



HAL
open science

Mitochondrial vasculopathy due to the m.3243A > G mutation is not restricted to the carotid artery

Josef Finsterer, Sinda Zarrouk-Mahjoub

► **To cite this version:**

Josef Finsterer, Sinda Zarrouk-Mahjoub. Mitochondrial vasculopathy due to the m.3243A > G mutation is not restricted to the carotid artery. *Molecular Genetics and Metabolism Reports*, 2016, 8 (9), pp.34. 10.1016/j.ymgmr.2016.10.002 . pasteur-01457109

HAL Id: pasteur-01457109

<https://riip.hal.science/pasteur-01457109>

Submitted on 6 Feb 2017

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

Mitochondrial vasculopathy due to the m.3243A>G mutation is not restricted to the carotid artery


Keywords:

mtDNA
m.3243A>G
Carotid artery
Dissection
Arteriopathy
Gene
Mitochondrial disorder
Stroke-like episode

With interest we read the article by Mancuso et al. about two unrelated, adult females carrying the m.3243A>G mutation, in whom carotid-artery-dissection was regarded as a manifestation of the genetic defect [1]. We have the following comments and concerns.

Mitochondrial vasculopathy is well appreciated and may not only manifest as migraine, stroke-like-episode, rupture of the aorta, or carotid-artery-dissection or occlusion, but also as atherosclerosis, vascular leucoencephalopathy, vascular retinopathy, ectasia of the aortic-root or of cerebral vessels, or as aneurysm formation [2].

In addition to the present cases and the case by Ryther et al. 2011, mitochondrial carotid-artery-dissection was reported by Kalashnikova et al. 2012 [3] and by Sakharova et al. 2012 [4].

It is extremely unusual that cerebral MRI was normal, in the acute stage in patient-1 [1]. Were DWI, ADC, and PWI images carried-out? Usually, acute high-grade carotid-artery-stenosis or occlusion results in ischemic stroke with hyperintensity on DWI, hypointensity on ADC, hyperintensity on T2 and hypoperfusion of PWI. Was MRS carried out to see if CSF lactate was elevated?

Dissection is not convincing on conventional carotid angiography [1]. Were also T1-weighted transverse sections with contrast medium carried out to confirm the presence of two lumina? Could the occlusion be due to acute cardiac embolism? Were patients screened for cardiac involvement, frequently found in MELAS? Was ever paroxysmal atrial-fibrillation recorded on ECG? Was there noncompaction on echocardiography as a source of embolism? Patient-2 had bilateral dissection but only left-sided stroke [1]. How do the authors explain this discrepancy?

Concerning treatment of ischemic stroke, we should be informed if both patients underwent acute systemic thrombolysis, acute stenting, or mechanical thrombus extraction? Which was the outcome?

Overall, essential supplemental information is required to assess if stroke was truly ischemic and due to the dissection. The association between m.3243A>G and carotid-artery-dissection remains vague.

Conflict of interest

There are no conflicts of interest.

Contribution

Both authors contributed equally.

Funding

No funding was received.

References

- [1] M. Mancuso, V. Montano, D. Orsucci, L. Peverelli, L. Caputi, P. Gambaro, G. Siciliano, C. Lamperti, Mitochondrial m.3243A>G mutation and carotid artery dissection, *Mol. Genet. Metab. Rep.* 9 (2016) 12–14.
- [2] J. Finsterer, S. Zarrouk-Mahjoub, Mitochondrial vasculopathy, *World J. Cardiol.* 8 (2016) 333–339.
- [3] L.A. Kalashnikova, L.A. Dobrynina, A.V. Sakharova, R.P. Chaïkovskaia, M.A. Nazarova, M.F. Mir-Kasimov, N.L. Patrusheva, L.I. Patrushev, R.N. Konovalov, S.V. Protskiĭ, The A3243G mitochondrial DNA mutation in cerebral artery dissections, *Zh. Nevrol. Psikhiatr. Im. S S Korsakova* 112 (2012) 84–89.
- [4] A.V. Sakharova, L.A. Kalashnikova, R.P. Chaïkovskaia, M.F. Mir-Kasimov, M.A. Nazarova, T.N. Pykhtina, L.A. Dobrynina, N.L. Patrusheva, L.I. Patrushev, S.V. Protskiĭ, Morphological signs of mitochondrial cytopathy in skeletal muscles and microvessel walls in a patient with cerebral artery dissection associated with MELAS syndrome, *Arkh. Patol.* 74 (2012) 51–56.

Josef Finsterer MD, PhD
Krankenanstalt Rudolfstiftung, Vienna, Austria
Corresponding author at: Postfach 20, 1180 Vienna, Austria.
E-mail address: fipaps@yahoo.de.

Sinda Zarrouk-Mahjoub PhD
Genomics Platform, Pasteur Institute of Tunis, Tunisia

3 October 2016