



Correspondence

Mitochondrial myopathy plus due to the variant m.586G > A in MT-TF



To the Editor: Sir,

We have read with interest the Letter of Dr. J. Finsterer [1] regarding the report of Barcia et al. on a novel case of *de novo* MTTF mutation [2]. Our report was aimed at emphasizing the striking clinical differences between two individuals carrying the same mitochondrial MTTF mutation (m.586G > A), namely a childhood-onset form of mitochondrial myopathy [2] and a late-onset form of progressive neurodegenerative disorder with akinesia-rigidity, abnormal movements, dementia and psychiatric disorder in a 57 yrs-old woman [3]. Whether intrinsic mitochondrial genetic factors (such as heteroplasmy and mitotic segregation) or hitherto unknown nuclear factors accounted for those strikingly different presentations remains unanswered. Similarly, whether brisk deep tendon reflexes of our patient should be considered as “quadri-spasticity” and episodes of migraine-like headaches and vomiting episodes, be regarded as the hallmark of “CNS involvement” is also debatable, especially as our patient had consistently elevated plasma lactate, normal brain MRI and normal schooling.

Whatever the opinions, this issue offers the opportunity to recall that mitochondrial respiratory chain deficiency can give rise to virtually any symptom, in any organ or tissue, at any age, from the neonatal and even antenatal period [4] to late childhood and adulthood. In early-onset pediatric forms of the disease, the clinical expression of the primarily affected organ is at the forefront of the scene, possibly hiding CNS involvement (bone marrow in Pearson syndrome, liver in hepatic failure, heart in mitochondrial cardiomyopathy). In our pediatric cohort, CNS involvement frequently appears later in the course of the disease, especially in chronic forms or after transplantation of the life-threatening organ [5]. Also, as far as clinical management of the

children is concerned, putting forward CNS involvement at early stages of the disease inevitably leads to a form of discouragement that we do our best to postpone.

References

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