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Research Article

Assessing Cognitive and Behavioral Deficits in Children with Myotonic Dystrophy Type 1: A Pilot Study

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Abstract

Myotonic Dystrophy Type 1 (DM1), a rare autosomal dominant disorder caused by a CTG triplet repeat expansion in the *DMPK* gene, manifests with diverse physical, cognitive, and behavioral symptoms. While its neuromuscular features are well-documented, cognitive and behavioral aspects remain underexplored. This study piloted a neuropsychological test battery for children with DM1, assessing its feasibility and sensitivity in eight participants (ages 11-19) from the Neuromuscular Reference Center at University Hospitals Leuven.

The test battery evaluated intelligence, memory, visual-motor integration, and executive functions, supplemented by parent, teacher, and self-report questionnaires measuring behavior and executive functioning. Results showed 75% of participants scored below average on intelligence tests, with total IQ negatively correlated with CTG repeat size ($\rho = -0.69$, $p < .05$). Distinct memory patterns emerged: visual memory deficits correlated negatively with CTG repeat size ($\rho = -0.76$, $p < .05$), while verbal memory showed a positive correlation ($\rho = 0.74$, $p < .05$). Behavioral assessments revealed significant deficits in attention (75%, $p < .001$), working memory (71.5%, $p < .01$), and initiative-taking ($p < .01$).

The findings underscore the utility of this battery in identifying DM1-associated cognitive and behavioral deficits and suggest distinct neural pathways for visual and verbal memory processing. These results highlight the need for more neuropsychological and imaging studies to better understand the neural mechanisms underlying DM1.

Keywords: *Cognitive and Behavioral Deficits; Myotonic Dystrophy Type 1 (DM1); CTG Triplet Repeat; DMPK Gene; Children*

Introduction

Myotonic Dystrophy Type 1 (DM1) is a rare autosomal dominant disorder caused by a CTG triplet repeat expansion in the *DMPK* gene on chromosome 19q13.3 [1]. The disease manifests with a wide range of physical, cognitive, and behavioral symptoms. Although the incidence is approximately 1 in 8,000 newborns, the prevalence among children under 16 is estimated at 5 per 100,000, with symptoms often emerging later in life [1]. DM1 is a multisystem disorder affecting skeletal muscles, the eyes, heart, smooth muscles, and the endocrine and central nervous systems.

Cognitive impairments in DM1 are diverse, and intelligence assessments have traditionally classified patients into two groups: those with normal intelligence (mean IQ = 91, SD = 13) and those with mild intellectual disability (mean IQ = 58, SD = 9) [2,3]. More recent studies have reinforced this variation in cognitive performance across the different subtypes of DM1 (congenital, infantile, and juvenile) [4]. A consistent finding across studies is the negative correlation between the size of the CTG repeat expansion and IQ, particularly in maternally inherited cases [5,6].

Neuropsychological deficits, particularly in working memory and attention, are among the most frequently reported cognitive impairments in children with DM1. Up to 71% of children show working memory deficits, while 66% experience attention impairments [6]. These deficits are linked to structural brain abnormalities, including white matter lesions, as revealed by advanced imaging techniques [7]. The role of these brain abnormalities in the cognitive profile of DM1 patients is further emphasized by studies indicating a higher vulnerability to executive dysfunction, which significantly affects academic performance and daily living skills [8].

Behavioral and psychiatric comorbidities, including ADHD, anxiety, and autism spectrum disorders, are common [9-11]. These behavioral issues often interfere with educational and social functioning, making early recognition and management crucial [12,13].

Neuroimaging studies have provided deeper insight into the structural changes that underlie cognitive deficits in DM1. Recent findings suggest that DM1-related brain abnormalities are not merely the result of focal lesions but rather a disruption in brain connectivity, which may help explain the wide variability in cognitive and behavioral outcomes [5,14,15]. These studies propose that the effects of DM1 on cognitive functioning should be viewed as a disorder of connectivity, highlighting the complex interaction between neurodevelopmental and neurodegenerative processes. Sweere., *et al*. (2023) corroborate these findings, emphasizing the need for longitudinal neuropsychological assessments to guide clinical management and optimize care strategies for children with DM1 [6]. However, international consensus about how this longitudinal neuropsychological follow-up should be organized and which tests should be included, is lacking. In this study, we propose such a neuropsychological test battery and perform a pilot studie on a sample of MD1 patients to explore the feasibility.

