Fahr's disease: A rare neuropsychiatric disorder

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Abstract

Fahr's disease is a rare clinical neurodegenerative entity, occurring mainly in 4th or 5th decade, showing gradually progressive bilateral symmetric calcifications in basal ganglia, subcortical white matter, thalami or cerebellum, which can lead to movement disorder and/or neuropsychiatric manifestations. We present two cases in the same family; a 68-year-old brother had involuntary jerky movements of hand and dysarthria for 10 years while the 44-year-old sister had right lower limb spasticity and decreased vision for 2 years. The serial MRI scans showed slow progression in the bilateral subcortical white matter and cerebellar dentate nuclei calcifications along with surrounding reactive gliosis.

Keywords: Symmetrical basal ganglia calcifications, Neuropsychiatric disorder, Familial.
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Both of the cases were referred to our diagnostic service as undiagnosed cases of involuntary movements. The finding of high diagnostic importance in these siblings is the symmetry of brain calcifications as well as its pattern i.e.; these are coarse in the globus pallidus and white matter while serpentine in the dentate nuclei better appreciated on the MRI scan as areas of T1/T2/Flair low signal with GRE blooming artefact and on unenhanced CT scan as punctate hyperdense foci.

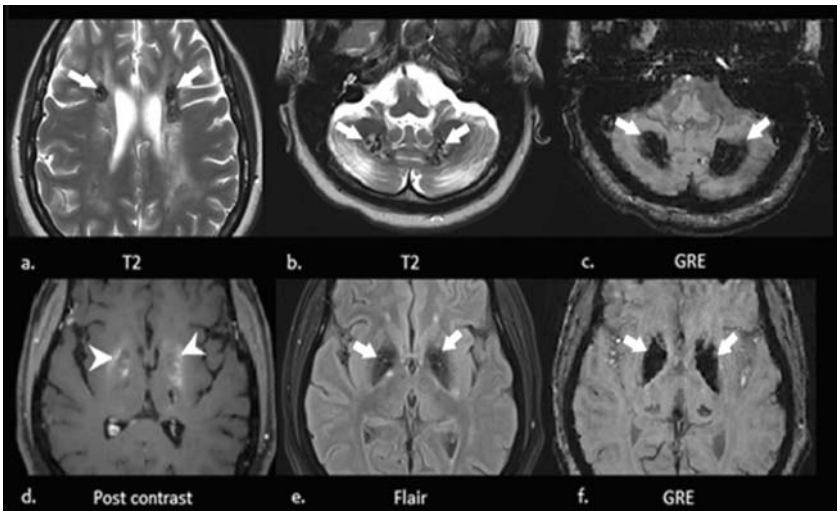

Figure: The T2 images demonstrating bilateral symmetrical calcifications (arrows) in subcortical deep white matter (a) and cerebellar dentate nuclei (b) in addition to the coarse basal ganglia calcifications (e) showing corresponding GRE blooming artefacts (c & f) There is mild post contrast enhancement (arrow heads in "d") and flair hyperintense surrounding gliosis (e).

Fahr's disease also called primary familial brain calcification or bilateral striatopallidodentate calcinosis typically presents with a movement disorder (extrapyramidal symptoms, dysarthria) and/or neuropsychiatric manifestations (psychosis, dementia, sensory changes).[1,2] These findings may correlate with the site of calcifications.[3] Diagnostic criteria consists of bilateral symmetrical coarse calcifications (typically in corpus striatum, globus pallidus and dentate nuclei), progressive neurologic dysfunction, genetic abnormality (positive family history, usually autosomal dominant pattern), typical age and no secondary causes of calcification (biochemical, infectious, traumatic or toxic cause).[4] The case presented here fulfilled the criteria with typical age, family history and characteristic imaging and clinical features.

References

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