Psychological characteristics of patients with myotonic dystrophy type 1.

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Abstract

OBJECTIVES: Myotonic dystrophy type 1 (DM1) is the most common adult-onset muscular dystrophy. It is associated with motor symptoms but patients also display non-motor symptoms such as particular personality traits. Studies have reported mixed results about personality characteristics which may be attributable to small sample sizes, different disease severity of groups studied, and use of different questionnaires or method. This study aimed to describe the psychological characteristics of a large cohort of patients with DM1, to characterize those at risk of developing a psychiatric disorder, and to compare characteristics between two DM1 phenotypes, a mild and more severe adult-onset phenotype.

METHODS: Two hundred patients with DM1 (152 adult-onset; 48 mild) were asked to complete questionnaires assessing personality traits, psychological symptoms, self-esteem, and suicidal risk. Neurological and neuropsychological assessments were performed to compare personality characteristics to clinical and cognitive measures.

RESULTS: Patients with DM1 globally showed personality traits and psychological symptoms in the average range compared to normative data, with normal levels of self-esteem and suicidal ideation. However, 27% of patients were found to be at high risk of developing a psychiatric disorder. Moreover, psychological traits differed across phenotypes, with the most severe phenotype tending to show more severe psychological symptoms. The presence of higher phobic anxiety and lower self-esteem was associated with lower education, a higher number of CTG repeats, more severe muscular impairment, and lower cognitive functioning (P < 0.001).

CONCLUSIONS: Different phenotypes should thus be taken into account in clinical settings for individual management of patients and optimizing therapeutic success.

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KEYWORDS: adult-onset myotonic dystrophy type 1; mild myotonic dystrophy type 1; myotonic dystrophy type 1; personality traits; psychological symptoms

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Publication Types, Grant Support


2. **Comparisons of intellectual capacities between mild and classic adult-onset phenotypes of myotonic dystrophy type 1 (DM1).**

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Author information

Abstract

**BACKGROUND:** Myotonic dystrophy type 1 (DM1) is an autosomal dominant genetic multisystem disorder and the commonest adult-onset form of muscular dystrophy. DM1 results from the expansion of an unstable trinucleotide cytosine-thymine-guanine (CTG) repeat mutation. CTG repeats in DM1 patients can range from 50 to several thousands, with a tendency toward increased repeats with successive generations (anticipation). Associated findings can include involvements in almost every systems, including the brain, and cognitive abnormalities occur in the large majority of patients. The objectives are to describe and compare the intellectual abilities of a large sample of DM1 patients with mild and classic adult-onset phenotypes, to estimate the validity of the Wechsler Adult Intelligence Scale-Revised (WAIS-R) in DM1 patients with muscular weakness, and to appraise the relationship of intelligence quotient (IQ) to CTG repeat length, age at onset of symptoms, and disease duration.

**METHODS:** A seven-subtest WAIS-R was administered to 37 mild and 151 classic adult-onset DM1 patients to measure their Full-Scale (FSIQ), Verbal (VIQ) and Performance IQ (PIQ). To control for potential bias due to muscular weakness, Standard Progressive Matrices (SPM), a motor-independent test of intelligence, were also completed.

**RESULTS:** Total mean FSIQ was 82.6 corresponding to low average IQ, and 82% were below an average intelligence. Mild DM1 patients had a higher mean FSIQ (U=88.7 vs 81.1, p<0.001), VIQ (U=87.8 vs 82.3, p=0.001), and PIQ (U=94.8 vs 83.6, p<0.001) than classic adult-onset DM1 patients. In both mild and classic adult-onset patients, all subtests mean scaled scores were below the normative sample mean. FSIQ also strongly correlate with SPM (r s =0.67, p<0.001), indicating that low intelligence scores are not a consequence of motor impairment. FSIQ scores decreased with both the increase of (CTG)n (r s =-0.41, p<0.001) and disease duration (r s =-0.26, p=0.003).

**CONCLUSIONS:** Results show that intellectual impairment is an extremely common and
important feature in DM1, not only among the classic adult-onset patients but also among the least severe forms of DM1, with low IQ scores compared to general reference population. Health care providers involved in the follow-up of these patients should be aware of their intellectual capacities and should adapt their interventions accordingly.

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Publication Types, MeSH Terms, Grant Support


Health supervision and anticipatory guidance in adult myotonic dystrophy type 1.


