Author's Reply

Reply to "Mitochondrial tRNA Glutamic Acid Variant 14709T>C Manifesting as Myoclonic Epilepsy with Ragged Red Fibers"

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Thank you for your attention and interest for our study,[1] and thank you for your questions and opinions as well.

Our center has carried out muscle biopsy for more than 30 years, and we have conducted more than 5400 cases during these years. Among all the biopsy cases, we diagnosed more than 120 mitochondrial myopathy/encephalomyopathy cases, but only one of them is myoclonic epilepsy with ragged red fibers (MERRF) harboring a novel mutation. In China, there are a total of 36 reported cases of MERRF,[2-7] which indicates that MERRF is rather rare. MERRF can be clinically diagnosed by myoclonus, seizures, cerebellar ataxia, and mitochondrial myopathy with ragged-red fibers; if m.A8344G mutation was found, MERRF would be perfectly diagnosed; however, even without gene mutations, MERRF can still be diagnosed by clinical presentations.

For mitochondrial myopathy/encephalomyopathy patients, we mainly collected their clinical presentations, muscle biopsy, and blood/muscle gene information; this also meets the interest of patients. The patient presented with obvious episode of epilepsy (absence seizure), myoclonus, and ataxia. Electroencephalograph (EEG) did not show epileptiform discharges, but it is not so unreasonable since there are almost 10–20% epileptic patients have normal EEG presentation. A complete checkup was done, and electrocardiogram and ultrasonic cardiogram showed normal findings; he did not really tell any histories of vomiting, profuse sweating, looking pale, feeling flustered, short of breath, dizziness, chest pain, angor pectoris, or cold in fingers and feet. Based on this, we say that he suffered definite epilepsy in clinical aspect but not any cardiac or autonomic nervous system problems.

The patient and his family members as well as previous doctors did not realize his episode of epilepsy; therefore, no treatment was given, but we then gave topiramate as an antiepileptic regimen, with no obviously curative effect shown.

Our patient had presented with red maculopapules on his face, double ears, and neck, which were itchy and blanch on pressure and were ulcerated and scabby; dermatologist considered them as allergic dermatitis and prescribed mizolastine, loratadine, and ketotifen; and the maculopapules almost disappeared after the treatment. Hence, we think his skin presentation was just nonspecific but not due to mitochondrial dysfunction.

Among 123 mitochondrial myopathy/encephalomyopathy cases in our center, there are three cases occurred in scoliosis, including two cases of mitochondrial myopathy and one case of MERRF – this indicates that scoliosis and skeleton deformity are rarely seen in myopathy/encephalomyopathy. Data from the Peking Union Medical College Hospital revealed that the prevalence of scoliosis among patients with mitochondrial myopathies was 5% (3/60), much higher than that among the general population (2%).[8]

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It is a pity that we did not get the genetic information from patient's mother. Hence, we would like to classify the variant as sporadic.

References

1. Ban R, Guo JH, Pu CQ, Shi Q, Liu HX, Zhang YT, *et al.* A novel mutation of mitochondrial T14709C causes myoclonic epilepsy with ragged red fibers syndrome in a Chinese patient. Chin Med J 2018;131:1569-74. doi: 10.4103/0366-6999.235120.
2. Chen G, Li W, Du WD, Cao HM, Tang HY, Tang XF, *et al.* Development of a DNA biochip for detection of known mtDNA mutations associated with MELAS and MERRF syndromes (in Chinese). Hereditas (Beijing) 2008;30:1279-86. doi: 10.3724/SP.J.1005.2008.01279.
3. Gao F, Li XD, Chen QT. Analysis of mitochondrial DNA A8344G point mutation in 6 cases with myoclonic epilepsy and ragged-red fibres disease (in Chinese). J Apoplexy Nerv Dis 2003;20:117-9.
4. Song D, Zhang Y, Shi J, Lü Q, Chen J, Zhang H, *et al.* Point mutations of muscle mitochondrial DNA from patients with mitochondrial encephalomyopathies. Chin Med J 2001;114:1273-5.
5. Zhao MM, Zhang Y, Bao XH. Myoclonus epilepsy with ragged-red fibers: A case report and literature review (in Chinese). J Peking Univ 2015;47:1034-6. doi: 10.3969/j.issn.1671-167X.2015.06.027.
6. Lu H, Wu LY, Ye J, Wang M, Liu H, Da YW, *et al*. A family of MERRF associated with scotosensitive seizures (in Chinese). J Brain Nerv Dis 2014;22:245-8.
7. Zhao J, Zhao DH, Zhang W, Lü H, Yuan Y, Qi Y, *et al.* Clinical heterogeneity associated with mitochondrial DNA A8344G point mutation (in Chinese). Natl Med J China 2012;92:2835-8.
8. Li Z, Shen J, Liang J. Scoliosis in mitochondrial myopathy: Case report and review of the literature. Medicine (Baltimore) 2015;94:e513. doi: 10.1097/MD.0000000000000513.