital deafness. Laboratory tests, which ruled out the possibilities of endocrine abnormalities, infection and toxification as the causes of calcification,were normal.Computed tomography (CT) showed calcification in bilateral thalamus, basal gangia, cerebral cortex and cerebellar dentate nucleiconsistent with Fahr's disease (Figure 1). Additionally, hypodense lesion areas with edema were observed in the right frontotemporal lobe.T2-weighted magnetic resonance imaging (MRI) revealed two adjacent cystic lesions with white matter edema in the right frontotemporal region (Figure 2). Signal change indicating basal ganglia calcification and anteriorly located cyst calcification was observed on T1-weighted imaging

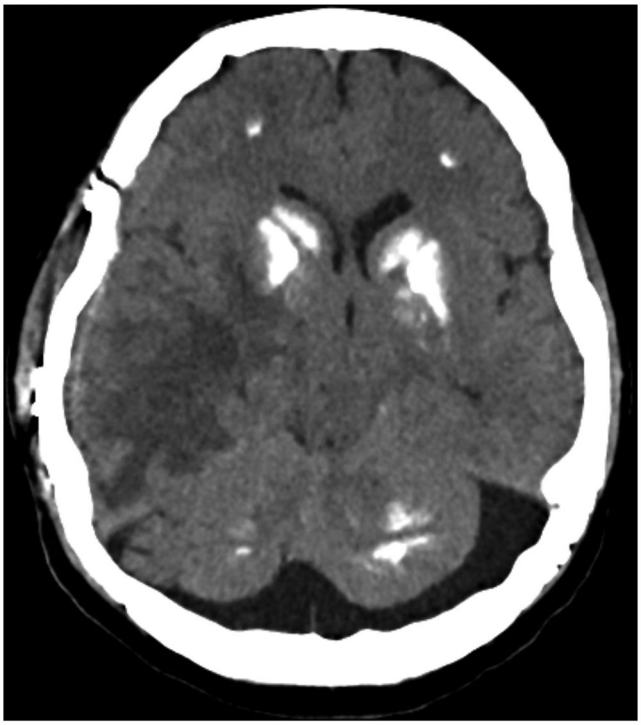

Figure 1. Axial non-enhanced computed tomography shows calcification in bilateral basal gangia, subcortical area and cerebellar dentate nuclei consistent with Fahr's disease.

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and susceptibility weighted imaging (SWI) (Figure 3(A,B)). Diffusion-weighted imaging did not show any restriction. On post-contrast T1-weighted imaging, marked peripheral ring-like enhancement was observed in cystic lesions (Figure 4(A)). MR perfusion examination conducted for the area of enhancement revealed increased relative cerebral blood volume (rCBV) values indicating increased vascularity (Resim 4B).MR perfusion examination conducted for the basal ganglia revealed no perfusion difference between calcified and noncalcified area in the thalamus (Resim 5). Conventional and advanced imaging findings were consistent with high grade tumor. The pathology result of the operated lesions was reported as anaplastic ependymoma (WHO Grade III) (Figure 5).