Collaborators (15)

Author information

Abstract
The complexity and variability of disease manifestations in myotonic dystrophy (DM1) pose a challenge for the clinical management of patients. The follow-up of DM1 patients has been described as fragmented, inadequate or even deficient for many patients. Through a systematic review of the medical and social literature and a validation process with a DM1 expert panel, we summarized systemic and social concerns clinically relevant to DM1 and revisited recommendations for treatment. This article summarizes common manifestations of the central nervous system, visual, respiratory, cardiac, gastro-intestinal, genito-urinary, muscular and metabolic impairments. In addition, we emphasized the social features of DM1 such as low education attainment, low employment, poor familial and social environment and poor social participation. While cardiac, respiratory and swallowing problems affect life expectancy, it is often excessive daytime sleepiness, fatigue, gastro-intestinal and cognitive behavioural manifestations that are the most disabling features of the disorder. A more holistic approach in the management of DM1 and a purposeful integrated organization of care involving all members of the patients' environment including family, clinicians, decision-makers and community organizations are needed to move out of the spiral of disease and handicap and move toward optimal citizenship and quality of life.

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Clinical and genetic knowledge and attitudes of patients with myotonic dystrophy type 1.


Author information

Abstract

AIMS: The goal was to assess clinical and genetic knowledge and attitudes in patients affected by myotonic dystrophy type 1 (DM1).

METHODS: Two hundred patients with molecular confirmation of the diagnosis of DM1 completed a multi-choice questionnaire. DM1 patients' knowledge and views were compared to clinically normal DM1 noncarriers (n = 264) and controls (n = 1,474).

RESULTS: Knowledge of the DM1 mode of inheritance was better in noncarriers than in patients (p < 0.001). Noncarriers were more aware than DM1 patients of the common clinical characteristics of DM1 such as limitations in physical activities and problems related to employment, schooling, activities of daily living, parenthood, peer relationships, and personality (p < 0.001). Compared to controls, DM1 patients felt less informed about the availability of clinical genetic services (p < 0.05) and new genetic technologies (p < 0.001). Among patients, logistic regression revealed that each additional year of education (p < 0.05) and each additional 100 CTG repeats (p < 0.01), respectively, increased and decreased the odds of knowing the DM1 mode of inheritance by about 23% and 18% respectively, independently of age, age at onset of symptoms, gender, severity of muscular impairment, and intellectual quotient.

CONCLUSIONS: DM1 patients' genetic knowledge is significantly dependent of the level of education and the number of CTG repeats. Healthcare providers should be aware of this situation in order to adjust counselling and education accordingly.

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5. **Fatigue and daytime sleepiness in patients with myotonic dystrophy type 1: to lump or split?**

Laberge L, Dauvilliers Y, Bégin P, Richer L, Jean S, Mathieu J.

**Author information**

**Abstract**

We assessed the relationship and clinical correlates of fatigue and Excessive Daytime Sleepiness (EDS) in 200 myotonic dystrophy type 1 (DM1) patients by means of questionnaire and neuropsychological evaluation. Fatigue levels were higher in patients with EDS and daytime sleepiness levels higher in patients with excessive fatigue. However, EDS without fatigue was rarely observed. Also, DM1 patients with fatigue (with or without EDS) showed greater muscular impairment, CTG repeats, abnormalities regarding personality, depressive symptoms and lower health-related quality of life (HRQoL) than patients without these symptoms. These findings do not readily support the contention that fatigue and EDS constitute distinct clinical manifestations in DM1. Clinicians should systematically evaluate both symptoms since fatigue and EDS have a greater impact on HRQoL than fatigue alone. However, specific rating scales for fatigue in DM1 have yet to be devised.

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**Publication Types, MeSH Terms**


6. **A polysomnographic study of daytime sleepiness in myotonic dystrophy type 1.**

Laberge L, Bégin P, Dauvilliers Y, Beaudry M, Laforte M, Jean S, Mathieu J.

**Author information**

**Abstract**

**OBJECTIVES:** To assess contributors to excessive daytime sleepiness (EDS) in myotonic dystrophy type 1 (DM1), to characterise subjects with sleep-onset REM periods (SOREMPs), and to verify whether self-reported instruments and respiratory function tests can predict multiple sleep latency test (MSLT) and sleep-disordered breathing.

**METHODS:** A sample of 43 DM1 patients without selection bias underwent polysomnography (PSG) for two consecutive nights and MSLT, completed a sleep diary and Epworth Sleepiness Scale (ESS), and were assessed for respiratory function and narcolepsy symptoms.

**RESULTS:** ESS scores (ES) > or =11 and MSLT mean sleep latency (MSL) < or =8 min were found in 21 (50.0%) and 19 (44.2%) subjects, and either in 30 (69.8%) subjects. ES did not
relate to MSL. Subjects with subjective sleepiness (ES> or =11) reported more cataplexy-like and sleep paralysis symptoms, longer habitual sleep times, and higher sleep efficiency and REM sleep per cent than those without. Subjects with objective sleepiness (MSL< or =8 min) had a higher stage 4 sleep per cent. Subjects with > or =2 SOREMPs (25.6%) showed higher muscular impairment, lower MSL, higher ES, and more cataplexy-like symptoms than those with < or =1 SOREMP. Apnoea-hypopnoea index (AHI) > or =5, predominantly obstructive, was found in 37 (86.0%) subjects, and AHI >30 in 12 (27.9%). Neither subjective nor objective sleepiness could be explained by AHI, nor satisfactorily predicted by daytime respiratory abnormalities.

