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Case Report

Early-Onset Multiple Sclerosis in Identical Twin Sisters: A Case Report

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Abstract

Background: Multiple Sclerosis (MS) is a chronic immune-mediated demyelinating disorder of the central nervous system. Early-onset MS is uncommon and may present with atypical clinical features. Genetic susceptibility plays a significant role, with higher concordance rates among monozygotic twins.

Case Presentation: We report a pair of identical twin sisters who developed early-onset MS at the age of 13 and 17 years. Case 1 presented initially with quadriparesis and later with longitudinally extensive transverse myelitis, mimicking neuromyelitis optica, while Case 2 exhibited progressive limb weakness more typical of MS. Both patients had positive CSF oligoclonal bands, abnormal MRI findings, and delayed visual evoked potentials. Serological markers for NMOSD and MOG-associated disease were negative.

Management and Outcome: Both twins responded to intravenous methylprednisolone followed by oral tapering of steroid and long-term azathioprine therapy. Case 1 experienced a relapse after discontinuing azathioprine, whereas Case 2 remained relapse-free.

Conclusion: These cases highlight the strong genetic component of MS and the phenotypic variability even among monozygotic twins. Sustained immunosuppressive therapy remains essential in resource-limited settings where disease-modifying therapies are unavailable.

Keywords: Multiple sclerosis, Early onset, Identical twins, Longitudinally extensive transverse myelitis, Genetics, Azathioprine

Introduction

Multiple Sclerosis (MS) is an autoimmune, inflammatory, demyelinating disease of the central nervous system with a multifactorial etiology involving genetic and environmental factors [1]. The global incidence is approximately 2.1 per 100,000 persons per year [2]. Although MS typically presents in early adulthood, pediatric and adolescent-onset MS account for 3-10% of all cases [3]. Genetic susceptibility is well established, with more than 200 risk variants identified. However, none are uniquely present in affected individuals [4]. Monozygotic twins have a significantly higher concordance rate than dizygotic twins, underscoring the strong heritable component [5]. We report early-onset MS in identical twin sisters with differing clinical manifestations, highlighting genetic and phenotypic considerations important for diagnostic and therapeutic decision-making.

Case Presentation

Case 1

A 13-year-old girl experienced sudden quadriparesis in 2017, which improved after treatment abroad. At age 17, she presented with two weeks of gait ataxia and progressive quadriparesis. Neurological examination revealed spastic quadriparesis (MRC 2/5), a sensory impairment up to C5 sensory level, and left cerebellar dysfunction. Treatment with intravenous methylprednisolone followed by oral tapering and azathioprine led to significant improvement, with mild residual deficits (MRC 5-/5). At age 21, she re-presented with right lower limb weakness (MRC 2/5) and persistent mild left cerebellar signs. No sensory or sphincter involvement was noted.

Case 2

Her identical twin sister presented at age 17 with two weeks of progressive limb weakness, predominantly right-sided, without sensory or sphincter involvement. She had no prior neurological illness.

Neither sister reported recent infection, vaccination, or toxin exposure.

Investigations

a) Cerebrospinal Fluid: Positive oligoclonal bands; normal cell counts and biochemistry

b) Serology: Negative aquaporin-4 IgG, anti-MOG IgG, ANA, ENA, and infectious serologies

c) Routine Biochemistry: Within normal limits

d) Visual Evoked Potentials: Bilaterally delayed P100 latency with reduced amplitude

e) MRI Brain and Spinal Cord: Shown in below.

6. Differential Diagnosis

a) Neuromyelitis Optica Spectrum Disorder (NMOSD): Considered due to longitudinally extensive transverse myelitis in Case 1 but excluded based on MRI findings and negative AQP4/MOG antibodies.

b) Acute Disseminated Encephalomyelitis (ADEM): Excluded due to absence of encephalopathy, relapsing–remitting course, and positive oligoclonal bands.

c) Metabolic, Infectious, or Toxic Myelopathy: Excluded by normal laboratory tests and clinical presentation.

Final Diagnosis: Relapsing–Remitting Multiple Sclerosis (RRMS) based on the 2017 McDonald criteria.

Treatment and Outcome

Both twins received:

i. Intravenous methylprednisolone 1 g/day for 5 days

ii. Oral steroid tapering

iii. Long-term azathioprine (2 mg/kg/day)

Case 1: Relapsed one year after discontinuing azathioprine but improved upon retreatment and resumption of therapy (Expended Disability Status Scale, EDSS 2).

Case 2: Remained relapse-free and has been off azathioprine for one year (EDSS 0).

Due to limited availability of disease-modifying therapies (DMTs), management relied on corticosteroids and azathioprine (Figure 1-6).

