80 Lower urinary tract symptoms amongst adult patients with genetically-confirmed mitochondrial disease

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Introduction & Objectives: Mitochondrial diseases are a rare group of genetic disorders leading to mitochondrial dysfunction, associated with a spectrum of clinical features due to multi-organ involvement. However, there remains a paucity of data on bladder function in this patient cohort. Since mitochondrial diseases are often characterised by a loss of smooth muscle function, it is plausible to hypothesise that mitochondrial dysfunction may also contribute to bladder dysfunction. A recent report focussing on patients with mitochondrial disease (m.3243 mutation) presenting with intestinal pseudo-obstruction (Ng et al, Annals of Neurology 2017) noted a concomitant incidental finding of urinary retention in 20% of patients, in support of this hypothesis. We therefore aimed to comprehensively evaluate lower urinary tract symptoms (LUTS) amongst patients with mitochondrial disease.

Materials & Methods: Adult patients with genetically-confirmed mitochondrial diseases were recruited from a specialist clinical service. Patients were asked to complete an ICIQ-LUTS questionnaire and a three-day bladder diary. Outpatient investigations included measurement of bladder voiding efficiency (BVE), serum urea and electrolytes, and urinalysis. Patients with ICIQ-LUTS severity > 60% of maximum score, BVE <70% or nocturnal polyuria index (NPI) > 35% were offered urodynamic studies, if clinically indicated.

Results: Between 31st October 2017 and 30th June 2018, 65 patients were recruited (42 females, median age 52.5 years), amongst which 61.5% carried the m.3243A>G mutation. The median ICIQ-LUTS severity score was 24.6% of maximum. High correlation between ICIQ-LUTS severity and bother scores was noted (R 0.84, $p < 0.001$). Female patients reported higher scores for storage and incontinence domains, whereas male patients reported higher scores for the voiding domain of the ICIQ-LUTS questionnaire ($p < 0.001$). Amongst female patients, ICIQ-LUTS severity score correlated with mitochondrial disease symptom severity scores (R 0.41, p 0.017). Reduced bladder voiding efficiency was noted amongst 18.1% patients. Evaluation of bladder diaries noted increased daytime urinary frequency amongst 5.7% patients (> 8x voids/day), and nocturia (>2x voids/night) amongst 39.6% of all patients, with 13.2% patients found to have nocturnal polyuria. Twenty percent of patients met our criteria for bladder dysfunction requiring onward urodynamic investigation.

Conclusions: This is the first study to evaluate LUTS in patients with mitochondrial disease, and finds that LUTS are under-recognised amongst this patient cohort. Patients with mitochondrial disease should therefore be routinely screened for LUTS, and specialist urological input sought where appropriate.