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URO-NEUROLOGICAL ASSESSMENT OF HUNTINGTON'S DISEASE

Hypothesis / aims of study

Huntington's disease (HD) is an autosomal dominant neurodegenerative disease, which is caused by an extended CAG repeat chain located within the IT15 gene on the short arm of chromosome 4. The clinical characteristics of HD include involuntary movement, behavioral abnormalities, personality changes, and dementia. In addition to motor and cognitive impairments, some patients show micturition symptoms, especially urinary incontinence. Thus we aimed to examine the micturition disorder of HD in this study.

Study design, materials and methods

We enrolled 5 patients (3 males, 2 females; mean age 49±7.7 years; mean duration of the disease 10.6±8.3 years) who met the diagnostic criteria for definite HD [1], all of whom were referred to our urodynamic laboratory for uro-neurological assessment. All patients had undergone brain magnetic resonance imaging or CT scanning that showed atrophy of the caudate nucleus. No patients had urological complications such as prostate hypertrophy, physical stress incontinence, or urinary tract infection. We administered a urinary symptoms questionnaire and performed an urodynamic study in all patients. In case 1, only free flowmetry and measurement of post-void residuals (PVR) could be performed because of urethral pain.

Results

The urinary symptoms questionnaire revealed that all 5 patients had lower urinary tract symptoms (LUTS). LUTS started after the onset of motor disorders in all patients; the average duration of LUTS in these patients was 3.2±1.9 years. LUTS included nocturnal urinary frequency in 4 patients (80%), urinary urgency in 3 patients (60%), urge urinary incontinence in 2 patients (40%), and voiding difficulty in 2 patients (40%). Measurement of PVR showed that none had large PVR (Table 1). In the filling phase of the cystometry, 2 patients had detrusor overactivity, and one patient had increased bladder sensation in the filling phase but without detrusor overactivity. In the voiding phase, 3 patients had decreased maximum flow rates and one patient showed weak detrusor contractility (Table 2). No patients had obvious urethral obstruction.

Interpretation of results

In the present study, our patients with HD had both storage and voiding disorders, with storage disorders being the most prominent. Urinary incontinence was noted in 3 of 5 patients. Neurogenic storage disorders are thought to be caused by three major mechanisms: 1) suprapontine lesions (such as basal ganglia and cerebral cortex) that normally suppress the micturition reflex; 2) spinal cord lesions below which novel sacral reflex emerges; and 3) irritation in the peripheral nerve/muscles. In patients with HD, the central pathology includes the basal ganglia (the striatum, e.g., the caudal nucleus and the putamen). The striatum is thought to have a significant role in the central control of micturition. In patients with PD, in which nigrostriatal fibers are degenerated, storage symptoms such as nocturnal frequency, urinary urgency, and urinary incontinence are commonly found [2]. Functional brain imaging studies have shown a close relation between the basal ganglia and micturition [3]. Therefore, it is thought that the striatum tonically suppresses the micturition reflex. In patients with HD, degeneration in the striatum may lead to not only motor disorder but also detrusor overactivity and increased bladder sensation.

Concluding message

Our HD patients had urinary storage dysfunction. The underlying mechanism for the urinary dysfunction seems to be degeneration in the striatum.

References

1) Neurology (1986) 36:1279-1283.
2) Auton Neurosci (2001) 92:76-85.
3) J Comp Neurol (2005) 493:27-32.

Table 1 Results of the questionnaire

Patient	Nocturnal frequency	Urinary urgency	Urgency incontinence	Voiding difficulty	Post-void Residuals (ml)*
1	4	+++	+++	—	10
2	2	—	—	—	5

3	1	+	+++	—	0
4	2	+	—	+	30
5	3	+++	+++	+++	0

+: monthly
++: weekly
+++: daily
*: normal<30 ml

Table 2 Results of the urodynamic study

Patient	Q max (ml/s)	FS (ml)	BC (ml)	DO	DSD	Urethral obstruction*	Detrusor contractility*
1	3	np	np	np	np	np	np
2	4	213	454	—	—	2	weak
3	38	270	420	+	—	0	normal
4	np	111	119	—	—	np	np
5	4	50	220	+	—	1	normal

Qmax: maximum flow rates
FDV: first desired volume MDV: maximum desired volume
>FS: first sensation, BC: bladder capacity
DH: detrusor hyperactivity >DO: detrusor overactivity
DSD: detrusor-sphincter dyssynergia
np: not performed

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