



**HAL**  
open science

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

Josef Finsterer, Sinda Zarrouk-Mahjoub

### ► To cite this version:

Josef Finsterer, Sinda Zarrouk-Mahjoub. Costs for mitochondrial medicine will remain high as long as mitochondrial disorders are misdiagnosed. *Molecular Genetics and Metabolism Reports*, 2017, 13, pp.41. 10.1016/j.ymgmr.2017.08.002 . pasteur-02010612

**HAL Id: pasteur-02010612**

**<https://riip.hal.science/pasteur-02010612>**

Submitted on 7 Feb 2019

**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.



Distributed under a Creative Commons Attribution - NonCommercial - NoDerivatives | 4.0 International License



## 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 located at all.

Fourth, a number of congenital MIDs may remain undiagnosed because patients die 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.

## References

- [1] S.E. McCormack, R. Xiao, T.J. Kilbaugh, M. Karlsson, R.D. Ganetzky, Z.Z. Cunningham, A. Goldstein, M.J. Falk, S.M. Damrauer, Hospitalizations for mitochondrial disease across the lifespan in the U.S. *Mol. Genet. Metab.* 121 (2017) 119–126.
- [2] J. Finsterer, A. Bastovansky, Multiorgan disorder syndrome (MODS) in an octogenarian suggests mitochondrial disorder, *Rev. Med. Chil.* 143 (2015) 1210–1214.
- [3] F.P. Bernier, A. Boneh, X. Dennett, C.W. Chow, M.A. Cleary, D.R. Thorburn, Diagnostic criteria for respiratory chain disorders in adults and children, *Neurology* 59 (2002) 1406–1411.
- [4] J. Poulton, J. Finsterer, P. Yu-Wai-Man, Genetic counselling for maternally inherited mitochondrial disorders, *Mol. Diagn. Ther.* 21 (2017) 419–429.

Josef Finsterer  
Krankenanstalt Rudolfstiftung, Vienna, Austria

Sinda Zarrouk-Mahjoub  
University of Tunis El Manar and Genomics Platform, Pasteur Institute of Tunis, Tunisia  
E-mail address: [fifigs1@yahoo.de](mailto:fifigs1@yahoo.de)

\* 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.

\* Corresponding author at: Postfach 20, 1180 Vienna, Austria.