Molecular Genetics and Metabolism Reports 13 (2017) 41

Contents lists available at ScienceDirect

Molecular Genetics and Metabolism Reports

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

Correspondence

Costs for mitochondrial medicine will remain high as long as mitochondrial disorders are misdiagnosed

ARTICLE INFO

Keywords:
Mitochondrial
Multiorgan
Epidemiology
Costs
Respiratory chain

Letter to the Editor

We read with interest the article by McCormack et al. about frequency and costs of hospitalisations of mitochondrial disorder (MID) patients in California [1]. There are several reasons why the figures provided are underestimations.

First, quite a number of MIDs go undetected or are misinterpreted as another disease. Particularly, patients with multiorgan disease are frequently in fact mitochondrial multiorgan disorder syndromes (MIMODSs) [2]. As soon as the cause of multisystem disease remains obscure, a MID should be suspected and considered as a differential diagnosis. Since work-up of suspected MID is time-consuming, logistically demanding, cost-intensive, and often associated with inconclusive or negative results, it is frequently not initiated at all, why many of these patients go undetected for years or forever.

Second, ICD codes do not cover the entire spectrum of MIDs. For example, MIRAS, LBSL, or PCH may be missed. Even ICD10 does not cover all specific and nonspecific MIMODS.

Third, coding of diagnoses is often insufficiently effectuated. Sometimes, only major diagnoses are encoded. Sometimes no ICD codes are allocated at all.

Fourth, a number of congenital MIDs may remain undiagnosed because patients decease during the first few days or months of life. During this short period it is often impossible to complete a comprehensive diagnostic work-up. Often these patients do not undergo autopsy.

Fifth, MIDs are often insufficiently diagnosed. According to various classification criteria, MIDs may be diagnosed as possible, probable, or definite [3]. Often MIDs are only diagnosed upon histochemical or biochemical investigations, without the inclusion of functional or genetic studies.

Accordingly, we do not agree with the figure 1:4300 for the prevalence of MIDs [1]. Nonspecific MIDs are regarded much more frequent occurring with a prevalence of 1:400 [4].

Overall, the costs in mitochondrial medicine will remain high if patients are misdiagnosed and thus mistreated. The longer a misdiagnosis is maintained, the more costs incur.

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☆ There are no conflicts of interest.
☆☆ Both authors contributed equally.
★ No funding was received.
★★ Author contribution: JF: design, literature search, discussion, first draft, SZ-M: literature search, discussion, critical comments.
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http://dx.doi.org/10.1016/j.ymgmr.2017.08.002
Received 1 August 2017; Accepted 1 August 2017
Available online 07 August 2017