Understanding cognitive impairments' impact on daily life in myotonic dystrophy: an exploratory sequential mixed design study

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Introduction

- Myotonic dystrophy type 1 (DM1) is a **rare neuromuscular disease**
- It presents with **multisystemic and progressive impairments:** muscles, heart, respiration, endocrine system, gastrointestinal system, ocular system and central nervous system (CNS) [1]
- There are five phenotypes based on age of symptom onset: congenital, infantile, juvenile, **adult-onset (most common)**, and late-onset [2]
- CNS impairments include cognitive disorders (memory, attention, executive functions, etc.) [3] and hypersomnolence [4]
- **Patients living with DM1 experience increasing restrictions in activities of daily living (ADL). In the adult-onset, restrictions mostly involves instrumental activities of daily living (IADL)** [5, 6] **:**

50% are unable or need help to maintain their home

68% to do their housework

52% to maintain a job

31% to prepare meals

Restrictions

Most studies have focused on describing impairments present in DM1, but none have specifically investigated the impact of CNS impairments on IADL performance.

Objective

To understand and describe the impact of CNS impairments on IADL performance in the adult-onset phenotype of DM1, from both objective (standardized assessments) and subjective (qualitative observations and discussions) perspectives.

Methodology

We used a exploratory sequential mixed design (QUAL → quan model) [7] in collaboration with a patient-partner and a relative research partner

Integration

	Qualitative		Quantitative
Phase 1	Multiple case study	→	Transversal observational study
Phase 2	Focus groups		

References

Website

GRIMN

Centre intégré universitaire de santé et de services sociaux du Saguenay–Lac-Saint-Jean Québec

Muscular Dystrophy Canada / Dystrophie musculaire Canada

UdS Université de Sherbrooke

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Methodology (continued)

Qualitative phase

Phase 1 Multiple case study [8]

Observation with the IADL Profile [9]

Assessment of the performance of five IADL: Communication, Financial management, Shopping, Meal preparation and Cleaning

Semi-structured interviews with the DM1 patient and with one of their relative (separately)

To get their perspective on the cognitive impairments (if present) and problems in daily life

Neuropsychological assessment

Assessment of the following cognitive functions: working memory, executive functions, attention, visuospatial abilities, apathy, hypersomnolence

Table I. Participants characteristics (n=6)

#	Age	Sex	CTG*	Mobility	Education	Living environment
1	56	W	400	No aid	Grade 10	Partner
2	62	M	1000	Rollator	College	Partner
3	37	M	250	No aid	Grade 11	Alone
4	49	W	550	No aid	Grade 11	Alone
5	56	M	150	No aid	College	Partner and children
6	36	W	350	No aid	Grade 9	Partner and children

** CTG corresponds to the number of repeats in the affected gene and correlates with the severity of the disease*

Phase 2 Focus groups [10]

Two focus groups

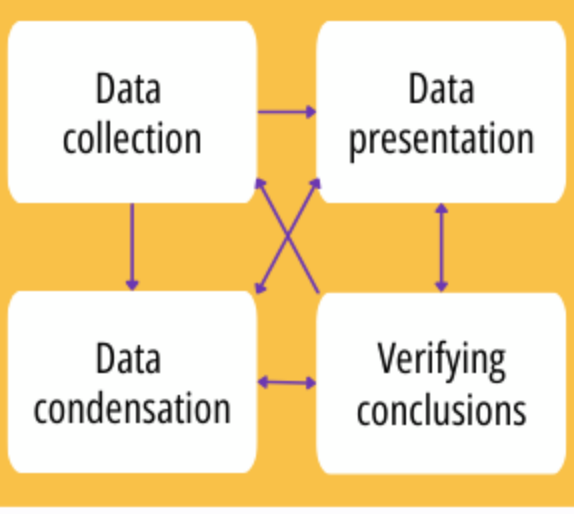
- Conducted sequentially, in presence
- Animated by one or two OT from the research team, lasting approximately 2 hours each

First group: OT working in community (primary care) (n=5):