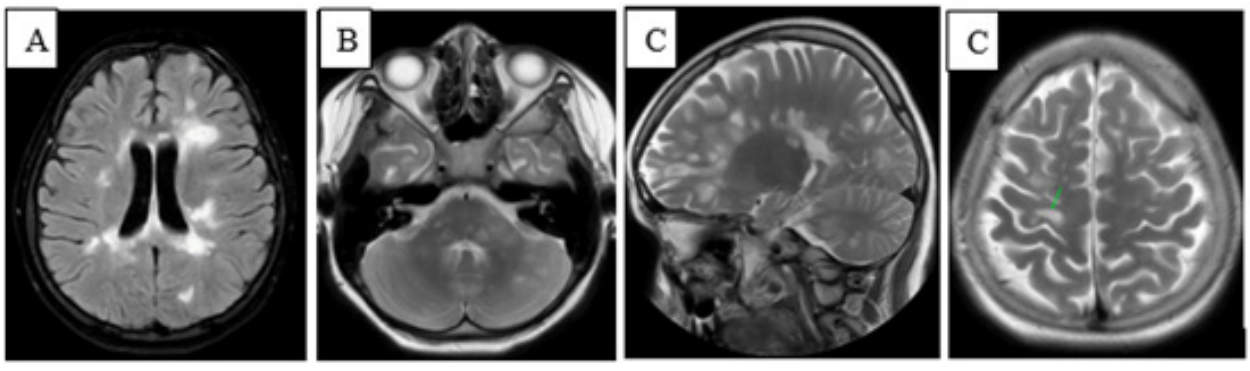

Figure 1: MRI Brain (2021) showed T2 FLAIR hyperintense lesions in both supra- and infratentoral brain A and B, Dawson fingers appearance of periventricular lesions (C) (Case 1).

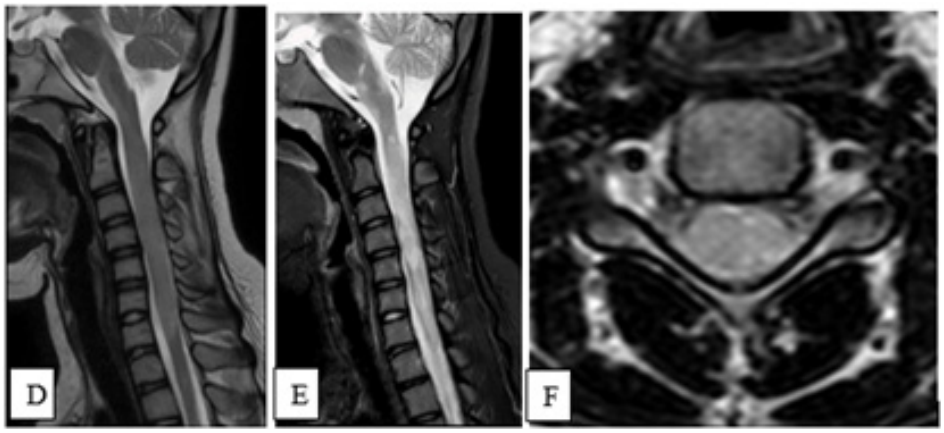

Figure 2: MRI Spine (2021) showed longitudinally extensive transverse myelitis (Case 1).

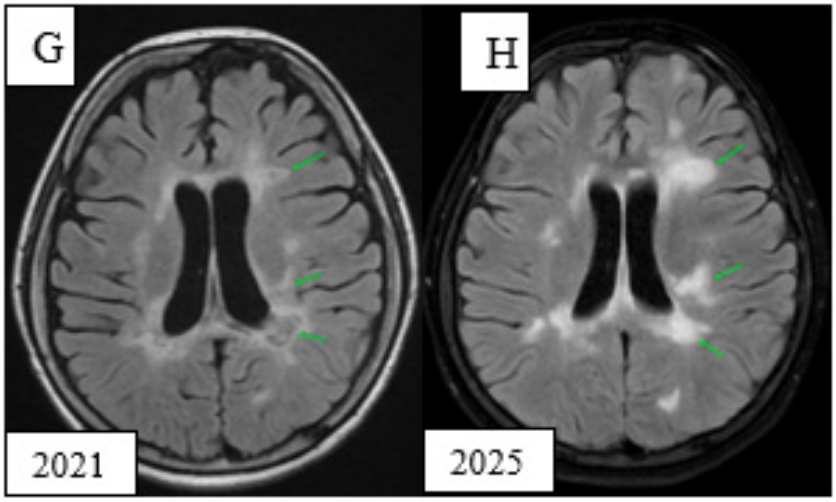

Figure 3: MRI Brain with contrast showed new contrast enhancing lesions (arrows) in periventricular white matter in (H) 2025 as compared to (G) in 2021 (Case 1).

