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Letter to the Editor

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# A Patient With Fahr's Disease Who Presented Prominent Visuospatial Dysfunction

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Fahr's disease (FD) is a rare neurological disorder characterized by symmetrical calcium deposition in various parts of the brain including the basal ganglia.[1] Movement disorders including Parkinsonism are the most common symptoms of FD, although convulsions and cognitive impairment are also known to be present.[2] We experienced an atypical FD with visuospatial dysfunction as the initial symptom.

A 64-year-old male patient with gradual worsening of difficulty in finding his usual path for about 4 years visited our hospital. The patient has been engaged in security work for two shifts from 4 years ago. He complained of visual hallucinations about six months ago. Right before visiting the clinic, loss of recent memory was reported by family members. His personality changes such as getting angry or impatient began to appear. He received medication for hypothyroidism. In a neurological examination performed one year after symptom onset, truncal ataxia and slow gait were observed. The remainder of the neurologic examination was unremarkable. Thyroid stimulating hormone increased to 6.229 uIU/mL but ionized calcium decreased to 1.13 mMol/L. Parathyroid hormone was normal at 54.3 pg/mL. The APOE genotype was confirmed as ε3/ε4. His total years of education was 15 years. In the Korean version of the Mini-Mental State Examination, his score was 21. In the Seoul neuropsychological screening test, marked deterioration in visuospatial ability, frontal lobe function, and executive function was observed. In particular, in the Rey Complex Figure Test, the time required to complete the task increased to 338 seconds (<0.01 percentile). The performance score decreased to 1 point (<0.01 percentile). Except for language and attention deficits, there were global cognitive impairments, including visuospatial ability, memory, frontal lobe and executive function impairments. Computed tomography showed extensive calcification in the bilateral basal ganglia, cerebellum, thalami, centrum semiovale, and occipital lobes (**Fig. 1A**). Magnetic resonance imaging revealed diffuse brain atrophy. In the lesions where high attenuation was observed on computed tomography, showed low-signal intensity lesions on T2-weighted images, fluid attenuated inversion recovery, and susceptibility weighted images, and high-signal intensity lesions on T1-weighted images, suggestive of calcification (**Fig. 1B**).

About four months after the diagnosis, bradykinesia developed. In single photon emission computed tomography of the dopamine transporter (FP-CIT PET), there were no abnormal findings except for calcification in bilateral basal ganglia (**Fig. 1C**). In the early phase, contrast

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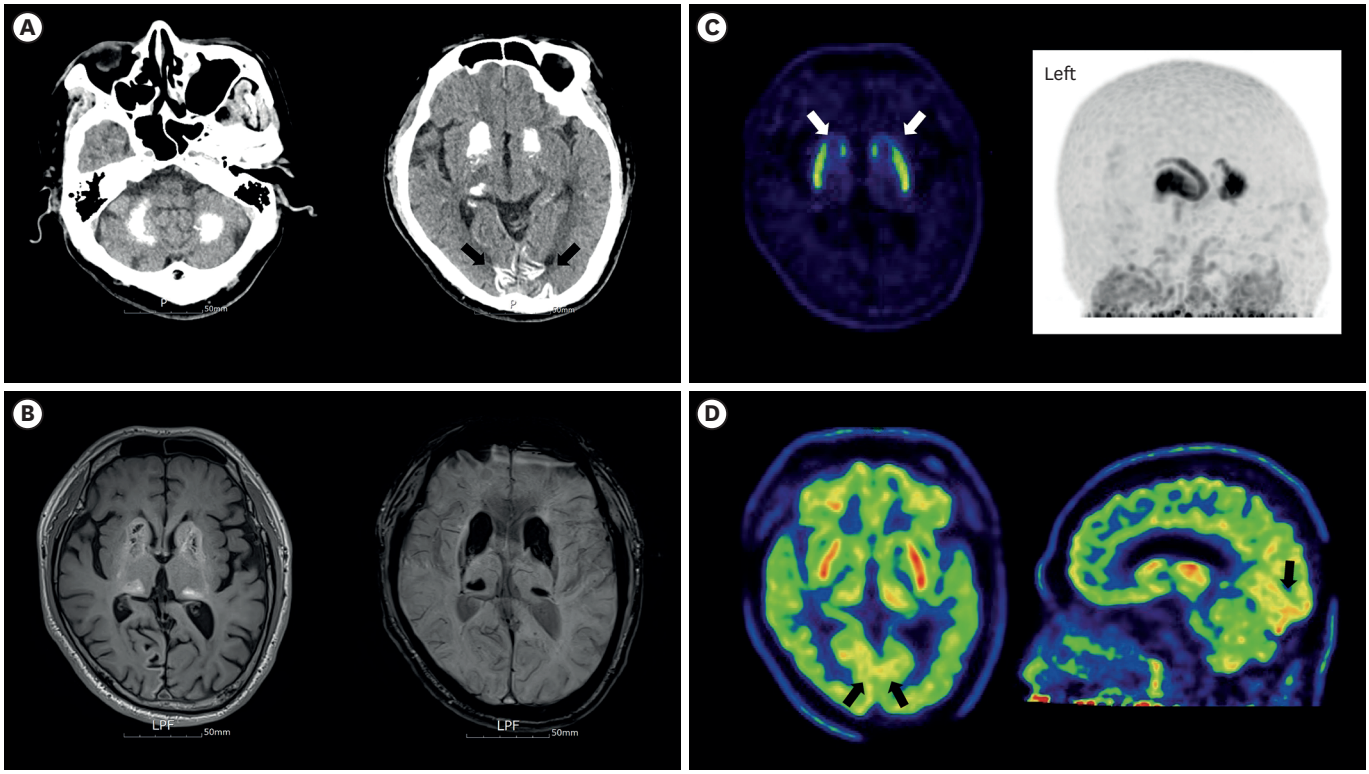


