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Labrune syndrome: A case Report

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Abstract

Labrune disease or Leukoencephalopathy with brain calcifications and cysts (LCC) is a rare genetic disease. It distinguishes by a triad of leukoencephalopathy, calcifications, and intracranial cysts. Recently, a mutation in the SNORD118 gene has been identified as the genetic basis involved in the disease. A 34-year-old woman presented with progressive left hemiparesis. Neuroimaging reveals multiple parenchymal cysts, diffuse white matter hyperintensities, and scattered calcifications. These radiological findings after ruling out other differential diagnosis lead to the diagnosis of LCC.

Introduction

Labrune disease is a rare autosomal recessive genetic disorder recognized by Leukoencephalopathy with brain calcification and cysts (LCC). Mutation in SNORD118 leads to this disease [1,2]. The disease manifests in different ways such as seizure, cerebellar ataxia, and cognitive and brainstem impairment [1,3,4]. The age of onset is different and it can occur in childhood, young or late adulthood [5,6].

A set of three imaging findings including diffuse white matter hyperintense signal on T2-weighted imaging, cerebral calcifications, and brain cysts in brain Magnetic Resonance Imaging (MRI) help in diagnosis [1]. In this report, we present a young woman with leukoencephalopathy, supratentorial calcifications, and scatter parenchymal cysts in the brain MRI.

Case presentation

A 34-year-old woman, without a previous past medical history of any disease, presented with progressive left hemiparesis in February 2021. She denied any remarkable familial history or drug abuse. Physical exams were normal except for the decreased force of the left limbs. A gadolinium-enhanced brain MRI study showed multiple cystic lesions, most notably in the brain stem and subcortical white matter, and diffuse white matter hyperintensity on T2-weighted images (Figure 1). The wall of cysts and some subcortical lesions showed enhancement.

Various infection, neoplastic and inflammatory work-ups were done and all of them were negative. She was treated with steroids without any improvement. The patient underwent two brain biopsies, the first biopsy showed astrogliosis, Rosenthal fiber, wall thickening in vessels, and bright eosinophilic material. The other revealed fibrinoid material, infiltration of histiocytes, and degenerative change.

When she was referred to our clinic in June 2022, based on the cystic lesion on MRI and extensive involvement of white matter, a brain Computed Tomography (CT) scan was done. A brain CT scan reveals calcification in the deep cerebral nuclei. Due to the simultaneous presence of cystic lesions, calcification, and leukoencephalopathy, Labrune disease was diagnosed.

Discussion

More than 100 cases of LCC were reported since 1996 [7]. Labrune disease is cerebral microangiopathy [1,2]. Biallelic mutations in the SNORD118 gene are considered to cause LCC [2]. In neuroimaging, multiple cysts, diffuse extensive white matter involvement with sparing corpus callosum and U-fiber, and the presence of calcification lead to LCC diagnosis [8-10]. Calcifica-

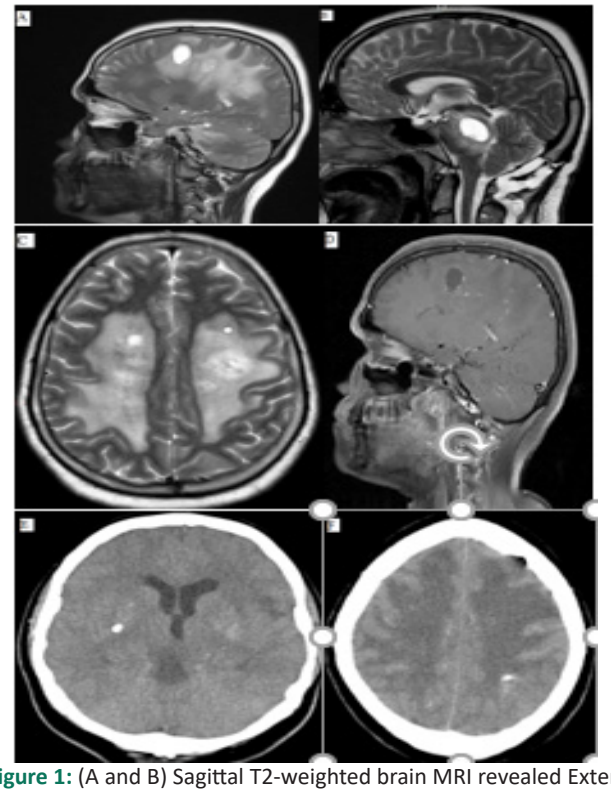

Figure 1: (A and B) Sagittal T2-weighted brain MRI revealed Extensive white matter hyperintensity with multiple cysts in subcortical white matter and (B) brainstem. (C) Axial T2-weighted brain MRI showed diffuse white-matter lesions with parenchymal cysts. (D) Sagittal T1-weighted brain MRI with contrast showed the wall of the cyst enhanced. It also showed scatter enhancement in the parenchyma. (E and F) Axial brain CT scan showed calcification in basal ganglia and subcortical white matter.

tion mostly is located in cerebral white matter or deep gray nuclei [11]. The cyst walls may be enhanced and also the mass effect is seen frequently [9]. The triad of extensive white matter T2 hypersignality, cerebral calcifications, and multiple cysts without the involvement of retinal or systemic presentation should propose the diagnosis of LCC [10].

Like our patient, in other Labrune-reported cases, sclerotic vessels with fibrinoid deposition, astrogliosis, Rosenthal fiber material, chronic microhemorrhage, and parenchymical calcification have been described [1,2,12,11,9,13,14,15].

Conclusion

In this article, we presented an adult-onset LCC. Clinical, radiological, and pathological findings are consistent with Labrune disease.

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