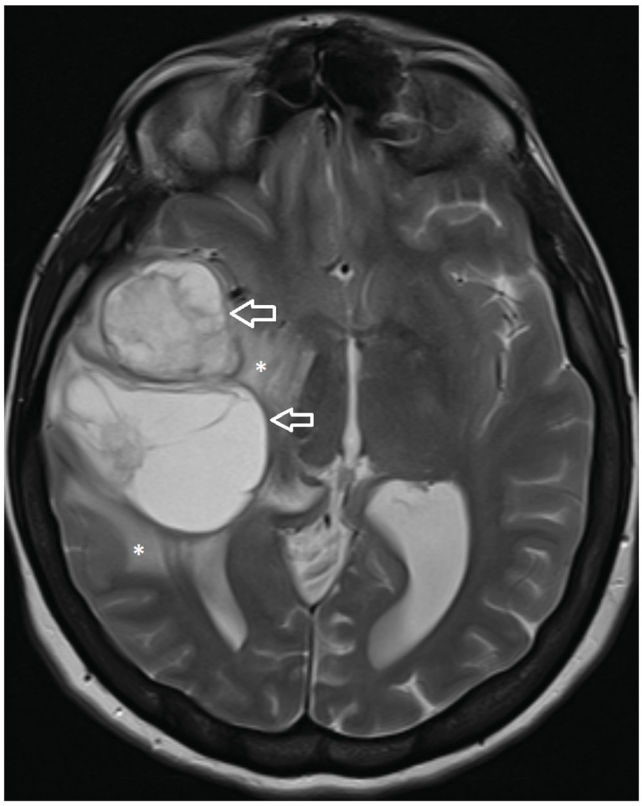

Figure 2. Axial T2-weighted magnetic resonance imagingshows two adjacent cystic lesions (arrows) with white matter edema (stars) in the right frontotemporal region.

Discussion

Fahr's disease is a rare disease, with genetic mutation and autosomal dominant inheritance in most cases.[8] The etiology of Fahr's disease is unknown but circulatory disturbances have been suspected.[9,10] Histopathological studies revealed concomitant hypertrophy and hyperplasia of astrocytes with calcification.[11] The diagnostic criteria are mainly based on exclusion of other diseases.[4,12] The symptoms usually occur between the fourth and sixth decade of life, but they have been also reported in children as well as in young adults.[11,13] There is no specific treatment of Fahr's disease, except for the calcium antagonist treatments considered theoretically.[14]

Supratentorial ependymomas (SEs), typically located within the brain parenchyma, are thought to originate from the remaining ependymal cells that have been retained within the brain parenchyma after embryonic development.[15] The WHO classification system categorizes ependymomas into either Grade I, II, or III. Anaplastic ependymomas, also known as Grade III ependymomas have high proliferation and infiltration capacity, and poor prognosis.[16] Pajtler *et al.* reported that supratentorial tumors were more commonly anaplastic in comparison with their infratentorial counterparts.[17] Peritumoral edema was reported as to be always minimal by Van Gompel *et al.*,[18] whereas Wang *et al.* reported it as a common situation.[5] In our case, two adjacent cystic masses with extraventricular location accompanied by pronounced edema was observed.

In the literature, the association of Fahr's disease and brain tumor is not frequently defined. In a recent study by Lin *et al.* the fifth case of brain tumor associated with Fahr's disease was

