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Male sexual function in presymptomatic gene carriers and patients with Huntington's disease

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ABSTRACT

Aims: To report sexual dysfunction in a systematically studied cohort of men with Huntington's disease (HD), and compare them with control men of a similar age.
Methods: In men with HD and asymptomatic HD gene carriers, the male sexual dysfunction questionnaire (International Index of Erectile Function – IIEF, covering erectile and orgasmic function, sexual desire, intercourse satisfaction and overall satisfaction), neurologic assessment using the Unified Huntington's Disease Rating Scale (UHDRS) and the Total Functional Capacity (TFC) Score were utilized.
Results: Responses were obtained from 23 HD patients and 2 HD gene carriers. HD patients reported more problems with erection, intercourse satisfaction and overall satisfaction ($p < 0.05$) compared to 41 controls. HD patients generally reported reduced sexual desire and performance. Sexual dysfunction progressed in parallel with patients' decline in motor (UHDRS) and TFC, but was not related to patients' age and duration of disease.
Conclusions: Our study demonstrated a significant impact of HD on male sexual function that progressed in parallel with motor and total patient (TFC) dysfunction. Physicians helping HD patients should also consider this largely neglected aspect of the disease.

1. Introduction

Although Huntington's disease (HD) most prominently affects patients' movement, cognition, and personality, symptoms of autonomic dysfunction [1] and specifically, lower urinary tract dysfunction [2], are also common. However, in contrast to Parkinson's disease (PD) [3] and multiple system atrophy (MSA) [4], data obtained in larger, systematically studied populations of HD patients are limited. It is well recognized that chronic neurologic diseases, and particularly neurodegenerative disorders affecting the basal ganglia, may also affect sexual function. Furthermore, HD patients often complain of sexual problems even at the onset of the disease [5]. Although the age of onset of HD depends primarily on the number of CAG triplets, in the majority of patients the disease starts during their reproductive period (i.e., peak incidence at 38–45 years [6]).

Nevertheless, only a few studies of sexual dysfunction in HD have been published. Furthermore, the diagnosis of HD was confirmed by genetic testing in only half of the published studies [7]. George Huntington described hypersexuality in two men with HD [8]. Other early studies also reported hypersexuality more frequently than hyposexuality (18.6% vs. 10.8%, respectively) [9]. By contrast, in newer studies, up to 85% of men reported sexual problems, with reduced sexual desire, inhibited orgasm [10] and problems with erection or ejaculation [1] being the most common. Only 5–30% of HD men in these newer studies reported hypersexual or sexually demanding behaviors [10,11]. However, none of the published studies correlated sexual (dys)function with the HD stage and functional disability (TFC) obtained by the United Huntington's Disease Rating Scale (UHDRS) [12].

The aim of the present study was therefore to present sexual (dys)function in a population of genetically confirmed HD men and asymptomatic HD gene carriers using the International Index of Erectile Function (IIEF) [13]. The findings in HD patients were compared to control men of similar age. We hypothesized that sexual dysfunction is more prevalent in men with HD than in controls. We also hypothesized that sexual dysfunction progresses in parallel with HD.

Abbreviations: HD, Huntington's disease; MSA, multiple system atrophy; PD, Parkinson's disease; SD, standard deviation; TFC, total functional capacity; TMS, Total Motor Score; UHDRS, United Huntington's Disease Rating Scale; IIEF, International Index of Erectile Function; SSRI, selective serotonin reuptake inhibitors.

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Table 1
General data on 23 men with Huntington's disease (HD) and 41 controls included in the study.

	Men with HD	Control group
Number	23	41
Age (years)	46 (25–68)	42 (25–64)
Disease (years)	4 (0–15)	/
Number of CAG triplets	44 (40–51)	/
TMS	36 (7–79)	/
TFC	10 (2–13)	/
BECK depression score	4 (0–33)	0.5 (0–21)

Except for numbers of subjects, and age (mean), median values (ranges) are presented. All questionnaires/tests were administered to all subjects, except Beck's depression questionnaire, which was administered to 18 men with HD during follow-up examination. The groups were not statistically different in age ($p = 0.25$). TFC — total functional capacity; TMS — Total Motor Score (motor part of the Unified Huntington's Disease Rating Scale — UHDRS).

2. Materials and methods

Men from our inventory of patients with genetically proven HD (inclusion criterion: ≥ 38 CAG repeats in the huntingtin gene [7]) were asked to participate in a study assessing sexual function during their regular outpatient visit or by written invitation. Patients not able to assist with toileting (e.g., bedridden) were excluded (exclusion criterion). A control group of men of similar age was also recruited (Table 1). Control subjects with disorders possibly affecting sexual function (e.g., spinal injuries, severe diabetes), and severely ill subjects (e.g., heart failure, active cancer, liver cirrhosis) were also excluded (exclusion criteria). The National Ethics Committee of Slovenia approved the study that has therefore been performed in accordance with the ethical standards laid down in the 1964 Declaration of Helsinki and its later amendments. After a comprehensive explanation of the study protocol, all included patients and controls provided written informed consent.

