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NEUROPSYCHOLOGICAL PROFILE OF AN ADOLESCENT WITH DUCHENNE MUSCULAR DYSTROPHY: A CASE STUDY

PERFIL NEUROPSICOLÓGICO DE UN ADOLESCENTE CON DISTROFIA MUSCULAR DE DUCHENNE: UN ESTUDIO DE CASO

PERFIL NEUROPSICOLÓGICO DE UM ADOLESCENTE COM DISTROFIA MUSCULAR DE DUCHENNE: UM ESTUDO DE CASO

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Palavras-chave: Estudo de caso; Transtorno de Tourette; Comorbidade; TDAH; Deficiência Intelectual.

A B S T R A C T

Duchenne Muscular Dystrophy (DMD) is one of the most common diseases that leads to muscle wasting. It is a recessive and hereditary congenital disorder, affecting mostly males. It is caused by alterations in the expression of the Xp21 segment of the X chromosome, which is responsible for regulating the production of the dystrophin protein. Due to the association between the lack of dystrophin and cortical post-synaptic neural atrophy, DMD is associated with a high likelihood of cognitive impairment, especially in reasoning, executive functions, and academic performance. This study presents a case report detailing a neuropsychological assessment conducted on a 14-year-old boy with DMD. The main objective was to comprehend the underlying reasons for the patient's low academic performance. The assessment included measures of reasoning, attention, language skills, executive functioning, memory, and academic performance. The findings are on par with international literature, demonstrating that there are neuropsychological impairments even in the absence of a deficit in IQ. Notably, this study identified that language and academic achievement may be significantly impacted, which had not been thoroughly explored in the scientific literature. Thus, it can be concluded that understanding the impacts of DMD beyond the physical symptoms could enhance support for affected individuals worldwide.

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R E S U M E N

La distrofia muscular de Duchenne (DMD) es una de las enfermedades más frecuentes que conducen a la pérdida progresiva de masa muscular. Se trata de un trastorno congénito, hereditario y recesivo ligado al cromosoma X, que afecta principalmente a varones. Está causada por alteraciones en la expresión del segmento Xp21 del cromosoma X, responsable de regular la producción de la proteína distrofina. Debido a la asociación entre la ausencia de distrofina y la atrofia neural cortical postsináptica, la DMD se vincula con una alta probabilidad de compromiso cognitivo, especialmente en áreas como el razonamiento, las funciones ejecutivas y el rendimiento académico. Este estudio presenta un reporte de caso que describe una evaluación neuropsicológica realizada a un adolescente de 14 años con DMD. El objetivo principal fue comprender los factores subyacentes al bajo rendimiento académico del paciente. La evaluación incluyó medidas de razonamiento, atención, lenguaje, funciones ejecutivas, memoria y desempeño académico. Los resultados son concordantes con la literatura internacional, evidenciando la presencia de alteraciones neuropsicológicas incluso en ausencia de un déficit en el coeficiente intelectual (CI). De manera destacada, este estudio identificó que el lenguaje y el rendimiento académico pueden verse significativamente afectados, aspectos que no han sido explorados en profundidad en la literatura científica. En consecuencia, se concluye que comprender los efectos de la DMD más allá de los síntomas físicos podría mejorar el apoyo brindado a las personas afectadas a nivel mundial.