**Fig. 1.** (A) CT image of the patient showing extensive calcification in both basal ganglia, dentate nucleus of the cerebellum, and bilateral occipital cortices (black arrows). (B) High signal intensity lesions were observed in bilateral basal ganglia, thalami, and occipital cortices in T1 weighted images. Low signal intensity lesions were observed in SWIs. However, occipital cortices lesions were not prominent in the SWI scan. (C) Normal dopamine transporter image except for calcified caudate head lesion (white arrows). (D) Early phase study of single photon emission computed tomography of the dopamine transporter showing relatively normal tracer uptake in both basal ganglia, slightly increased uptake in bilateral occipital cortices (black arrows), and diffusely decreased bilateral cerebral cortices.
CT: computed tomography, SWI: susceptibility weighted image.

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was enhanced for lesions in calcified occipital lobes compared to that in other non-calcified cortices, along with a decrease in the uptake of contrast at the whole brain (**Fig. 1D**). Finally, he was diagnosed with FD, showing visuospatial dysfunction and visual hallucinations.

This case was a relatively young patient who presented with visual hallucinations due to impaired visuospatial function. Calcification of bilateral occipital cortices and decreased tracer uptake in the early phase of FP-CIT PET might affect the presentation of early visuospatial dysfunction. Furthermore, calcified occipital cortices could produce abnormal signals or lead to a reduction in sensory input that can lead to the production of spontaneous images from the visual association cortex, resulting in visual hallucination. In the early stage, the workup was performed with a possibility of neurodegenerative diseases including early-onset Alzheimer's disease (EOAD). The possibility of EOAD was considered low for the following two reasons. First, there were no typical AD patterns in cognitive function test. Atypical AD symptoms such as Blint syndrome, disinhibition, and impaired repetition deficit were not observed. Second, normal tracer uptake in posterior-cingulate and precuneus in the early stage of PF-CIT PET.

Since there was no clear family history, genetic diseases were not considered. FD is generally expressed as motor symptoms in most (55%) cases, whereas cognitive impairment is extremely rare in FD.[3] Since calcium deposition mainly occurs in the basal ganglia, it is


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thought to be related to the occurrence of symptoms. However, previous studies have shown that there is insufficient evidence that the degree of calcium deposition is directly related to the onset or severity of symptoms.[4] However, in this case, initial findings of FP-CIT PET are thought to be helpful in explaining the clinical course of patients with PD. Further research is needed to determine whether the degree and intensity of calcification could be related to a patient's symptoms.

## REFERENCES

1. Saleem S, Aslam HM, Anwar M, Anwar S, Saleem M, Saleem A, et al. Fahr's syndrome: literature review of current evidence. Orphanet J Rare Dis 2013;8:156.
PUBMED | CROSSREF
2. Manyam BV, Walters AS, Narla KR. Bilateral striopallidodentate calcinosis: clinical characteristics of patients seen in a registry. Mov Disord 2001;16:258-264.
PUBMED | CROSSREF
3. Heo JH, Choi YJ, Lee KM. A case of Fahr's disease presenting with frontal lobe dysfunction. J Korean Neurol Assoc 2007;25:549-552.
4. Calabrò RS, Spadaro L, Marra A, Bramanti P. Fahr's disease presenting with dementia at onset: a case report and literature review. Behav Neurol 2014;2014:750975.
PUBMED | CROSSREF