A 4-year-old female child with an uneventful perinatal history, presented with complaints of hypotonia, delayed cognitive development, and abnormal eye movements since birth. Clinically, she had hypotonia, grade 3/5 power in all limbs, absent deep tendon reflexes, and nystagmus. Serum creatinine kinase level was mildly elevated (560 U/L). Fundoscopy was normal, and electromyography showed a myopathic pattern.

Brain magnetic resonance imaging (MRI) of the child showed a markedly hypoplastic pons with a small notch on its ventral surface giving a Z-shaped appearance to the brainstem [Figure 1]a. The other findings were right anterior frontal lobe polymicrogyria [Figure 1]b, hypogenesis of vermis, cerebellar polymicrogyria with cysts [Figure 2]a, bilateral occipital cobblestone lissencephaly [Figure 2]b, and patchy areas of T2 prolongation in right temporal and periventricular white matter. Based on the MRI findings, we classified our patient as having Fukuyama congenital muscular dystrophy (CMD). [Figure 1]• [Figure 2]

CMDs are a heterogeneous group of disorders characterized by hypotonia, weakness, and congenital contractures. [1]

Three well-defined syndromes, Fukuyama CMD, Walker-Warburg syndrome and muscle-eye-brain disease, which
are all associated with structural brain changes, were separated from a “pure,” or classical form of CMD, not associated with structural brain changes by an International Consortium in 1993.[2]

The information obtained from high-quality MRI imaging of the brain is useful to properly classify children with CMDs.[1]

The following patterns of MR abnormalities are seen in CMD as described by Barkovich:[1] (1) Patients with pure CMDs have diffuse central cerebral hypomyelination with mild pontine and cerebellar vermian hypoplasia; (2) patients with Fukuyama CMD have a diffuse central cerebral hypomyelination, cerebellar polymicrogyria (with or without cysts), frontal polymicrogyria, hypoplasia of pons and cerebellar vermis, and occasionally occipital cobblestone cortex; (3) patients with muscle–eye–brain disease have cerebellar polymicrogyria (with or without cysts), absence of septum pellucidum, diffuse cerebral cortical dysplasia, pontine and cerebellar vermian hypoplasia, patchy hypomyelination, and variable callosal hypogenesis and hydrocephalus; and (4) patients with Walker–Warburg syndrome have severe diffuse cobblestone cortex, complete absence of cerebral and cerebellar myelin, cerebellar polymicrogyria (with or without cysts), pontine and cerebellar vermal hypoplasia, hydrocephalus, and variable callosal hypogenesis.

The MRI features can be used to facilitate the diagnosis of CMDs, particularly if central nervous system involvement dominates the clinical picture and there is a lack of technological skill to establish the molecular diagnosis.[3]

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Conflicts of interest

There are no conflicts of interest.

References

