Neuroradiology

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A 5-year-old boy with complaints of impaired vision, gait disturbance, and delayed milestones was referred for MRI of the brain. On examination, the child was mentally retarded. He had no perception of light. He had pendular nystagmus and convergent squint. He could stand with support. Fundoscopic examination of the eye showed bilateral hyperemic discs with blurred margins and whitish stippling of the retina, with attenuated vessels. There was no evidence of optic atrophy. MRI of the brain was performed [Figures 1 and 2].

What is the diagnosis?

Figure 1: Axial T2W MRI of the brain

Figure 2: Axial T2W MRI of the brain