Methodology

Participants

Children with MD1 were recruited from the Neuromuscular Reference Center of the University Hospitals Leuven, Belgium. 10 children between 11 and 20 years of age were invited for a neuropsychological examination. Eight of them agreed for participation. Both children and parents filled in an informed consent approved by the ethics committee of the University Hospitals Leuven (s57182).

Instruments

An overview of the included tests is given in table 1. Intelligence is measured by means of the Wechsler Intelligence Scale for Children III (WISC-III NL) or the Wechsler Adult Intelligence Scale IV. In case of the WISC-III we added a fluid intelligence task, Matrix Reasoning, from the Nonverbal Wechsler Scale (NWS). We used the computerized Amsterdam Neuropsychological Tasks to investigate attention and executive functioning. Memory was tested by means of the Children Memory Scale and visual working memory were investigated with the Spatial Orientation task from the NWS. Visual-motorial integration was measured by means of the VMI 5th edition.

Testing was done by a trained neuropsychologist who was familiar to the patients. To exclude an effect of fatigue, we split the testing moments in blocks of maximum 2 hours. Parents and teachers were asked to fill in the Child Behavior Checklist (CBCL) and the Behaviour Rating Inventory of Executive Functioning (BRIEF) for parents and the Teachers Report Form and BRIEF for teachers, respectively. Children were asked to complete the Youth Self Report (YSR) and BRIEF for adolescents.

Neuropsychological function	Test	Subtest	Age limits
Intelligence	WISC-III	- Picture completion - Information - Coding - Similarities - Picture arrangemaent - Arithmetic - Block Desing - Vocabulary - Object Assembly - Comprehension - Symbol Search	7- 16 years
Attention/Executive functioning	ANT	- Baseline speed - Focused Attention 4 letters - Memory Search Letters - Sustained attention dots - Set Shift Visual	
Memory	Children Memory Scale	- Dots Location - Verbal paired associate learning - Digit span	5-16 years
Fluid Intelligence	Nonverbal Wechsler Scale	- Matrix Reasoning	4-22 years
Visual working memory	Nonverbal Wechsler Scale	- Spatial orientation	4-22 years
Visual-motorial integration	VMI		
Behavioral questionnaires	CBCL YSR TRF BRIEF parents BRIEF teachers BRIEF adolescents		6 - 18 years 11-18 years 6-18 years 5-18 years 5-18 years 11-18 years
WISC-III = Wechsler Intelligence Scale for Children 3rd edition; ANT = Amsterdam Neuropsychological Tasks; CBCL = Childbehavior Checklist; YSR = Youth Self Report; TRF = Teacher's Report Form; BRIEF = Behaviour Rating Inventory of Executive Functioning			

Table 1: *Selected neuropsychological tests.*

Statistics

Raw data were transformed into standard scores using the age-adapted norms of the different tests. SPSS version 29 was used to analyze normative data. Descriptive statistics were calculated, and a one-sample t-test was used to compare our study sample with population norms. Analyses were performed on a 95% confidence-level. Pearson's Rho correlation coefficient was used to investigate the relation between outcomes on the different instruments and number of CTG-repeats.

Results

Participants

Eight children and their parents consented to participate in the study and signed informed consent forms. Demographic variables are shown in table 2. Two participants did not return their YSR and BRIEF questionnaires for adolescents and one participant was unable to complete the entire test battery, resulting in missing results for the ANT. Another participant, due to age, could not be tested with the CMS and instead received age-adapted questionnaires, specifically the Adult Self Report and Adult Behavior Checklist.

Nr	Gender	Age	CTG-repeats	Inheritance
1	F	14 yrs	600	Paternal
2	M	15 yrs	930	Paternal
3	M	11 yrs	/	Maternal
4	M	14 yrs	400	Maternal
5	M	16 yrs	140	Maternal
6	M	19 yrs	400	Maternal
7	M	14 yrs	400	Paternal
8	F	14 yrs	730	Maternal
Mean (SD)		14,6 (2,2)		

Table 2: *Demographic variables of participants.*

Neuropsychological functioning

Intelligence

The majority of the sample (75%) scored below average on the intelligence test, with IQs ranging from 54 to 111. Total IQ was significantly lower than the average ($t = -3.30$, $p < .05$), as were verbal IQ ($t = -3.65$, $p < .05$), performance IQ ($t = -3.37$, $p < .05$), and fluid intelligence, as measured by an abstract reasoning task ($t = -2.91$, $p < .05$).