R E S U M O

A Distrofia Muscular de Duchenne (DMD) é uma das doenças mais comuns que levam à degeneração muscular. Trata-se de um distúrbio congênito, hereditário e recessivo, que afeta predominantemente indivíduos do sexo masculino. A DMD é causada por alterações na expressão do segmento Xp21 do cromossomo X, responsável por regular a produção da proteína distrofina. Devido à associação entre a falta de distrofina e a atrofia neural pós-sináptica cortical, a DMD está relacionada a uma alta probabilidade de comprometimento cognitivo, especialmente no raciocínio, nas funções executivas e no desempenho acadêmico. Este estudo apresenta um relato de caso detalhando uma avaliação neuropsicológica realizada em um menino de 14 anos com DMD. O principal objetivo foi compreender as razões subjacentes ao baixo desempenho acadêmico do paciente. A avaliação incluiu medidas de raciocínio, atenção, habilidades linguísticas, funcionamento executivo, memória e desempenho acadêmico. Os achados estão alinhados com a literatura internacional, demonstrando que há prejuízos neuropsicológicos mesmo na ausência de um déficit no QI. Notavelmente, este estudo identificou que a linguagem e o rendimento acadêmico podem ser significativamente impactados, um aspecto que ainda não havia sido amplamente explorado na literatura científica. Assim, conclui-se que compreender os impactos da DMD para além dos sintomas físicos pode aprimorar o suporte oferecido aos indivíduos afetados em todo o mundo.

Duchenne muscular dystrophy (DMD) is an X-linked recessive disorder caused by mutations in the Xp21 segment, which lead to dystrophin deficiency and progressive muscle and neural degeneration, primarily affecting males (Darras et al., 2000; Verhaart & Aartsma-Rus, 2019). The lack of this protein is associated with post-synaptic neuron atrophy and a lack of production of the protein chain labeled PSD-95, which is essential for building structures such as carrier proteins and ionic channels (Chieffo et al., 2015; Aranmolate et al., 2017). DMD is characterized by a progressive, severe, and irreversible loss of strength and muscle mass, a high mortality rate, and a life expectancy of approximately four decades post-diagnosis (Darras et al., 2000; Birnkrant et al., 2018). DMD has a prevalence of 15.9 to 19.5 per 100,000 live births (Ryder et al., 2017), and its prevalence is the highest in North America (Trindade et al., 2024).

Impairments due to DMD extend across various systems of the human body, including the cardiovascular, respiratory, muscular, and nervous systems (Anikiej et al., 2018). The impairment of the nervous system associated with DMD is linked with dystrophin deficiency (Connoly et al., 2013). McKusick (1986) claimed that other diseases linked to the alteration of the Xp21 segment in the X chromosome may result in a lower IQ. Of the 349 phenotypes associated with mutations in Xp21, 80 presented alterations in reasoning, with 39 categorized as syndromic, which includes cases of DMD (McKusick, 1986).

Estimates suggest that between 15 to 63% of individuals affected by DMD present some degree of intellectual impairment (Cotton et al., 2005). Between 15% and 60% of people with DMD have some degree of cognitive impairment, ranging from borderline functioning to mild or moderate deficits (Cotton et al., 2005; Nardes et al., 2020). According to Bresolin et. al (1994), intellectual difficulties constitute an intrinsic component of DMD's symptomatology, being manifested before the onset of any physical symptoms and contributing to delay in language acquisition. Furthermore, the same study indicated that reasoning deficits are non-progressive when met with age-dependent parameters. Overall, reasoning deficits are frequent, albeit not universally observed.

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The concept of intelligence represents general cognitive ability, which means that an intelligence deficit is not specific enough to lay down a neuropsychological profile (Schelini, 2006). It is also documented that DMD patients might present cognitive impairment without having any global deficit (Battini et al., 2018). Overall, DMD patients have shown low to average range performance in neuropsychological instruments that measure selective attention and inhibition. It has been suggested that such patients have difficulties with tasks that require adaptability and frequently present impulsive behavior (Anikiej et al., 2018).

In accordance with Anikiej et al. (2018), a study by Battini et al. (2018) similarly concluded that even in boys with no intellectual impairments, DMD exerts notable effects on cognition. This study demonstrated that DMD patients with no intelligence deficits performed below average on tasks that required planning and rule-following. Additionally, patients performed below-average on WISC-IV's Digit Span and Letter Number Sequencing subtests, resulting in a Working Memory Index below the average range. The same pattern emerged among patients with low IQ.

