

## Muscle Weakness in CPEO Patients May Not only be Determined by Myopathy Smoking or Gender

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### 1. Editorial

In a recent article, Heighton et al. [1] Reported about the influence of sex and smoking on knee-extension strength in 116 patients with CPEO [1]. We have the following comments/concerns.

The study has several shortcomings. The first is that muscle strength not only depends on the presence/absence of a neuromuscular disorder (NMD), sex, and smoking, but on a number of other factors, such as metabolic status (diabetes, lactate, pyruvate levels, cholesterol, and thyroid/parathyroid, suprarenal, and pituitary function), on the training status, cardiac functions, and on the innervation of the muscles. How many of the included patients performed regularly sport, went to the fitness studio, or had a physically demanding job? How many patients had cardiomyopathy or systolic dysfunction? Occasionally, cardiomyopathy is a phenotypic feature of CPEO [2].

In a study of 15 CPEO patients one had cardiomyopathy [3]. Which were the pro BNP values in the 116 patients? Systolic dysfunction/heart failure can be phenotypic manifestations of the underlying genetic defect [4] was neuropathy, plexopathy, radiculopathy, and neuronopathy excluded in all 116 patients? How many had a previous stroke?

A second shortcoming is that the genetic defect of the 116 patients has not been reported. CPEO may not only be due to single mtDNA deletions or mtDNA point mutations but also due to multiple mtDNA deletions or mtDNA depletion [5]. Multiple mtDNA deletions or mtDNA depletion may be due to mutations in nDNA located genes, such as *POLG1*, *POLG2*, *C10orf2* (*TWINK*), *SL-C25A4*, *TFAM*, *RNASEH1*, *MGME1*, *DNA2*, *TK2*, *DGUOK*, *SUCLG1*, *SUCLA2*, *ABAT*, *RRM2B*, *TYMP*, *AGK*, *MPV17*, *OPA1*, *MFN2*, or *FBXL4*. How many had a single deletion, how many a mutation in a nuclear gene?

A third shortcoming is the retrospective design. How was it guaranteed that muscle strength was measured consistently in each patient? To which degree did investigational procedures influence the study results? Data were collected over a period of 11y.

A fourth shortcoming is that smoking was not encountered in controls, which prevents comparison between CPEO and controls regarding smoking. Was the prevalence of atherosclerosis/stroke different between CPEO smokers and CPEO non-smokers?

Fifth, reduced strength in females could be attributed to a more advanced stage of the disease in females, to higher heteroplasmy rates, or to mutant genes different between females and males.

Overall, this study would profit from assessing all determinants of muscle strength, from providing the genetic cause of CPEO, and from a prospective design. The conclusion that smoking determines muscle strength in female CPEO patients is not justified.

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