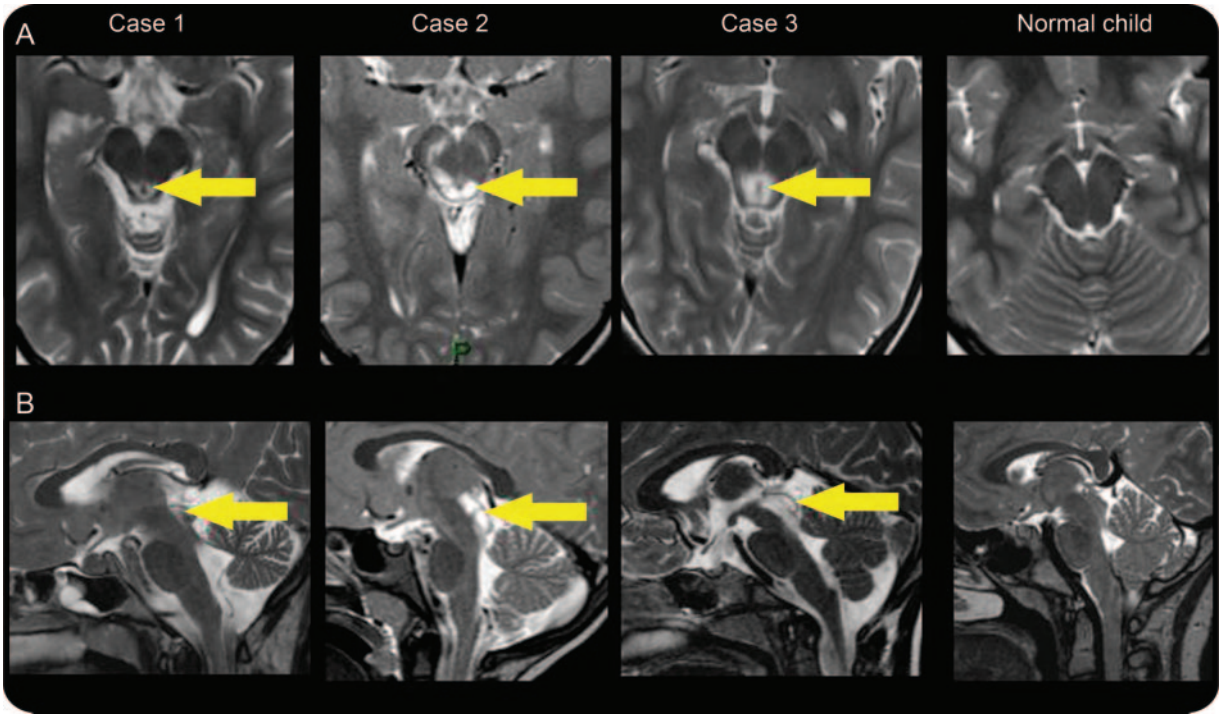
Mitochondrial ND5 mutations mimicking brainstem tectal glioma

Figure MRI showing a periaqueductal high T2 signal on axial T2 (A) and sagittal T2 (B) and an enlargement of the tectum that mimicked tectal glioma in all 3 children (case 1, 9 years; case 2, 6 years; case 3, 7 years)



We report MRI periaqueductal T2 hypersignal suggestive of tectal glioma in 3 unrelated children with reduced vision and normal mental development (figure). Increased CSF lactate and optic atrophy in the first case suggested mitochondrial dysfunction. Muscle biopsy revealed complex I deficiency. A heteroplasmic mt-ND5 mutation was found (m.13513G>A).[1] The second case presented with similar clinicoradiologic features, complex I deficiency, and the same heteroplasmic mutation. The third case had visual disturbance without optic atrophy, normal muscle enzyme activities, and a heteroplasmic mt-ND5 mutation (m.13514A>G).[2] Even in absence of optic atrophy, mental retardation, or multiorgan dysfunction, the combination of visual disturbance and periaqueductal T2 hypersignal should prompt the search for mitochondrial DNA mutation.

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Disclosure: The authors report no disclosures.

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