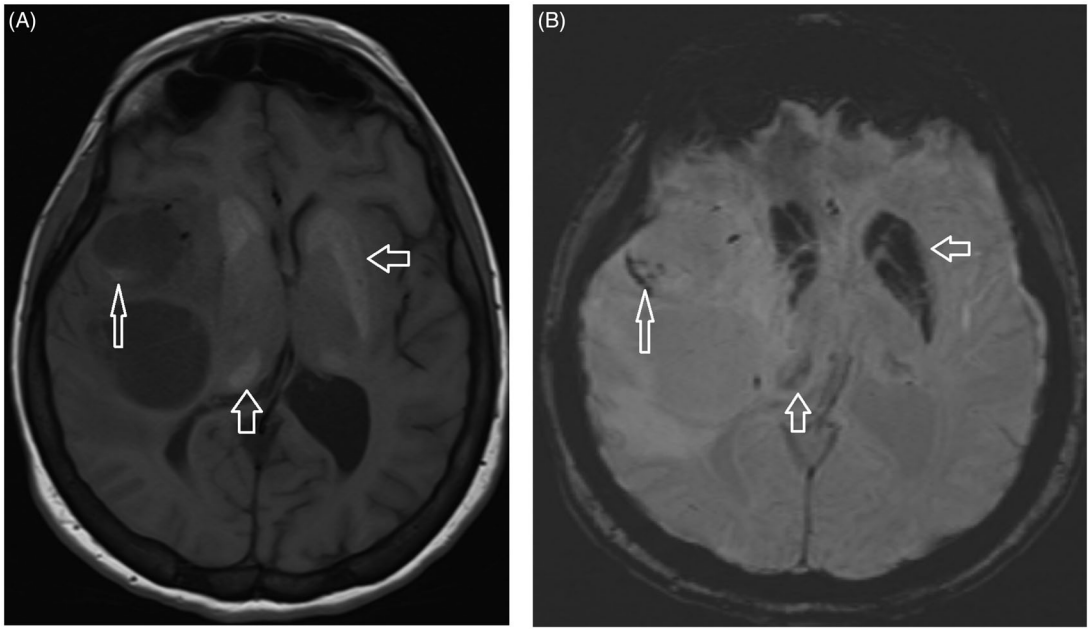

Figure 3. Signal change indicating basal ganglia calcification (thick arrows) and anteriorly located cyst calcification (thin arrows) was observed on (A) T1-weighted imaging and (B) susceptibility weighted imaging (SWI).

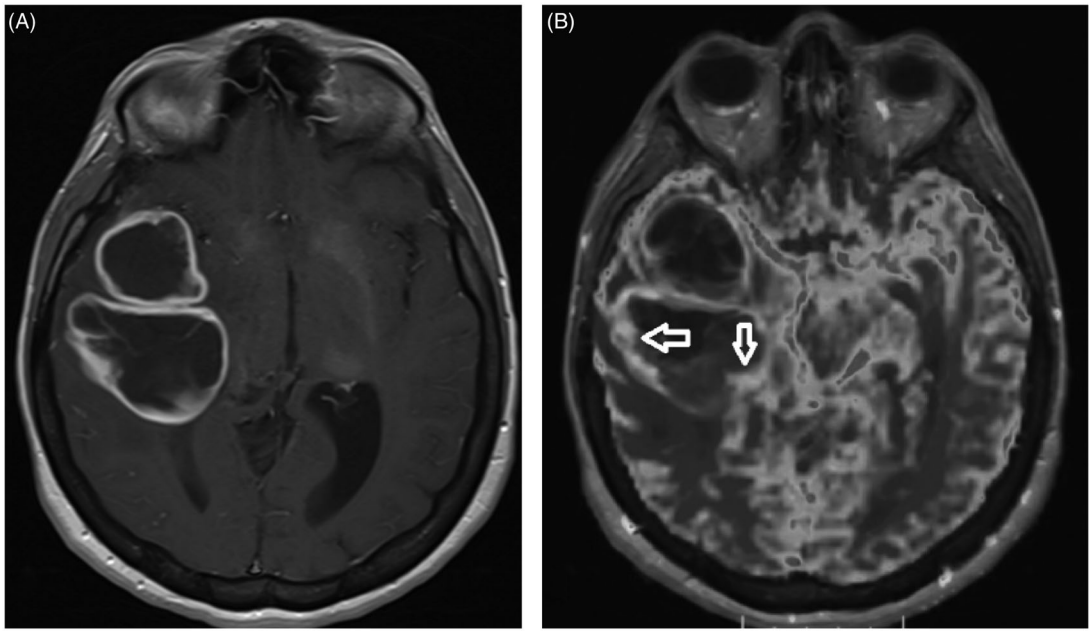

Figure 4. (A) Axial post-contrast T1-weighted imaging shows peripheral ring-like enhancement in cystic lesions, (B) color-coded perfusion map (overlaid on the corresponding post-contrast T1-weighted image)conducted for the area of enhancement shows increasedrelative cerebral blood volume (rCBV) values indicating increased vascularity (arrows).

