Teaching NeuroImages: Neuroradiologic evolution of Leigh disease

OPEN

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Figure 1 MRI head (age 2 years)

Axial T2-weighted images (A-C) reveal hyperintensities involving the left globus pallidus, midbrain, and medulla, with diffusion restriction evident on diffusion-weighted sequences (D-F).

A 2-year-old girl with no significant family history presented with motor developmental delay and strabismus. MRI revealed unilateral basal ganglia and brainstem lesions (figure 1). Eighteen months later, she developed acute onset right arm weakness, leading to a diagnosis of multiphasic disseminated encephalomyelitis. Treatment with steroids and mycophenolate produced no symptomatic or imaging improvement. Her condition progressed, with ataxia, multifocal dystonia, spasticity, and loss of ambulation at age 6 years. Serial imaging showed evolution of bilateral basal ganglia changes, compatible with

Leigh syndrome¹ (figure 2). A heteroplasmic mutation, m.12706T>C in the *MTND5* gene, was identified in muscle.

AUTHOR CONTRIBUTIONS

Y.S.N. conceptualized this work, interpreted the data, and wrote the first draft of the manuscript. M.L. collected and interpreted the data and critically reviewed the manuscript. G.T. collected and interpreted the data and critically reviewed the manuscript. R.M. conceptualized this work, interpreted the data, and revised and critically reviewed the manuscript.

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Figure 2 Serial MRI head images (T2 and diffusion-weighted)

Serial T2 (A–C) and diffusion-weighted (D–F) images reveal an evolving hyperintense lesion in the right globus pallidus (age 4 years). This becomes symmetrical by age 5–6 years.

DISCLOSURE

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