CONCLUSIONS: DM1 entails frequent EDS but with different phenotypes and distinct mechanisms involved. The high prevalence of daytime sleepiness and severe sleep apnoeas found in this study supports the routine use of clinical sleep interviews, PSG and MSLT in DM1, and emphasises the need for more randomised trials of psychostimulants.

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**Publication Types, MeSH Terms**


**Predictors of disrupted social participation in myotonic dystrophy type 1.**

Gagnon C¹, Mathieu J, Jean S, Laberge L, Perron M, Veillette S, Richer L, Noreau L.

**Author information**

**Abstract**

**OBJECTIVE:** To identify personal and environmental predictors of the most disrupted participation domains in people with myotonic dystrophy type 1 (DM1).

**DESIGN:** Cross-sectional study.

**SETTING:** Outpatient neuromuscular clinic.

**PARTICIPANTS:** Adults (n=200; 121 women), age 18 years or older (mean age, 47 y), with a confirmed diagnosis of DM1 were selected from the registry of a neuromuscular clinic (N=416). Fifty-two participants had the mild phenotype and 148 the adult phenotype.

**INTERVENTIONS:** Not applicable.

**MAIN OUTCOME MEASURES:** Social participation in mobility, housing, employment, and recreation was assessed with the Life Habits Measure. Disrupted participation was based on whether help was needed in performing most life habits because of incapacities or environmental barriers. Environmental factors were assessed by using the Measure of the
Quality of the Environment. Personal factors were assessed with standardized instruments including the Berg Balance Scale, the Krupp Fatigue Severity Scale, and manual muscle testing.

**RESULTS:** A large proportion of participants (45%-61%) reported disrupted participation in all 4 domains. Lower-extremity strength (odd ratios [OR], 15.0-5.5; P<.050) and higher fatigue (OR, 6.0-2.6; P<.05) were present in participants with disrupted participation. With regard to environmental factors, family support (OR, 3.6-2.5; P<.05) and public services (OR, 2.8-2.2; P<.05) were perceived as barriers for participants with disrupted participation in most domains.

**CONCLUSIONS:** This study identified personal and environmental factors that may influence the trajectory toward disrupted participation in individuals with DM1. Fatigue, strength, family support, and public services were found to be independent predictors of disrupted participation.

**Comment in**

*Pain in myotonic muscular dystrophy, type 1.* [Arch Phys Med Rehabil. 2008]

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**Publication Types, MeSH Terms**

8. **Towards an integrative approach to the management of myotonic dystrophy type 1.**
   Gagnon C ¹, Noreau L, Moxley RT, Laberge L, Jean S, Richer L, Perron M, Veillette S, Mathieu J.

**Author information**


**Publication Types, MeSH Terms**

9. **Fatigue and daytime sleepiness rating scales in myotonic dystrophy: a study of reliability.**
   Laberge L ¹, Gagnon C, Jean S, Mathieu J.

**Author information**

**Abstract**
OBJECTIVES: To assess the reliability of the Epworth Sleepiness Scale (ESS), Daytime Sleepiness Scale (DSS), Chalder Fatigue Scale (CFS), and Krupp's Fatigue Severity Scale (KFSS) in patients with myotonic dystrophy type 1 (DM1).

METHODS: In total, 27 patients with DM1 were administered the questionnaires on two occasions, with a 2 week interval. Internal consistency and test retest reliability were measured using intraclass correlation coefficients (ICCs), and Cronbach's alpha, Cohen's kappa, and Goodman-Kruskal's gamma coefficients.

RESULTS: Internal consistency of the CFS and KFSS were adequate (alpha > 0.70) but that of the ESS was weak (alpha = 0.24). Both daytime sleepiness and fatigue rating scales showed significant test retest reliability. Test retest reliability for individual items revealed inconsistencies for some ESS and CFS items.

CONCLUSIONS: Reliability of the CFS, DSS, and KFSS was high, allowing their use for individual patients with DM1, but that of the ESS was lower, rendering its current usage in DM1 questionable. Fatigue rating scales such as the KFSS, which are based on the behavioural consequences of fatigue, may constitute a more accurate and comprehensive measure of fatigue severity in the DM1 population.

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Publication Types, MeSH Terms