

Mitochondrial disease

Bowel symptoms are well documented in mitochondrial disease. However, data concerning other pelvic organs is limited. We have undertaken a large, case-control study to determine the presence of lower urinary tract symptoms (LUTS) and sexual dysfunction in adults with genetically confirmed mitochondrial disease.

METHODS: Adults with genetically confirmed mitochondrial disease and control subjects were recruited from a specialist mitochondrial clinic. The presence and severity of LUTS and their impact on quality of life, in addition to sexual dysfunction and bowel symptoms, were captured using four validated questionnaires. Subgroup analysis was undertaken in patients harbouring the m.3243A>G MT-TL1 mitochondrial DNA (mtDNA) mutation. A subset of patients underwent urodynamic studies to further characterise their LUTS.

RESULTS: Data from 58 patients and 19 controls (gender and age matched) was collected. Adults with mitochondrial disease had significantly more overactive bladder (81.5% versus 56.3%, $p=0.039$) and low stream (34.5% versus 5.3%, $p=0.013$) urinary symptoms than controls. Urodynamic studies in 10 patients confirmed that bladder storage symptoms predominate. Despite high rates of LUTS, none of the patient group was receiving treatment. Female patients, and those harbouring the m.3243A>G MT-TL1 mutation, experienced significantly more sexual dysfunction than controls (53.1% versus 11.1%, $p=0.026$ and 66.7% versus 26.3%, $p=0.011$, respectively).

LUTS are common but undertreated in adult mitochondrial disease, and female patients and those harbouring the m.3243A>G MT-TL1 mutation experience sexual dysfunction. Given their impact on quality of life, we recommend screening and treating LUTS and sexual dysfunction in adults with mitochondrial disease ¹⁾.

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