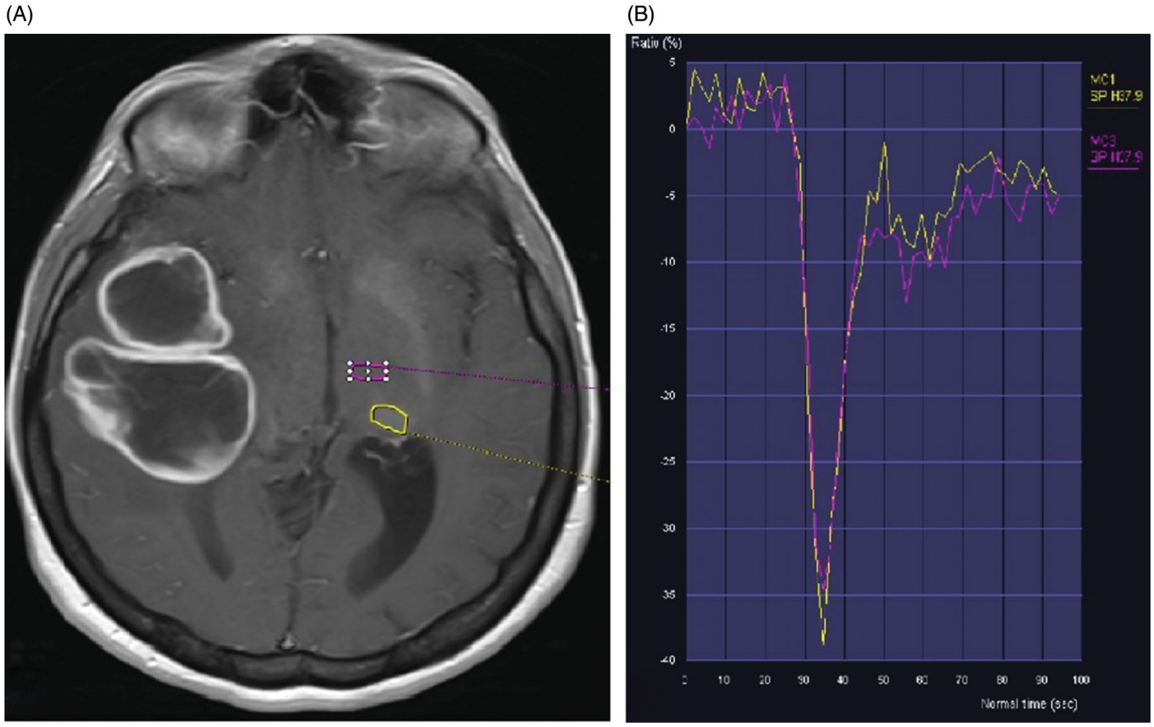

Figure 5. (A) Axial post-contrast T1-weighted imagingand (B) signal intensity time curvesshows no perfusion difference between calcified (yellow) and noncalcified area (purple) in the thalamus.

presented and all were reported as low-grade glioma.[3] Also, Jaworski *et al.* described coexistence of Fahr's syndrome with low-grade brain tumors such as astrocytoma and pineal body gangliocytoma.[19] Histopathology studies in patients with Fahr's disease have noted extensive calcification accompanied by hypertrophy and hyperplasia of astrocytes. Furthermore, several genetic alterations such as platelet-derived growth factor β polypeptide gene, have also been demonstrated to be involved in the development of glioma.[3] For all that, whether the occurrence of astrocytoma could be linked etiologically to long-standing astroglial proliferation remains speculative. According to our knowledge, the study showing the association of anaplastic ependymoma and Fahr's disease in the literature has not been previously presented. However, we do not have sufficient data neither in the literature to prove that the cause of the tumor is Fahr's disease. There are publications stating that the disease is associated with astrocyte proliferation, but we could not reach data to show its relationship with ependymia.

In conclusion,the diagnosis of brain tumor should be taken into account when patients with Fahr's disease develop emerging headaches or blurred consciousness.The occurrence of two adjacent cystic masses may predispose anywhere else to the formation of another cystic tumor, which warrants the close monitoring and follow-up of these patients.

Disclosure statement

No potential conflict of interest was reported by the author(s).

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