In Battini et al. (2021), researchers recruited DMD patients with no reasoning deficits, revealing that, despite the absence of such deficits, executive functioning remained below average. Notably, working memory tests such as WISC-IV's Digit Span and Letter and Number Sequencing, NEPSY-II maintenance set, and Modified Wisconsin Card Sorting Test (rule violation) indicated deficits.

In measures that demand visual and verbal learning, individuals with DMD typically exhibit a similar performance to their non-DMD peers. This indicates that attention to the stimuli, memorization, long-term storage, recall, and recognition abilities on pictorial tasks are intact. However, concerning episodic memory tasks such as story recall and verbal digit span tasks, DMD children performed substantially worse. Such a discrepancy implies that while their visual working memory remains relatively preserved, verbal memory is significantly impaired. This observation suggests that the verbal working memory impairment stems from a reduced capacity within the phonological loop of working memory (Hinton et al., 2001; Wingeier et al., 2011). The same pattern was observed by Anikiej et al. (2018) and Thangarajh et al. (2020), who remarked that DMD children performed poorly in working memory tasks and on the selective aspect of inhibitory control, indicating difficulties in following rules when presented with other stimuli and a propensity to forget instructions during tasks.

It must be considered that living with motor disabilities may also impact social, emotional, and cognitive development, which may be intensified by issues such as social isolation, cardiovascular or respiratory pathologies, and hormonal dysfunctions (Anikiej et al., 2018; Thangarajh et al., 2020). Despite limited knowledge of psychological and social outcomes, some research has been conducted regarding comorbidities. Attention deficit hyperactivity disorder (ADHD) was found to be the most common neurobehavioral comorbidity associated with DMD, affecting as muny as 50% of patients (Hinton et al., 2006; Steele et al., 2008). Thus, studies suggest that attentional and executive functioning deficits may be common in DMD.

Pane et al. (2012) found that 32% of the DMD sample met the criteria for ADHD, as opposed to 3% among the general population. Higher rates of Autism Spectrum Disorder and Obsessive-Compulsive Disorder were also observed in this population (Poysky, 2007). The development of theory of mind and social skills may also be affected (Crowe et al., 2017). Further studies suggest that boys with DMD are at higher risk of developing learning disabilities, as they have also shown deficits in phonological awareness and processing. Such information may contribute to the explanation of the higher rates of reading problems, which could be as high as 40% for boys with DMD (Leibowitz & Dubowitz, 1981; Billard et al., 1998). Overall, studies suggest that while visuospatial abilities remain relatively preserved, verbal and executive functions show consistent deterioration, even in the absence of global intellectual deficits.

Despite the fact that DMD is a severe and incapacitating disorder, it should not impede affected individuals from leading eventful and fruitful lives (Birnkrant et al., 2018). In order to properly aid the child to do so, it is recommended that the health professional should adopt a rehabilitation approach. In this regard, rehabilitation is defined as a process achieved by an active change, where the person who is disabled can accomplish an optimal lifestyle through the acquisition of proper social, physical, and psychological skills (Wilson et al., 2017).

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Furthermore, a fundamental aspect of elaborating a good rehabilitation program is a comprehensive neuropsychological assessment (Wilson et al., 2017). Such an assessment involves a systematic collection, categorization, and interpretation of the patient's functioning (Wilson et al., 2017). A comprehensive assessment may provide information on the deficits associated with the greatest day-to-day challenges and intervention possibilities, while looking beyond psychometric-restricted analyses (Cristiano et al., 2022). Additionally, a neuropsychological assessment may identify coping strategies, psychological factors associated with the difficulties, and family support (Wilson et al., 2017).

While international literature has documented general cognitive deficits in DMD, analyses at the individual level remain scarce. To contribute to this line of research, we present below the neuropsychological assessment of an adolescent with DMD, aimed at understanding his cognitive profile and guiding educational support strategies. Thus, the present study presents a case report of a neuropsychological assessment of a Brazilian boy with DMD. We aimed to identify his strengths and weaknesses in order to establish a guideline for parents and the school, which could potentially enhance the child's quality of life.