- Age: 35-50 years old
- Experience with DM1 patients (years): M = 18,8 ; SD 9,9

Second group: HCP of various discipline, working in the neuromuscular clinic (NMC) (tertiary care) (n=6): neurologist, neuropsychologist, nurse, OT, physical therapist and social worker

- Age: 35-57 years old
- Experience with DM1 patients (years): M = 13,7 ; SD 7,4



Thematic content analysis [11]

- **Iterative process**: throughout data collection (intra-case → inter-case → focus group)
- Analysis mainly carried out by student researcher and co-analysis with research director and **patient-partners**
- Coding and mixed categorization approach: based on objectives and conceptual frameworks (deductive) and what emerges from the data (inductive)
- Use of qualitative data processing software (**NVivo**)

Quantitative phase

Information from the qualitative results

+ Scoping review of all available OT assessments

+ Consultation with neuropsychologists and DM1 experts

Quantitative analysis to come

Selection of variables for a transversal observational study

Dependent variables → **1 First half-day assessment (DM1 patients)**

Selection of two OT assessments:

- IADL Profile (scenarios 2 and 3)
- Executive Function Performance Test
- ADA

Independent variables → **2 Second half-day assessment (DM1 patients)**

Neuropsychological functions:

- Visuospatial abilities
- Attention
- Executive functions
- Self-awareness
- Fatigue and hypersomnolence
- Anxiety and depression

3 Third half-day assessment (relatives)

Results

Qualitative phase

Phase 1

In the ADL Profile, difficulties were observed across all executive operations

In the neuropsychological testing, impairments were identified across all assessed domains (working memory, selective and sustained attention, executive functions, visuospatial perception, apathy and sleeping profile)

In the individual interviews, CNS symptoms related to working memory, attention, executive functions, hypersomnolence and visuospatial deficits were raised by DM1 participants and their relatives. Problematic performance concerned cleaning, financial management, house-related procedures, meal preparation and driving

Phase 2

In the focus groups, CNS symptoms related to impulsivity, abstraction abilities (generalization, anticipating consequences), high-level executive functions (judgment, problem-solving skills), social cognition, irritability and perseveration were reported. Issues related to IADL performance were identified in financial management (indebtedness), shopping, cleaning (unsanitary homes), health management (refusing recommendations and/or help), interpersonal relationships (isolation)

Triangulation and integration of all data allowed us to identify possible factors to these difficulties and profiles, such as:

Issues related to HOUSE CLEANING emerged in case 3:

Assessment allowed to observe **difficulty to initiate the task** (i.e., can tell what they need to do, but required verbal assistance to start)

Possible contributing factors

Patients reported **decreased motivation**, while relatives talked about **"laziness"**

"Starting is the worst" (Part. 3)

*That's it, she would be able to do it, but it's because she is lazy; **if she wouldn't be lazy, she would do it"*** (Relative of part. 4)

Presence of moderate apathy was observed (Lille Apathy Rating Scale)

Issues related to MEAL PREPARATION emerged in case 1, 2 and 5:

Assessment allowed to observe **difficulty to accomplish the task** (i.e., needed many breaks)

Possible contributing factors

*"[they] have a good deal of daytime sleepiness, so they get up at noon, or 2 a.m., so they're **decycled**, I'll put it that way, and that's why there are appointments where they don't show up. And I think they cut short just about everything. You know, **I'm just going to buy a loaf of bread because the grocery store is closing**, or the convenience store, you know. Then I'll eat some toast. **They cut that short too because of that."*** (NMC focus group)

Patients and relatives reported needing **many hours of sleep** and **waking up late**

Disrupted chronotype and **low-quality sleep** (Pittsburgh Sleep Quality Index)

Contribution to the OT profession

- This project used an innovative methodology which allowed a deep, rigorous, and structured in-depth understanding of the impact of CNS impairments on IADL performance, with the perspective of patients, relatives, and healthcare providers
- This study highlights the significant contribution of the OT in this type of assessment and, ultimately, will provide specific guidelines for OT working with this disease and other similar conditions

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Website

Thank you!