East African Scholars Journal of Medical Sciences
Abbreviated Key Title: East African Scholars J Med Sci
ISSN 2617-4421 (Print) | ISSN 2617-7188 (Online) |
Published By East African Scholars Publisher, Kenya

OPEN ACCESS

Volume-2 | Issue-9| Sept -2019 | DOI: 10.36349/EASJMS.2019.v02i09.020

Letter to the Editor

Manifestations of Zaspopathy in Skeletal Muscle, Peripheral Nerves, and Heart

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Keywords: electromyography (EMG), ZASP, myotilin, αB-crystallin, hypertrabeculation / noncompaction.

In a recent article, Selcen and Engel's reported about 11 patients with Zaspopathy, presenting with distal, proximal, or diffuse muscle weakness (n=11), cardiac involvement (n=3), or neuropathy (n=5) (Selcen, D., & Engel, A.G. 2005). We have the following comments and concerns:

How can one be sure about peripheral nerve involvement when assessed only by electromyography (EMG) and not by nerve-conduction-studies? A neuropathic EMG may also occur in a myopathic patient (Zalewska, E. *et al.*, 2004). Which are the unequivocal, clinical and histological features of neuropathy that 5 patients had peripheral nerve involvement, although only 1 patient had a neurogenic but 8 patients a myogenic EMG? Was neuropathy axonal, demyelinating, or mixed, symmetric or asymmetric, motor, sensory, or autonomic, and were upper / lower limbs equally affected? Arguments for an exclusive affection of the skeletal muscle are the myogenic EMGs in 8 and the mixed neurogenic / myogenic EMGs in 2 patients. How severe was muscle weakness? Which were the cerebrospinal fluid (CSF)-findings in the patient with poly-radiculo-neuropathy and was there any indication for conduction-block? Was poly-radiculo-neuropathy diagnosed solely upon histological or EMG findings? Were causes other than ZASP mutations systematically excluded as causes of neuropathy?

In how many patients was the mutation found in muscle, blood, or both? How do the authors explain genetic heterogeneity of the phenotype resulting from ZASP, myotilin, αB-crystallin, or desmin mutations? Was affection of family members defined upon neurologic, cardiac or genetic findings? Concerning ZASP expression in the brain it is worthwhile to present results of imaging studies and to report if there was clinical evidence of encephalopathy.

Cardiologic findings are only scarcely presented and definition of cardiac involvement is lacking. Was history, clinical examination, ECG, 24h-ECG, and echocardiography carried out in all patients, which abnormalities did the authors look for, and which were the results? Did any of the patients have hypertrophic, dilative, or restrictive cardiomyopathy (Vatta, M. *et al.*, 2003)? Was left ventricular hypertrabeculation / noncompaction, previously described in association with ZASP mutations (Finsterer, J., & Stollberger, C. 2004), found in any patient? How many patients had symptomatic cardiac involvement? Did patient 3 with reduced ejection faction also suffer from heart failure? Did the patient with QT-prolongation, present with a history of syncope, vertigo, or palpitations? Why were cardiac abnormalities attributable to ZASP mutations and not to other causes? Were cardiovascular risk factors and concomitant systemic diseases assessed? The actual medication of any patient is not given. Drugs should be excluded as causes of the cardiac findings.

To assess degree and type of muscle, nerve, and myocardial involvement in ZASP-mutations, more detailed and thorough investigations are warranted.

Quick Response Code

Journal homepage:
http://www.easpublisher.com/easjms/

Article History
Received: 24.08.2019
Accepted: 05.09.2019
Published: 19.09.2019

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