MATERIALS, METHODS, AND PROCEDURES

The present study is a case report, entailing a comprehensive description and discussion of a neuropsychological assessment, in accordance with the guidelines set forth by the Brazilian National Committee on Research Ethics (CONEP; Brasil, 2018). The information regarding the present study was obtained during the routine practice of a professional neuropsychologist, as usual in case reports (Minayo, 2001).

Clinical Case

John (pseudonym) is a 14-year-old boy diagnosed with DMD. Two out of his three brothers were also diagnosed with DMD. John presented no immediate post-birth abnormalities, and he was able to develop his early motor skills within the expected timeframe. He started walking at 15 months, and there was no delay in his speech development. Mobility difficulties began when John was around the age of 7 and have been progressing ever since, necessitating wheelchair use for mobility assistance. He also showed limitations in gross motor and day-to-day skills such as jumping, running, self-dressing, and maintaining balance.

John was referred to the clinical practice, where the assessment took place, by his school. Both the family and the school requested the neuropsychologist to identify the reasons associated with the academic difficulties. This information was needed to determine whether school adaptations or enrollment in special classes were necessary. The patient uses medication to control low blood pressure and supplements such as calcium and vitamins, and is reported to have chronic muscular pain. John reported having school difficulties, particularly with math and reading comprehension, often requiring assistance from teachers or parents to complete school tasks. He is an affectionate boy with no observed limitations in the social domain aside from those related to his mobility limitations.

Instruments

The assessment protocol was carefully selected, considering John's mobility issues. The psychometric segment of the assessment was tailored to discern preserved and deficitary functions. Cognitive functions such as intelligence, attention, memory, and executive functioning were assessed with the use of neuropsychological tasks. Since a psychometric approach may be insufficient in providing enough information for a rehabilitation plan (Wilson et al., 2017), scales and ecological tasks served as complementary instruments.

The neuropsychological assessment encompassed various standardized instruments. The Wechsler Intelligence Scale for Children (WISC-IV; Wechsler et al., 2011) served as a comprehensive battery of multiple cognitive functions to derive coefficients such as the intelligence coefficient (IQ) and domain-specific coefficients (verbal comprehension, perceptual reasoning, working memory, and processing speed indices). We employed the Visual Attention Test-4 (TAVIS-4; free

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translation; Mattos et al., 2018) and the d2-R (Brickenkamp et al., 2018) to assess attention, requiring the patient to provide accurate responses following the identification of stimuli across contexts of sustained, selective, and alternating attention.

Episodic memory was evaluated using the Rey Auditory Verbal Learning Test (RAVLT; de Paula & Malloy-Diniz, 2018), involving the repeated reading of a word list followed by immediate and delayed recall attempts. Rey-Osterrieth Complex Figure Test (Rey et al., 2014) was used to assess nonverbal memory, involving the drawing of a complex figure through copying and subsequently from memory. The Five Digit Test (FDT; Sedó et al., 2015) was used to assess executive functioning, particularly inhibition and cognitive flexibility, requiring the patient to inhibit distractions in incongruent stimuli and to rule-switch. Executive functioning was further assessed with a verbal fluency test (Fonseca et al., 2016).

The School Achievement Test II (TDE II; free translation; Stein et al., 2018) was used to assess academic performance and to compare it with the expected score for the patient's age. Additionally, the Behavior Rating Inventory of Executive Function 2 (BRIEF 2; Gioia et al., 2015) was utilized to assess day-to-day executive functioning, while the Assessment Behavior System-3 (ABAS-3; Harrison & Oakland, 2015) was employed to investigate the patient's autonomy and adaptive skills, assessing conceptual (functional communication, academic skills, and self-direction), social (leisure and socialization) and practical (daily life, self-care, health, and safety) domains. We considered norm-referenced scores from each test according to the manual, encompassing percentiles, standard scores, T scores, and z scores. When not specified in the manual, we adopted the American Academy of Clinical Neuropsychology consensus to interpret results (Guilmette et al., 2020).