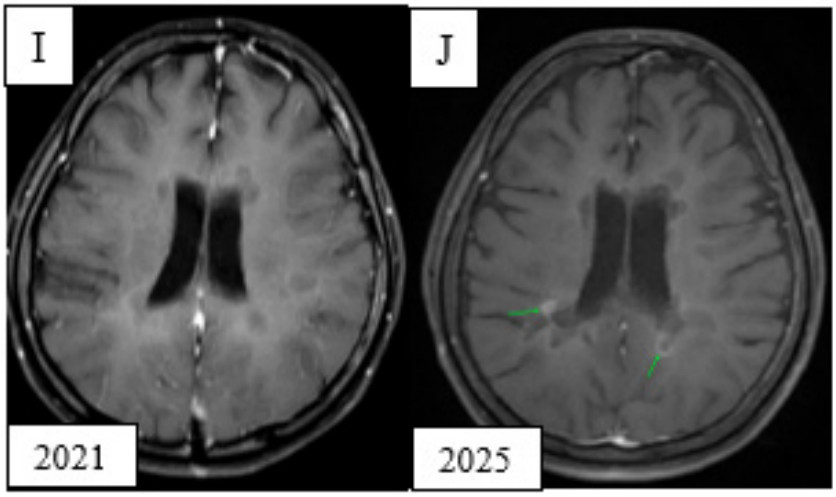

Figure 4: MRI Brain with contrast showed interval resolution of oedema in periventricular lesions (arrows) shown in (J) 2025 as compared to (I) 2021 (Case 1).

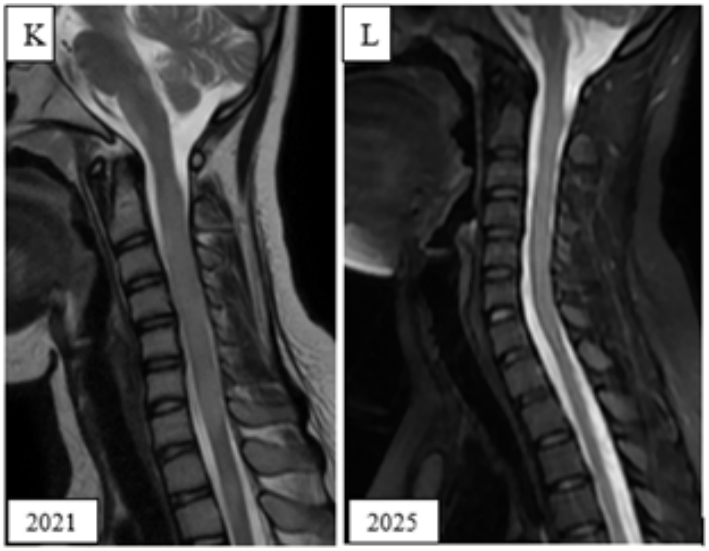

Figure 5: MRI spine: Interval resolution of transverse myelitis (2021 vs 2025) (Case 1).

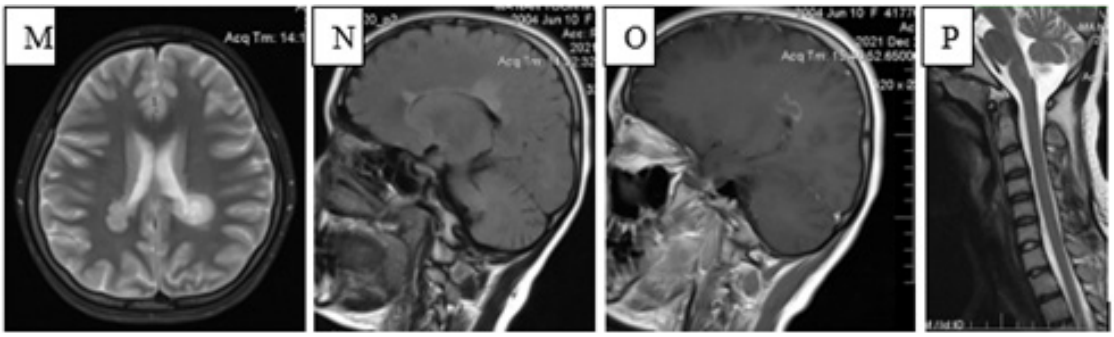

Figure 6: MRI (Brain and spine) showed multiple periventricular white matter lesions with active lesion and Dawson's finger appearance and normal spine MRI (Case 2).

Discussion

These identical twin sisters developed MS during early adolescence, an age group where MS is relatively uncommon. Case 1 exhibited longitudinally extensive transverse myelitis, an atypical manifestation that can mimic NMOSD, while Case 2 showed more typical MS features. The 25–30% concordance rate for MS among monozygotic twins compared with less than 5% in dizygotic twins or siblings highlights the strong genetic contribution to disease pathogenesis [5]. However, the phenotypic variability observed suggests environmental and epigenetic factors modulate clinical expression. These cases emphasize the importance of early recognition of MS in patients with a strong genetic background and highlight the need for accessible immunosuppressive therapy in resource-limited settings.

Learning Points

a) MS can present at an unusually early age in genetically predisposed individuals.

b) Longitudinally extensive transverse myelitis may occur as an atypical feature of MS and mimic NMOSD.

c) Identical twins with MS demonstrate strong genetic susceptibility but variable phenotypic expression.

d) Sustained immunosuppressive therapy such as azathioprine is beneficial where DMT access is limited.

e) Genetic testing and advanced immunological evaluation are important for understanding familial MS.

Ethical Considerations

Written informed consent for publication was obtained from both patients and their mother.

Conflicts of Interest

The authors declare no conflicts of interest.

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