

Transient symmetric T2-hyperintensities of basal ganglia and brainstem not only point to Leigh syndrome

Josef Finsterer, Sinda Zarrouk-Mahjoub

► **To cite this version:**

Josef Finsterer, Sinda Zarrouk-Mahjoub. Transient symmetric T2-hyperintensities of basal ganglia and brainstem not only point to Leigh syndrome. *Annals of Indian Academy of Neurology*, Medknow Publications, 2016, 19 (3), pp.419 - 419. <10.4103/0972-2327.186857>. <pasteur-01446489>

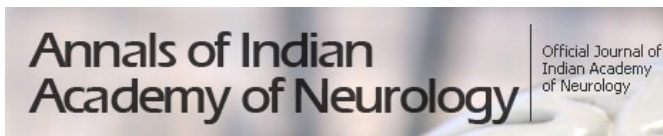
HAL Id: pasteur-01446489

<https://hal-riip.archives-ouvertes.fr/pasteur-01446489>

Submitted on 2 Oct 2017

HAL is a multi-disciplinary open access archive for the deposit and dissemination of scientific research documents, whether they are published or not. The documents may come from teaching and research institutions in France or abroad, or from public or private research centers.

L'archive ouverte pluridisciplinaire **HAL**, est destinée au dépôt et à la diffusion de documents scientifiques de niveau recherche, publiés ou non, émanant des établissements d'enseignement et de recherche français ou étrangers, des laboratoires publics ou privés.



LETTER TO THE EDITOR

Year : 2016 | Volume : 19 | Issue : 3 | Page : 419-420

Transient symmetric T2-hyperintensities of basal ganglia and brainstem not only point to Leigh syndrome

Josef Finsterer¹, Sinda Zarrouk-Mahjoub²,¹ Krankenanstalt Rudolfstiftung, Vienna, Austria² Genomics Platform, Pasteur Institute of Tunis, Tunis, Tunisia

Correspondence Address:

Josef Finsterer
Postfach 20, 1180 Vienna
Austria

How to cite this article:

Finsterer J, Zarrouk-Mahjoub S. Transient symmetric T2-hyperintensities of basal ganglia and brainstem not only point to Leigh syndrome. *Ann Indian Acad Neurol* 2016;19:419-420

How to cite this URL:

Finsterer J, Zarrouk-Mahjoub S. Transient symmetric T2-hyperintensities of basal ganglia and brainstem not only point to Leigh syndrome. *Ann Indian Acad Neurol* [serial online] 2016 [cited 2017 Oct 2];19:419-420Available from: <http://www.annalsofian.org/text.asp?2016/19/3/419/186857>

Full Text

Sir,

With interest we read the article by Jabeen et al. about a 37-year-old female with suspected adult-onset Leigh syndrome who profited from the administration of a mixture of thiamine, riboflavin, coenzyme-Q, L- carnitine, and L-arginine. [1] We have the following comments and concerns.

We doubt the diagnosis of adult Leigh syndrome. Arguments against Leigh syndrome are that the central nervous system lesions and the clinical manifestations completely resolved, that no mtDNA mutation was detected, that the family history was negative for a mitochondrial disorder (MID), and that ragged-red fibers were absent on muscle biopsy. Immunohistological findings on muscle biopsy could be secondary, as well as elevated serum lactate and the magnetic resonance spectroscopy findings. Bilateral T2-hyperintense lesions of the thalamus, [2] the periaqueductal grey, [3] the hypothalamic region [3] have also been reported in patients with thiamine deficiency. The patient did not undergo lactate stress testing or investigations for mutations in nuclear DNA-encoded genes.

Which were the results of the biochemical investigations of the muscle homogenate? Was there isolated Complex I, Complex II, Complex III, Complex IV deficiency or multiple complex deficiencies as has been previously reported in Leigh syndrome? [4],[5] Which were the muscle biopsy findings on electron microscopy? Was there cristae destruction, swelling of mitochondria, intramitochondrial glycogen deposition, or paracrystalline inclusions?

Which was the cause of abdominal pain and vomiting since 3 months? Did the patient suffer from gastrointestinal pseudoobstruction as has been reported in patients with Mitochondrial neurogastrointestinal encephalomyopathy syndrome. [6] Was aseptic pancreatitis, repeatedly reported as a manifestation of MIDs, the cause of her complaints?

Which was the cause of breathlessness and shock when the patient was readmitted shortly after dismissal? Which type of shock was diagnosed, cardiac or septic? Could an unwitnessed seizure or self-limiting status epilepticus (e.g., aborted sudden unexpected death in epilepsy) explain metabolic acidosis or lactate elevation? Was an electroencephalogram recorded during this episode? Was creatine kinase, troponine-T, or proBNP elevated? Which were the results of the electrocardiogram and echocardiography at that time? Were there any indications for acute heart failure due to a Takotsubo syndrome, which also could explain the acute deterioration? Was the cause of respiratory insufficiency cardiac, infectious, or muscular? For how long did the patient require artificial ventilation?

Overall, the presented data strongly argue against an MID. MIDs are usually progressive in nature and only marginally respond to vitamin/cofactor cocktails. The only MID, which potentially responds to such compounds is primary coenzyme-Q deficiency, [7] which often responds favorably to coenzyme-Q, and should be excluded in this interesting patient as well.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

References

- Jabeen SA, Sandeep G, Mridula KR, Meena AK, Borgohain R, Sundaram C. Adult-onset Leigh's disease: A rare entity. *Ann Indian Acad Neurol* 2016;19:140-2.
- Huertas-González N, Hernando-Requejo V, Luciano-García Z, Cervera-Rodilla JL. Wernicke's encephalopathy, wet beriberi, and polyneuropathy in a patient with folate and thiamine deficiency related to gastric phytobezoar. *Case Rep Neurol Med* 2015;2015:624807.
- Busani S, Bonvecchio C, Gaspari A, Malagoli M, Todeschini A, Cautero N, et al. Wernicke's encephalopathy in a malnourished surgical patient: A difficult diagnosis. *BMC Res Notes* 2014;7:718.
- Cameron JM, MacKay N, Feigenbaum A, Tarnopolsky M, Blaser S, Robinson BH, et al. Exome sequencing identifies complex I NDUFV2 mutations as a novel cause of Leigh syndrome. *Eur J Paediatr Neurol* 2015;19:525-32.
- Ferdinandusse S, Waterham HR, Heales SJ, Brown GK, Hargreaves IP, Taanman JW, et al. HIBCH mutations can cause Leigh-like disease with combined deficiency of multiple mitochondrial respiratory chain enzymes and pyruvate dehydrogenase. *Orphanet J Rare Dis* 2013;8:188.
- Blondon H, Polivka M, Joly F, Flourie B, Mikol J, Messing B. Digestive smooth muscle mitochondrial myopathy in patients with mitochondrial-neuro-gastro-intestinal encephalomyopathy (MNGIE). *Gastroenterol Clin Biol* 2005;29:773-8.
- Quinzii CM, DiMauro S, Hirano M. Human coenzyme Q10 deficiency. *Neurochem Res* 2007;32:723-7.

Monday, October 2, 2017

[Site Map](#) | [Home](#) | [Contact Us](#) | [Feedback](#) | [Copyright and Disclaimer](#)