Ethical Considerations

Written consent was obtained from the participant and his parents, in accordance with the guidelines of the National Research Ethics Committee for situations that emerge spontaneously and contingently in professional practice (CONEP, 2016). The participant's privacy, data confidentiality, and human dignity were preserved.

Procedures

The assessment occurred in a private clinical practice in a quiet room free of visual and auditory distractions. We conducted a total of nine sessions, each lasting 50 minutes, encompassing anamnesis, testing procedures, and subsequent feedback sessions to the family and the school. Following the signing of informed consent, we seated John at the desk for interviews and testing sessions, which a senior neuropsychologist (L.B.C.) and an assistant psychologist (G.M.S.) administered.

RESULTS

Norm-referenced scores are presented in Table 1. The results for different cognitive functions are presented within different sections (reasoning, executive functioning, attention, memory, and language).

Reasoning

A standard deviation (SD) of >1,5 was found between WISC-IV indices, hindering the interpretation of the IQ score as a unified measure. General Intelligence by WISC-IV results had a >1,5 SD variability between indices, which makes it impossible to interpret the results as a unique measure of IQ. Therefore, we used General Ability Index (GAI) and Cognitive Proficiency Index (CPI) analyses. The GAI represents intelligence (both crystalized and fluid) while disregarding attention and motor functions. John obtained a GAI score of 103 (97-109; 95% C.I.), indicating a preserved capacity to reflect and select responses in problem-solving situations.

Attentional deficits impacted his general performance and may impact his learning, working memory, and processing speed. Such difficulties, classifiable as borderline intellectual functioning (CPI=79; 73-88; 95% C.I.), impose significant deficits regarding academic skills. Furthermore, despite the attention deficits that skewed John's performance to the point of hindering interpretation, motor deficits were also below average. Given the Processing Speed Index (PSI) on WISC-IV demands motor skills, John required more time to complete the task, as observed in other tasks that demanded the use of a pencil and paper.

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Table 1.
John's neuropsychological assessment scores

Test	Measure	Score	Classification
WISC-IV (Wechsler Intelligence Scale for Children-IV)	Verbal comprehension index (VCI)	104	Average
	Perceptual reasoning index (PRI)	102	Average
	Working memory index (WMI)	80	Low average
	Processing speed index (PSI)	83	Low average
	Full-scale IQ (FS-IQ)	92	Average
FDT (Five Digit Test)	Reading	25-50%	Average
	Counting	25-50%	Average
	Choosing	25-50%	Average
	Alternating	50-75%	Average
	Inhibition score	25-50%	Average
	Flexibility score	50-75%	Average
d2-r	Concentration performance	89	Low average
	Speed	86	Low average
	Acuracy	97	Average
ROCFT (Rey-Osterrieth Complex Figure Test)	Copy	<10%	Below average
	Short term memory	<10%	Below average
RAVLT (Rey Auditory Verbal Learning Test)	A1 – first attempt	<5%	Below average
	B1 – disctraction list	25-50%	Average
	A6 – post-distraction	25%	Average
	A7 – delayed recall	75%	High average
	Recognition	95%	Above average
TDE II (School Performance Test-II)	Writting	<1%	Exceptionally low
	Written efficiency	1%	Exceptionally low
	Words written per minute	10%	Low average
	Reading	80%	High average
	Reading efficiency	5%	Below average
	Reading per minute	1%	Exceptionally low
	Maths	<1%	Exceptionally low
	Maths efficiency	1%	Exceptionally low
Verbal Fluency test	Phonemic	z=0,97	High average
	Semantic	z=1,28	High average

Note: Scores accompanying "%" indicate percentile. Scores accompanying z indicate z-score. The remaining scores indicate a composite score (where 100 is the mean). IQ=intelligence quotient. All scores were calculated based on normative data and interpreted according to Guilmette et al. (2020).

