Teaching NeuroImages: The Putaminal Eye, A Highly Characteristic Imaging Feature of MEGDEL Syndrome

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A 22 month-old female product of a consanguineous marriage presented with recurrent nausea, diarrhea, elevated liver enzymes, metabolic acidosis and developmental regression. Urine analysis revealed elevated 3-methylglutaric acid and 3-methylglutaconic acid. Brain MRI showed bilateral basal ganglia injury (Figure 1A-C) and cerebellar atrophy (Figure 1D). Auditory brainstem response assessment demonstrated bilateral profound sensorineural hearing loss. DNA analysis revealed pathogenic homozygous variant of the SERAC1 gene, diagnostic of MEGDEL syndrome (3-MElthylglutaconic aciduria, Deafness, Encephalopathy, Leigh-like syndrome).

MEGDEL syndrome is an infantile onset syndrome characterized by dystonia, deafness, progressive spasticity, developmental delay or regression, and 3-methylglutaconic aciduria. The causative SERAC1 gene encodes a phosphatidylglycerol remodeler, essential for mitochondrial function and intracellular cholesterol trafficking. When imaging at the appropriate stage (1 to 4 years of age), the pattern of basal ganglia injury on T2-weighted images sparing the middorsal putamina, called “putaminal eyes,” is pathognomonic. Life expectancy is unknown. Some patients die in infancy.
Figure 1 Classic MRI brain findings in MEGDEL

Axial T2-weighted image (A) showing symmetrical T2 hyperintensity with atrophy of the caudate nuclei (short arrows) and T2 hyperintensity of the putamina with sparing of their middorsal portions (long arrows). Axial diffusion image (B) and ADC map (C) showing mixed diffusion characteristics in the areas of injury: restricted diffusion (long arrows, sites of active injury) and facilitated diffusion (short arrows). Coronal T2-weighted image (D) showing cerebellar atrophy (arrow).
References

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