2.1. Sexual (dys)function questionnaire in men

We asked all studied HD patients about erectile and ejaculatory dysfunction. Afterwards, the patients answered the validated Slovene translation of the International Index of Erectile Function (IIEF) [13]. The responses to 15 questions of the IIEF were scored using five or six point scales, with better sexual function scoring higher. The first ten questions (Q1–10) were scored 0–5, and the last five questions (Q11–15) scored 1–5. Six questions (Q1–5, Q15) concerned erectile function, three (Q6–8), intercourse satisfaction, two (Q9–10), orgasmic function, two (Q11–12), sexual desire, and two (Q13–14) concerned overall satisfaction (Tables 2 and 3). Each area of sexual activity was first scored separately, and then a general score of sexual function was obtained by summation of the individual scores. Sexual dysfunction in the individual domains and the general score were described as absent, mild, moderate, or severe sexual dysfunction (Fig. 1).

Because the IIEF questionnaire is not designed for hypersexual and psychiatric dysfunction, we also asked all HD patients about hypersexual behavior.

2.2. Neurological evaluation

The Total Motor Score (TMS) part of UHDRS [12] was determined in all patients (Table 1). According to the TMS (range: 0–124 points), HD patients were divided into four groups: (1) presymptomatic HD gene carriers (0–4 points); (2) early HD patients (5–24 points); (3) intermediate HD patients (25–49 points); and (4) late HD patients (≥50 points) [12].

Table 2
Sexual function in 23 men with Huntington's disease (HD) and 41 controls.

Severity score	Attempts (%)	Satisfaction (%)	Enjoyment (%)	Ejaculation (%)	Orgasm (%)	Desire freq. (%)	Desire level (%)	Overall satisfaction (%)	Partner satisfaction (%)
0	35/7	35/10	35/7	17/10	4/10	-	-	-	-
1	26/22	0/2	9/2	0/5	17/2	17/2	9/2	26/5	22/7
2	17/22	4/5	4/2	4/5	4/2	0/12	17/10	0/7	4/2
3	4/15	13/5	4/12	9/5	9/2	26/12	35/29	9/10	9/10
4	4/17	22/17	39/48	4/12	4/12	22/24	35/37	39/39	35/37
5	13/17	26/61	9/27	65/63	61/71	35/49	4/22	26/39	30/44
p-Value	**0.01**	**0.004**	**0.005**	0.928	0.383	0.184	**0.045**	0.140	0.054

Sexual function was studied using the International Index of Erectile Function (IIEF) questionnaire that evaluates sexual activity in the previous four weeks. The frequency of attempts to have sexual intercourse (Q6), satisfaction (Q7), and enjoyment (Q8) with sexual intercourse were used to evaluate intercourse satisfaction. Frequency of ejaculation (Q9) and frequency of a feeling of orgasm (Q10) evaluated ejaculatory function. Frequency (Q11) and level (Q12) of sexual desire evaluated that domain of sexual life. Overall satisfaction with sexual life (Q13) and satisfaction with sexual relationship with partner (Q14) evaluated overall sexual satisfaction. Note that Q6–10 were scored 0–5 and Q11–14 were scored 1–5. Better sexual function scored higher. In Q7, Q9, Q10, and Q11, the scale was the same as in Q1–5, and in Q12 the scale was the same as in Q15. Q6 was scored as follows: 0, no attempts; 1, one to two attempts; 2, three to four attempts; 3, five or six attempts; 4, seven to ten attempts; 5, eleven or more attempts; Q8 was scored as follows: 0, no intercourse; 1, no enjoyment; 2, not very enjoyable; 3, fairly enjoyable; 4, highly enjoyable; 5, very highly enjoyable; Q13 and Q14 were scored as follows: 1, very dissatisfied; 2, moderately dissatisfied; 3, about equally satisfied and dissatisfied; 4, moderately satisfied; 5, very satisfied.

Table 3
Erectile function in 23 patients with Huntington's disease (HD) and 41 controls.

Score	Frequency (%)	Quality (%)	Penetration (%)	Maintenance (%)	Completion (%)	Confidence (%)
0	30/10	35/10	35/10	35/10	35/10	4/0
1	4/2	4/5	0/2	4/2	0/2	17/2
2	0/5	0/2	9/2	9/2	0/2	4/7
3	17/5	13/2	9/5	13/2	13/2	35/20
4	13/2	9/5	9/0	13/12	17/7	26/34
5	35/76	39/76	39/81	26/71	37/76	22/37
p-Value	**0.003**	**0.004**	**0.001**	< **0.001**	**0.002**	0.280

Erectile function was studied using the International Index of Erectile Function (IIEF) questionnaire. Six responses in this questionnaire inquire about erectile dysfunction: frequency of obtaining an erection (question 1, Q1), having an erection hard enough for penetration (Q2), ability to penetrate the partner (Q3), ability to maintain erection after penetration (Q4), and difficulty in maintaining erection until completion of intercourse (Q5), together with confidence to achieve and maintain an erection (Q15). Q1–5 were scored 0–5 (0, no sexual activity; 1, almost never/never; 2, rarely; 3, sometimes; 4, usually; 5, almost always/always). Q15 was scored 1–5 (1, very low; 2, low; 3, moderate; 4, high; 5, very high). In the IIEF, all questions evaluate sexual activity in the previous four weeks. Better sexual function scored higher.
Significant p -values ($p < 0.05$) are printed in bold.