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Executive functioning

John exhibited the ability to inhibit irrelevant information and displayed sufficient cognitive flexibility to change behavior patterns according to environmental demands. However, he has demonstrated that attentional lapses indeed impacted his working memory capacity, causing him to forget information on how to execute a task. These findings reflected the manifestation of perseveration behaviors and inefficient information decoding, causing frequent planning errors. To compensate for the difficulties, we suggested repetition strategies to promote enhanced engagement in the task and memory consolidation. John exhibited high average performance in the verbal fluency task, thus demonstrating the ability to organize words in clusters, and additionally presented average scores in the FDT.

Attention

John had an average performance on the selective attention task, which is the capacity to identify stimuli amidst irrelevant ones, but he had significant signs of attentional fluctuations during the assessment. Behavioral observations suggested that those fluctuations were attributable to fatigue and demotivation. As expected, his errors increased in frequency when his sustained attention was recruited. We also observed target-type errors, associated with inhibitory control failure, and omission-type errors, associated with inattention.

Memory

John displayed an enhanced performance when presented with more concrete information, such as visual stimuli, in contrast to tasks solely reliant on auditory input. We hypothesized that the patient possessed greater proficiency in the visuospatial loop of working memory compared to the phonological loop. Furthermore, he demonstrated a satisfactory learning capacity but borderline performance in a coding task requiring immediate memory. This performance allowed the inference that he may require more exposure time to the same stimuli to achieve a satisfactory performance.

Considering his difficulty in keeping information in his short-term memory, it was advised that the school should allow John to take longer to transcribe information from the blackboard. Moreover, we advised teachers to redundantly administer supplementary instruction repetitions, thus creating a verbal and visual pathway, especially considering his greater proficiency in the latter. We proposed a top-down methodology, wherein an overview of the content could be presented both before and after the explanation of the concepts. Without such accommodation, John may struggle to retain and properly connect information due to his limited short-term memory capacity.

Inattention-type errors represented a mild impact on his learning skills. Despite this, he could store information and subsequently was able to consolidate it in his long-term memory. It is possible to infer that with more repetitions and time, he could learn and retrieve the information when required. Notably, his performance substantially improved when presented with a priming stimulus, indicating that multiple-choice activities in school or day-to-day life might be advantageous for John, because even when he knows the content, he may fail to retrieve the information voluntarily.

Language

John presented an adequate ability to use words and to define concepts. However, there were difficulties related to connecting ideas in unfamiliar contexts. His verbal comprehension performance also indicated difficulties. However, such difficulties were considered secondary to deficits in attention and short-term memory, causing a lack of initial comprehension of the stimuli and a reduced ability to keep the information in mind in order to understand the meaning of complex phrases.

Academic skills

In the assessment of vocabulary, John presented a good performance, but he could not voluntarily access his knowledge in specific situations. Regarding mathematical skills, he performed poorly, not showing proper reasoning and comprehension to solve basic math operations and interpret written problems.

He also presented difficulties in expressing himself in speaking and writing. Difficulties were also evident in reading comprehension, but were milder on themes that he had previous knowledge of. In written assessments and in tasks where

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some words were dictated out loud, he struggled with converting phonemes into graphemes. He added syllables and committed various errors in the written phonemes of irregular words and during tasks that required following grammatical norms. However, these errors do not characterize a learning disorder. In terms of reading proficiency, John demonstrated the ability to read, but his reading was slow when compared to the expected level for adolescents in his age group.

Scales

According to the ABAS-3 results in comparison with his peers, John's autonomy was mostly preserved, with a General Adaptive Index of 88. While his conceptual and social domains were found to be average, the practical domain indicated greater challenges. This practical domain covers autonomy for everyday situations, with John's lowest score attributed to domestic life tasks. His motor limitations may impact the execution of some activities. Notably, the social domain was preserved, which could potentially serve as compensation for other abilities in challenging situations. John actively engages in social activities, presents empathic behavior, and can adjust his behavior according to the environment and social norms.

