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Letter to the Editor

Causes of Lower Urinary Tract Symptoms In Mitochondrial Disorders May Be Complex

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In a recent article, Poole *et al.*, reported about a case control study of 58 patients with a mitochondrial disorder (MID), which was genetically confirmed in 91%, for urogenital manifestations (Poole, O. V. *et al.*, 2019). Fours scores (USP, FSQ, ASEX, and NBD) were applied (Poole, O. V. *et al.*, 2019). Mutation carriers had more frequently an over-reactive bladder and low stream urinary symptoms than controls, and female carriers had more frequently sexual dysfunction than controls (Poole, O. V. *et al.*, 2019). We have the following comments and concerns.

The main shortcoming of the study is that heteroplasmy rates were determined in blood lymphocytes and not in urinary epithelial cells. We thus, do not agree with the notion that disease burden (heteroplasmy rate, mutation load) does not influence lower urinary tract symptoms (LUTS) (Poole, O. V. *et al.*, 2019). More appropriate for correlating disease burden with LUTS is heteroplasmy rate in cells from organs clinically affected than from organs which are not clinically affected at all. Furthermore, heteroplasmy rates were available in only 16/58 patients.

A further shortcoming is that it was not mentioned if the mutation carriers were clinically affected or not and if affected how severely they were affected. This point is crucial since cerebral or spinal involvement in the disease may be responsible for LUTS. We thus should know in how many of the included patients a cerebral or spinal MRI was carried out and in how many of them a cerebral lesion could explain LUTS. Differences in the SFQ score could be attributed to affection of organs other than the urogenital organs.

Since patients carrying the m.3243A>G mutation frequently manifest with endocrine abnormalities such as hypopituitarism, hypothyroidism, hypoparathyroidism, diabetes, hypocorticism, or hypogonadism, we should know if appropriate hormone levels were determined and if they were reduced in any of the patients with LUTS. Did females with sexual dysfunction more frequently suffer from hypogonadism than those who did not report sexual dysfunctions.

Another shortcoming of the study is that questionnaires were used to assess LUTS. Since patients carrying the m.3243A>G mutation may manifest with cognitive impairment or psychiatric disease (Salsano, E. *et al.*, 2011), the answers may not always be reliable. We thus should know in how many of the 52 patients cognitive impairment and in how many psychiatric disease was present.

Another shortcoming of the study is that the included patients did not carry the same mtDNA variant and that some patients were even included that did not carry any mutation at all. We thus should know how the authors in the latter cases knew that they had indeed a MID. Furthermore, the variants detected in the patient with a genetic cause should be mentioned to assess if these variants were truly pathogenic.

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In the method section it is indicated that the current medication was registered but nothing is mentioned about the potential relation to LUTS. We thus should know the co-medication the included patients were taking. Did those with LUTS take more tablets than those without LUTS or tablets with known urogenital side effects?

Overall, this interesting study would be more meaningful if heteroplasmy rates were determined in urinary epithelial cells, if it was mentioned if the patients were clinically manifesting or not, if hormone levels were determined and correlated with LUTS, and if LUTS was assessed more objectively.

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