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Clinical Note

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Beta-thalassemia intermedia with ischemic transients misdiagnosed as multiple sclerosis: A case report

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Diagnosis of multiple sclerosis (MS) has shown to be a clinical challenge.[1] Different strategies have been proposed to minimize this potentially devastating mistake. It is crucial to pay attention to other problems that can mimic clinical and radiologic features of MS. Here we describe a case who received MS treatment, while the diagnosis was not MS at all.

A 35-year-old woman presented with repeated episodes of right-side numbness associated with neck pain and also low back pain. The episodes lasted for some minutes, repeated many times a day for two weeks. She mentions no previous or present visual symptom, skin lesion, or any other systemic symptom.

She was a known case of thalassemia intermedia and suffered major depression disorder. She was on hydroxyurea, folic acid, sertraline, quetiapine, and alprazolam. She had recently received a transfusion of one bag of packed cells. On physical exam, she was icteric and otherwise normal.

Complete blood count (CBC) was normal. Total bilirubin was 6.84 mg/dl (direct: 0.43). Fyb, Jka, Jkb antigens were positive, while s antigen was negative. Apart from wide diploic space due to thalassemia, magnetic resonance imaging (MRI) showed some nonspecific subcortical plaques, sparing corpus callosum (Figure 1).

Misdiagnosed with MS, she received interferon (IFN) beta-1a (Cinnovex®) injections for two months. She was referred to Sina MS clinic (Tehran, Iran) for a second opinion. The spinal MRI came out to be normal. Cerebrospinal fluid (CSF) was negative for oligoclonal bands (OCBs). As none of the plaques showed typical characteristics of MS (not touching cortex, no callosal, infratentorial, or spinal lesion), IFN was discontinued.

Cardiac evaluation, vasculitis and hypercoagulability panel, and Doppler ultrasound of cervical arteries were all normal. Aspirin was started for the patient. She has been symptom-free since then.

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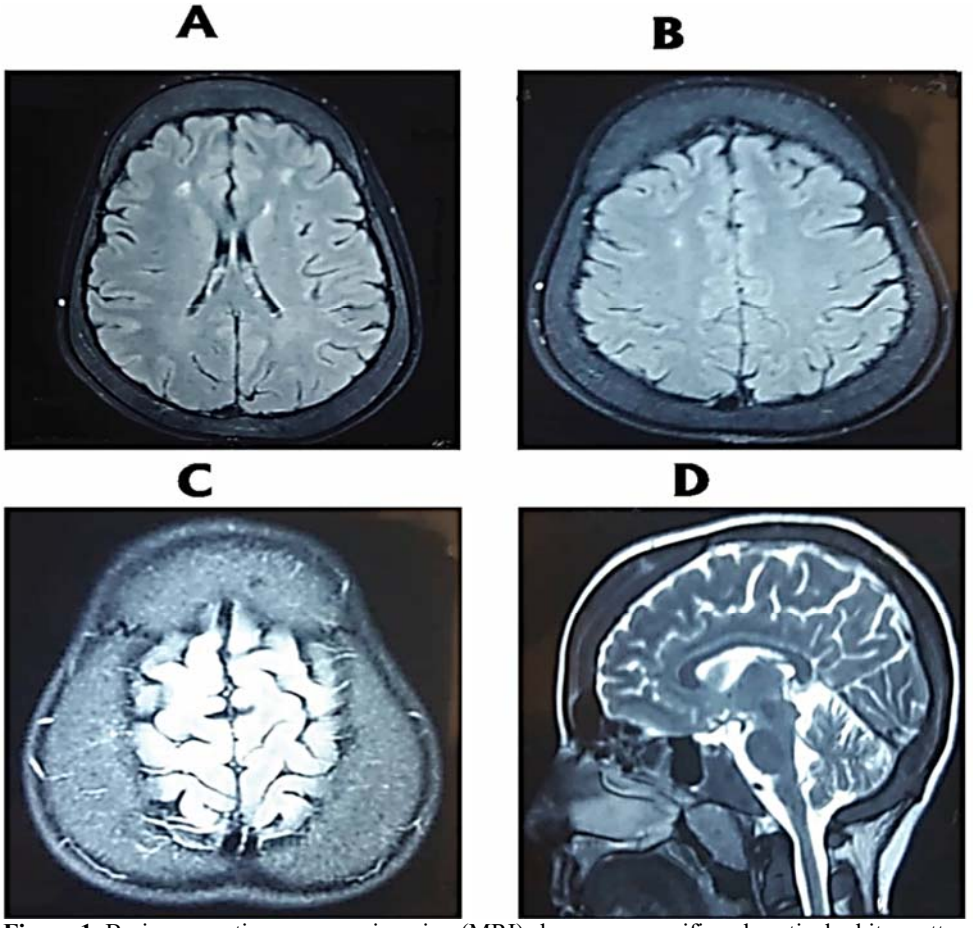