Memory

Only a small proportion of participants (approximately 30%) scored significantly below average on both visual and auditory memory tasks. Mean memory scores did not differ significantly from normative data. However, significantly lower scores were observed for both visual and auditory working memory tasks, with impairments identified in 75% and 100% of participants, respectively ($p < .05$).

Visual-constructive functioning

In this sample, 75% of participants demonstrated impairments in visual-constructive skills. Significant group effects were observed in the integration task ($p < .01$), the visual perception task ($p < .01$), and the motorial integration task ($p < .05$).

Amsterdam neuropsychological tasks (ANT)

Half of the MD1 patients exhibited slower reaction times compared to average. Reaction time did not appear to be further impacted in tasks with increased memory demands. In tasks requiring selective attention, inhibition, or sustained attention, MD1 patients were slower but maintained accuracy comparable to their peers. However, tasks requiring cognitive flexibility revealed that 50% of MD1 patients made more errors than average.

Behavioral functioning

Self-reports from MD1 patients and teacher assessments did not indicate problems that are considered clinically significant (T-score > 65). In contrast, 85% of parents reported reduced competencies in their children ($p < .05$). Parents specifically highlighted issues with attention (75%, $p < .001$), working memory (71.5%, $p < .01$), and taking initiative ($p < .01$).

Function	N	Mean (SD)	t	p-value (2-tailed)
Intelligence				
Full scale IQ	8	75,50 (21,02)	-3,30	.013*
Verbal IQ	8	78,13 (16,94)	-3,65	.008**
Performance IQ	8	76,63 (19,64)	-3,37	.012*
Fluid Intelligence (MR)	7	43,57 (5,86)	-2,91	.027*
Visual-constructive capacities				
Beery VMI	8	72,00 (22,00)	-3,61	.009**
Visual perception	8	74,00 (19,15)	-3,84	.006**
Motorial coordination	8	77,25 (20,56)	-3,13	.017*
Memory				
Immediate visual memory	7	95,57 (22,63)	-.52	.623
Deferred visual memory	7	97,29 (16,39)	-.44	.677
Immediate verbal memory	7	102,71 (30,90)	-.23	.824
Deferred verbal memory	7	94,43 (24,55)	-.60	.570
General Memory Index	7	94,00 (14,71)	-1,08	.322
Attention and Concentration	7	61,43 (28,94)	-3,53	.012*
Spatial Orientation	8	38,63 (8,31)	-3,87	.006**
Child Behaviour Checklist				
Total competence	6	25,33 (15,38)	-3,93	.011*
Anxious/Depressed	8	65,00 (10,54)	4,02	.005**
Withdrawn/Depressed	8	64,75 (9,93)	4,20	.004**
Somatic complaints	8	59,00 (7,11)	3,58	.009**
Social Problems	7	68,57 (6,68)	7,36	.000**
Thought problems	8	66,13 (7,75)	5,88	.001**
Attention problems	8	74,50 (9,29)	7,46	.000**
Rule-breaking behavior	8	58,00 (6,30)	3,59	.009**
Aggressive Behavior	8	58,63 (8,05)	3,03	.019*
Internalizing problems	8	64,50 (8,42)	4,87	.002**
Externalizing problems	8	57,75 (90,7)	2,42	.046*
Total problems	8	67,00 (6,26)	7,69	.000**
Youth Self Report				
Anxious/Depressed	6	56,67 (6,02)	2,71	.042*
Withdrawn/Depressed	6	61,00 (7,51)	3,59	.016*
Somatic complaints	6	55,83 (6,08)	2,35	.066

