Biochemical and Biophysical Research Communications 435 (2013) 513

Contents lists available at SciVerse ScienceDirect

Biochemical and Biophysical Research Communications

journal homepage: www.elsevier.com/locate/ybbrc

Letter to the Editor

Importance of muscle biopsy in diagnosis of muscle diseases

Dear Editor,

We have read the paper titled "A novel m.12908T>A mutation in the mitochondrial ND5 gene in patient with infantile-onset Pompe disease" with great interest [1]. In this article, authors have described a patient with Pompe disease. We understand that the patient has complex clinical symptoms including hypertrophic cardiomyopathy, hypotonia and hepatomegaly. Authors said that this case has been misdiagnosed as mitochondrial disorder, previously. In further analysis, they have revealed that a novel mutation m.12908T>A in the ND5 gene of the mitochondrial DNA. They also have shown the deleterious effects of this mutation on the ND5 protein by secondary structure analysis. We think that this case report is interesting and the methods they utilized are scientifically proper. Here, we would like to add some constructive information as follows:

We think that the authors had no chance to get and examine a muscle biopsy. They have pointed out that this case has been diagnosed as a mitochondrial disease in the first place. To our knowledge, however, it is not that difficult to reach a correct diagnosis in Pompe disease if a muscle biopsy is available for histopathological examination. Since mitochondrial diseases have a completely different sets of morphological findings than those in the Pompe disease, muscle biopsy would have facilitated the diagnosis period. For instance, there are glycogen accumulation and increased acid phosphatase activity in muscle fibers, a finding which is not to be detected in any forms of mitochondrial diseases, in Pompe disease. Even if in the absence of such a characteristic morphological appearance, acid phosphatase-positive globules can be used as a good and a reliable diagnostic marker [2]. Instead, tubular aggregate myopathy can be misdiagnosed as mitochondrial myopathy, because of the accumulations reminiscent ragged red fibers in modified gomori trichrome stain [3].

Although it is an invasive procedure, muscle biopsy may provide indispensable information for the most types of muscle diseases.

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Available online 9 May 2013

☆ DOI of original article: http://dx.doi.org/10.1016/j.bbrc.2012.10.105

http://dx.doi.org/10.1016/j.bbrc.2013.05.021