

Correspondence Unusual recovery of respiratory chain complex-III deficiency upon G-tube feeding and a cocktail

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Correspondence

Unusual recovery of respiratory chain complex-III deficiency upon G-tube feeding and a cocktail
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Letter to the Editor

With interest we read the article by Mori et al. about a 7 year old male with respiratory-chain complex-III-deficiency due to a 9 bp-deletion in the *cytb* gene, which initially manifested with failure to thrive, vomiting, diarrhea, constipation, weight-loss, fatigue, exercise-intolerance, muscle weakness, metabolic acidosis, and creatine-kinase elevation and who recovered almost completely by age 12 years [1]. We have the following comments and concerns.

In the majority of the cases mitochondrial disorders (MIDs) are progressive [2]. Thus, the marked clinical improvement is quite unusual. Unfortunately, the results of the clinical neurologic exam were not provided but obviously the patient initially had muscle weakness since he was wheelchair-bound [1]. How to explain complete recovery of weakness particularly with a heteroplasmy rate > 90% in the muscle? Why do the authors attribute recovery to G-tube-feeding and not to the cocktail of coenzyme-Q, L-carnitine, cornstarch, riboflavin, creatine-monohydrate, alpha-lipoic-acid, and medium-chain triglycerides? Is it conceivable that there was secondary coenzyme-Q-deficiency or carnitine-deficiency? Did consolidation of the phenotype persist even after discontinuation of G-tube-feeding and the cocktail? Was the

patient seen for follow-up after finishing treatment? Were coenzyme-Q levels reduced before substitution?

Nothing is reported about the clinical presentation of the mother and the patient's siblings [1]. Was the *cytb* deletion also found in the mother or any sibling? Were clinical manifestations of the index case variable from those of his siblings or his mother?

Did the patient ever experience a seizure or a stroke-like-episode? Was an EEG recorded? Were cognitive functions impaired? Did he have a history of migraine? Was there steatosis, cysts, or hepatomegaly?

Overall, this interesting case merits a broader discussion about the possible mechanisms of recovery and a more profound description of the phenotype and disease course. Furthermore, clinical and genetic data about his relatives are essential as well as follow-up data after discontinuation of G-tube-feeding and the cocktail.

References

- [1] M. Mori, J. Goldstein, S.P. Young, E.H. Bossen, J. Shoffner, D.D. Koeberl, Complex III deficiency due to an in-frame MT-CYB deletion presenting as ketotic hypoglycemia and lactic acidosis, *Mol Genet Metab Rep* 4 (2015) 39–41.
- [2] R. McFarland, R.W. Taylor, D.M. Turnbull, A neurological perspective on mitochondrial disease, *Lancet Neurol.* 9 (2010) 829–840.

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