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Letter to the Editor

In a 6-months-old m.3243A>G Carrier with Pleocytosis, Causes Other than the Mutation must be Ruled Out

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Letter to the Editor

We read with interest the article by Janiak *et al*. about a 3-year-old boy with a history of seizures, developmental delay, strabismus, spasticity, a Blake cyst, and cerebral atrophy at 6.5 months of age [1]. The patient benefited from vigabatrim and valproic acid respectively levetiracetam [1]. The study is interesting, but some points should be discussed.

The first point is that the index patient had pleocytosis (56/µl (n, <5/µl)), but no examination for pleocytosis was reported [1]. The cell count in cerebrospinal fluid (CSF) is usually normal in m.3243A>G carriers [2]. Based on these considerations, we do not agree that all clinical symptoms, especially the seizures, are attributable to the m.3243A>G variant, but rather to infectious encephalitis, autoimmune encephalitis (AIE), or a paraneoplastic disorder. If cerebrospinal fluid samples are still available, they should be tested for neurotropic viruses, tuberculosis, antibodies associated with AIE, and malignancy.

The second point is that lactate was not measured in either serum or cerebrospinal fluid [1]. In m.3243A>G carriers with central nervous system involvement, lactate levels in cerebrospinal fluid are often elevated [3]. Elevated lactate levels in CSF can be detected not only by direct measurement of lactate in CSF, but also by magnetic resonance spectroscopy (MRS) [3]. If CSF samples are still available, lactate levels in CSF should be determined. In addition, serum lactate levels should be determined if the patient is still alive and blood sampling is possible.

The third point is that a stroke-like lesion (SLL) was not ruled out as the cause of the seizures. Since m.3243A>G carriers often have stroke-like episodes (SLEs) [4], it would have been crucial to rule out an SLL, the imaging equivalent of an SLE, by multimodal MRI. SLLs have a typical pattern and dynamic course [5].

The fourth point is that we disagree with the diagnosis of “Blake's cyst” [1]. A Blake's cyst typically occurs in the posterior cranial fossa, but the location indicated in Figure 1 is not the posterior cranial fossa. Given that the patient had pleocytosis, a malignant tumor must be ruled out by MRS, biopsy, or resection.

The fifth point is that no long-term follow-up was reported [1]. Since the current age was stated as 3 years in the abstract, a follow-up of at least 2.5 years should be available. The quality of seizure control during this period and the possible occurrence of additional phenotypic manifestations of the m.3243A>G variant should be reported.

Finally, we should know whether the m.3243A>G variant was inherited or occurred de novo. Did the patient's mother carry the m.3243A>G variant or did show signs of mitochondrial disease?

In summary, pleocytosis is unusual in a carrier of the m.3243A>G variant, and encephalitis and malignancy must be ruled out. In addition, a SLE should be ruled out in carriers of the m.3243A>G variant with seizures.

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