Social Problems	5	60,00 (7,94)	2,82	.048*
Thought problems	6	59,00 (4,82)	4,58	.006**
Attention problems	6	67,67 (13,84)	3,13	.026*
Rule-breaking behavior	6	55,67 (3,67)	3,78	.013*
Aggressive Behavior	6	55,67 (7,17)	1,94	.111
Internalizing problems	6	57,50 (7,34)	2,50	.054
Externalizing problems	6	54,50 (7,06)	1,56	.179
Total problems	6	58,50 (7,18)	2,90	.034*
Teachers Report Form				
Anxious/Depressed	5	59,60 (9,92)	2,17	.096
Withdrawn/Depressed	5	60,40 (6,95)	3,35	.029*
Somatic complaints	5	56,80 (6,57)	2,31	.082
Social Problems	5	63,80 (9,09)	3,39	.027*
Thought problems	5	57,00 (4,95)	3,16	.034*
Attention problems	5	58,00 (5,79)	3,09	.037*
Rule-breaking behavior	5	56,00 (8,49)	1,58	.189
Aggressive Behavior	5	59,40 (14,21)	1,48	.213
Internalizing problems	5	59,60 (10,21)	2,10	.103
Externalizing problems	5	53,60 (15,92)	.51	.640
Total problems	5	60,80 (8,79)	2,75	.051
BRIEF Parents				
Inhibition	7	62,57 (4,58)	7,27	.000**
Cognitive flexibility	7	64,71 (3,86)	10,08	.000**
Emotion Regulations	7	58,71 (4,31)	5,35	.002**
Behavior regulation-index	7	63,29 (3,73)	9,43	.000**
Taking initiative	7	66,43 (6,37)	6,82	.000**
Working Memory	7	67,57 (7,55)	6,16	.001**
Planning and organising	7	61,43 (5,83)	5,19	.002**
Orderliness and neatness	7	59,14 (7,44)	3,25	.018*
Behavior evaluation	7	64,71 (6,70)	5,81	.001**
Metacognition-index	7	66,43 (4,65)	9,35	.000**
Total Score	7	66,14 (3,39)	12,61	.000**
BRIEF Teachers				
Inhibition	5	57,20 (11,43)	1,49	.232
Cognitive flexibility	5	59,20 (4,76)	4,32	.012*
Emotion Regulations	5	54,40 (10,19)	.97	.389
Behavior regulation-index	5	56,40 (10,97)	1,31	.262
Taking initiative	5	55,40 (10,23)	1,18	.304
Working Memory	5	57,00 (8,37)	1,87	.135

Planning and organising	5	59,20 (8,26)	2,49	.067
Orderliness and neatness	5	60,00 (3,16)	7,07	.002**
Behavior evaluation	5	58,40 (4,16)	4,52	.011*
Metacognition-index	5	59,20 (4,15)	4,96	.008**
Total Score	5	59,40 (6,19)	3,40	.027*
BRIEF adolescent				
Inhibition	5	51,60 (11,24)	.32	.766
Cognitive flexibility	5	57,40 (5,46)	3,03	.039*
Emotion Regulations	5	50,80 (13,68)	.13	.902
Behavior regulation-index	5	48,00 (11,64)	-.38	.720
Taking initiative	5	50,00 (8,49)	.00	1,000
Working Memory	5	50,00 (8,09)	.00	1,000
Planning and organising	5	53,60 (5,94)	1,36	.247
Orderliness and neatness	5	45,80 (8,35)	-1,13	.324
Behavior evaluation	5	47,80 (7,82)	-.63	.564
Metacognition-index	5	53,80 (5,85)	1,46	.220
Total Score	5	51,00 (9,85)	.23	.832
* p < .05 ** p < .01				

Table 3: *Results MD1-patients versus age-adapted norms.*

Relationships Between neuropsychological and behavioral outcomes with number of CTG repeats

The number of CTG repeats in the cohort was negatively correlated with total IQ ($\rho = -0.69$, $p < .05$), abstract reasoning ability ($\rho = -0.76$, $p < .05$), visual perception ($\rho = -0.90$, $p < .01$), and immediate visual memory ($\rho = -0.76$, $p < .05$). These findings support the hypothesis that a higher number of CTG repeats is associated with an increased frequency of comprehensive problems. Notably, immediate verbal memory displayed a strong positive correlation with the number of CTG repeats ($\rho = 0.74$, $p < .05$). Additionally, a significant negative correlation was identified between visual perception and immediate verbal memory ($\rho = -0.72$, $p < .05$). No significant correlations were found between the number of CTG repeats and the other behavioral or neuropsychological outcomes.

Discussion

In this study, we designed a neuropsychological test battery to assess cognitive functioning in children with Myotonic Dystrophy Type 1 (MD1). Historically, MD1 has been considered primarily a neuromuscular disorder, however, a growing consensus emerged that MD1 patients exhibit a distinct behavioral and cognitive phenotype [5]. While the specific phenotype can vary widely, shared neurobiological mechanisms are thought to underlie these presentations. Despite MD1 being one of the most common neuromuscular disorders, research into its cognitive and neuropsychological dimensions remains limited.