Figure 1. Brain magnetic resonance imaging (MRI) shows nonspecific subcortical white matter lesions. Note the significant widening of diploic space due to extramedullary hematopoiesis.

Silent ischemic lesions are reported in about 60% of patients with beta-thalassemia intermedia.[2] Multiple factors contribute to these ischemic insults, like increased platelet count and aggregation, alterations in coagulation factors, and iron overload.[3] As the pathology is ischemic, cerebral lesions mostly involve subcortical white matter. Here is the point where attention should be paid not to misdiagnose them with demyelinating plaques.

There are diagnostic criteria for MS (like the 2017 McDonald criteria and Barkhof criteria) to assist clinical judgment. Newer more specific radiologic characteristics are also defined (e.g. central vein sign, subpial demyelination, and lesional rims). But still, 30%-67% of referred cases to tertiary MS clinics with an initial diagnosis of MS, would be finally diagnosed with other differentials.[4] The most notable item leading to misdiagnosis was applying diagnostic criteria to an atypical neurologic syndrome for MS. Another mistake is not paying attention to the radiologic characteristics of the lesions.[5] The 2017 McDonald criteria insist on dissemination in time and space, typical sites of involvement, and "no better explanation", none of which was applied to our patient.

To remember is, although MS is increasingly diagnosed by enhanced diagnostic tools, its misdiagnosis leads to unnecessary psychological, economic, and safety burdens to both patients and society. One of its differential diagnoses that should be in mind, while facing a thalassemia patient, is silent or symptomatic ischemic white matter lesions. Detailed history taking and considering the site and characteristics of the MRI lesions could be helpful to diagnostic justification.

Conflict of Interests

The authors declare no conflict of interest in this study.

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References

1. Solomon AJ, Pettigrew R, Naismith RT, Chahin S, Krieger S, Weinshenker B. Challenges in multiple sclerosis diagnosis: Misunderstanding and misapplication of the McDonald criteria. Mult Scler 2021; 27(2): 250-8.
2. Musallam KM, Taher AT, Karimi M, Rachmilewitz EA. Cerebral infarction in beta-thalassemia intermedia: Breaking the silence. Thromb Res 2012; 130(5): 695-702.
3. Pazgal I, Inbar E, Cohen M, Shpilberg O, Stark P. High incidence of silent cerebral infarcts in adult patients with beta thalassemia major. Thromb Res 2016; 144: 119-22.
4. Solomon AJ, Naismith RT, Cross AH. Misdiagnosis of multiple sclerosis: Impact of the 2017 McDonald criteria on clinical practice. Neurology 2019; 92(1): 26-33.
5. Solomon AJ, Bourdette DN, Cross AH, Applebee A, Skidd PM, Howard DB, et al. The contemporary spectrum of multiple sclerosis misdiagnosis: A multicenter study. Neurology 2016; 87(13): 1393-9.