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# An m.8344A>G Carrier Does Not Have Dorsal Midbrain Syndrome If Pons Is Affected

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**Abstract:** Purpose: to comment on the paper by Kasetty, *et al*. Methods: comment on a recently published article. Conclusions and importance: Dorsal midbrain syndrome, also known as Parinaud syndrome, sylvian aqueduct syndrome, pretectal syndrome, and Koerber-Salus-Elschnig syndrome, is clinically characterised by the triad of impaired upward gaze, convergence retraction nystagmus, and pupillary hyporeflexia. It has not been reported in association with the variant m.8344A>G in MT-TK. However, if there is also a symmetric pontine lesion, the clinical presented should not be termed dorsal midbrain syndrome.

**Keywords:** mtDNA; mitochondrial disorder; MERRF; m.8344A>G; dorsal midbrain syndrome.

## BRIEF REPORT

We read with interest the article by Kasetty, *et al*., about a 10 year-old female with developmental delay, absence seizures, ataxia, and dorsal midbrain syndrome manifesting as accommodative insufficiency, impaired supraduction, and convergence retraction nystagmus [Kasetty, M. *et al*., 2023]. Cerebral MRI showed symmetrical lesions in the dorsal pons and genetic studies revealed the mtDNA variant m.8344A>G in *MT-TK* [Kasetty, M. *et al*., 2023]. The study is excellent but raises concerns that should be discussed.

We disagree that the variant m.8344A>G was "previously undiagnosed" as stated in the abstract [Kasetty, M. *et al*., 2023]. The m.8344A>G variant is a known pathogenic mutation in *MT-TK* and the most common cause of myoclonic epilepsy with ragged-red fibers (MERRF) syndrome, where it accounts for 80% of cases [Velez-Bartolomei, F. *et al*., 2003].

We also disagree with the conclusions that the index case is unique for her ophthalmologic abnormalities, as mentioned in the abstract [Kasetty, M. *et al*., 2023]. There are numerous reports, which describe patients with syndromic or non-syndromic mitochondrial disorders (MIDs) who present with complex optomotor abnormalities [Budumuru, U. *et al*., 2023].

We disagree with the statement n the discussion that MIDs are multisystem disease in general [Kasetty, M. *et al*., 2023]. Although we agree that in most cases MIDs are multisystem diseases, there are several MID patients who manifest only with cerebral involvement, skeletal muscle involvement or only cardiac muscle involvement. This is particularly the case at onset of the disease.

We disagree that the clinical presentation resembles MELAS syndrome [Kasetty, M. *et al*., 2023]. Cerebral lesions in MELAS are usually asymmetric and eye movements are usually preserved or minimally impaired.

There is a discrepancy between the clinical diagnosis of "dorsal midbrain syndrome" and the radiological finding of "symmetric lesions in the pons" [Kasetty, M. *et al*., 2023]. The pons is not a part of the midbrain, but is already part of the brainstem. How do the authors explain the discrepancy between the clinical presentation and the radiological findings?

Overall, the interesting study has limitations that call into question the results and their interpretation. Clarifying these weaknesses would strengthen the conclusions and could add value to the study. The pathogenicity of the m.8344A>G variant has been confirmed by many studies and several MIDs exhibit complex optomotor abnormalities. Ophthalmologists should be aware of the fact that optomotor abnormalities may reflect central or peripheral nervous system involvement in MIDs.

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