Our findings, which include deficits in intelligence, attention, working memory, and visuospatial functioning, align with the results of previous studies. For example, 75% of our participants scored more than one standard deviation below average on the IQ test, and FSIQ correlated significantly with the number of CTG repeats. Moreover, 75% of our participants exhibited significant impairments in visual perception, which showed the highest correlation with the number of CTG repeats. These results are consistent with previous findings which emphasized visuospatial deficits as a prominent feature of the cognitive profile in childhood MD1 [6,16]. This suggests that MD1

impacts not only cognitive but also perceptual abilities. Future research should explore the extent to which perceptual deficits influence overall development in children with MD1.

While intellectual impairments typically predict lower performance on memory tasks, our results revealed no significant differences in mean scores for visual or verbal memory tasks. However, a strong negative correlation between CTG repeat size and visual memory scores contrasted with a strong positive correlation between CTG repeat size and verbal memory scores. Variability in memory performance has been found before, suggesting that verbal memory may be relatively preserved in some subtypes of MD1 [6]. This paradox raises intriguing questions about underlying neurobiological mechanisms. One hypothesis is that impairments in visual memory might drive compensatory development of verbal memory. However, this explanation seems unlikely, as verbal learning capacity often surpasses general intellectual ability in these children, pointing to other possible mechanisms.

Earlier neuroimaging studies provide evidence of disruptions in frontotemporal and frontoparietal networks in adults with MD1, with more recent studies demonstrating microstructural alterations in brain connectivity [14,15]. These connectivity disruptions likely contribute to the heterogeneous cognitive and behavioral outcomes observed in MD1 [6]. Mutations in the *DMPK* gene appear to disrupt the development of functional brain networks, as suggested by both imaging findings and neuropsychological data. Future research should integrate advanced neuroimaging techniques with neuropsychological assessments to investigate potential links between affected brain networks and specific cognitive deficits, such as visual-perceptual impairments and verbal memory strengths [17].

In contrast to self-reports and teacher assessments, parents consistently report significant behavioral difficulties in children DM1. This discrepancy is noteworthy, as perceptions of behavior may differ across contexts and depend on the perspective of the observer. While teachers and peers may primarily evaluate behavior within structured educational environments, parents are more likely to observe behavior in unstructured home settings, which may be more sensitive to executive dysfunctions, emotional regulation, and social challenges. Research has highlighted that children with DM1 often exhibit internalizing problems such as anxiety, withdrawal, and depressive symptoms, as well as social difficulties and attention-related problems [3,9,18]. Parental reports in our study align with these findings, particularly in areas related to attention, working memory, and initiative-taking.

Despite this, behavioral functioning in DM1 remains less studied than cognitive and motor impairments. While some studies suggest an overlap with psychiatric disorder, others have linked behavioral difficulties to impairments in neuropsychological functioning and the underlying white matter abnormalities frequently observed in neuroimaging studies [5,19-22]. Given the significant impact that behavioral difficulties can have on daily functioning and family burden, this aspect warrants further exploration in future research. A deeper understanding of the specific behavioral phenotype of DM1 could provide insights for targeted interventions and support for both families and educational professionals [23].

This study represents a novel attempt to design and pilot a neuropsychological test battery for children with MD1, addressing a wide range of cognitive functions. However, we acknowledge several methodological limitations. First, the small and homogeneous sample size precludes generalizability to the broader MD1 population. Second, the absence of a control group limits comparative interpretation. Finally, while we observed significant correlations involving visual perception, we did not include other perceptual tasks, such as auditory perception, which could provide additional insights in future research.

Conclusion

MD1 has often been considered a predominantly neuromuscular disorder, leaving its broader impact on cognition and behavior underexplored. A better understanding of the neuropsychological functioning of children with MD1 can inform tailored interventions and enhance quality of care. While much remains to be done to unravel the true impact of MD1, systematic follow-up of these patients is critical

for both clinical and research purposes. In this paper, we have proposed a potential neuropsychological test battery and highlighted the possibility of distinctive information processing for visual and verbal information in children with MD1.

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Author Contributions

SG, JL, NG and LDW were involved in the conception and design of the study. SG was involved in the acquisition and analysis of the data. SG, JL, NG and LDW played an important role in the interpretation of the results. SG drafted a significant proportion of the manuscript. All authors reviewed the final manuscript.

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Conflict of Interest

No conflict of interest was reported.

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