Based on the BRIEF 2 results (answered by the patient's mother), the working memory and the self-monitoring components indicated difficulties. This indicates that John tends to present difficulties in keeping and assimilating information and may be confused during the execution of demanding activities or fail to complete them. Likewise, he may not perceive his own mistakes due to poor monitoring. Alternatively, no skills are below average if only the patient's answers are considered, indicating substantial deficits in monitoring.

DISCUSSION

This report was on par with the international literature. It is argued that even among patients with DMD with no intellectual impairment, the syndrome has an impact on neuropsychological functioning. John did not exhibit impairment in general reasoning; however, he did demonstrate a considerable gap between WISC-IV's Verbal Comprehension Index (VCI=104) and Working Memory Index (WMI=80) scales. Such a discrepancy required further analysis using the GAI. Although DMD is commonly associated with an impaired IQ (Bresolin et al., 1994), the GAI score indicated a normal range for IQ. However, tasks that demanded the manipulation of concomitant information revealed significant difficulties, rendering John fatigued and demotivated. To compensate for such deficits, John developed a slower processing speed.

This pattern can also be observed in executive functioning, in which deficits in organization and planning, especially when engaging with abstract concepts, were evident. WISC-IV analysis further showed that John possessed an adequate crystallized memory for his age but encountered challenges in accessing it voluntarily. According to Bresolin et al. (1994), DMD patients may present diverse impairments, such as in language, attention, and memory functions. It is important to note that such deficits may primarily stem from poor performance in executive functioning, especially working memory tasks (Bresolin et al., 1994).

Battini et al. (2018; 2021) reached similar conclusions in a study that excluded participants with psychiatric or neurodevelopmental comorbidities (e.g., ADHD) and identified that the DMD cognitive impairment may be mostly related to executive functioning, especially when it comes to inhibition, response switch, and planning. Anikiej et al. (2018) found that the most impaired functions in DMD are the selective aspects of executive functioning, as well as working memory, fostering maladaptive behaviors such as reduced motivation and abandoning tasks that are cognitively demanding.

John's performance was greatly enhanced following priming, enabling him to acknowledge concepts in a more concrete way in order to promote learning. In this sense, his attentional network was also compromised, hindering his ability to effectively address problems and filter extraneous stimuli. The present study supports the notion that executive functioning may be central to the understanding of the cognitive impairments in DMD, especially in terms of working memory, as also observed by Tyagi et al. (2021).

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In the field of cognitive impairments associated with DMD, it is imperative to recognize that this syndrome impacts not only cognition but also motor skills and the family routine. Therefore, it is essential to consider the lack of cognitive stimulation, especially during early infancy, along with the lack of ability to communicate with peers in a regular school setting. Poor environmental stimulation due to the symptoms may contribute to the observed cognitive impairment (Cabrera et al., 2020). Given his symptomatology, it is possible to categorize John's case as compatible with Attention Deficit/Hyperactivity Disorder (ADHD; American Psychiatric Association, 2022), thereby confirming international findings.

It is important to conduct further studies in the field of DMD. We suggest that comprehensive neuropsychological assessments could provide greater insight into symptom remission associated with emerging therapeutic approaches, such as the genetic treatment described by Mendell et al. (2020). Although this intervention demonstrated clinical efficacy, no neuropsychological data were collected (Mendell et al., 2021). Evidence suggests that medical treatments can lead to improvements in executive functioning and alterations in personality among individuals with motor disorders (Arten & Hamdan, 2025), reinforcing the need for continued investigation in this field.

CONCLUSIONS

Childhood represents a critical phase for neurodevelopment, with the brain deeply influenced by the stimuli it encounters. Therefore, a comprehensive neuropsychological assessment of children with neuromuscular conditions holds significant potential for promoting their development. DMD is associated with a lower IQ and a deficit in executive functioning, especially regarding working memory and monitoring. In the present case report, the observed neuropsychological profile aligns with findings in the international literature, highlighting how DMD may hinder academic achievement and language skills due to poor executive functioning.

Conflicts of interest

The authors declare that there are no conflicts of interest related to this study.

Taxonomía de contribuciones autoría CRediT (Contributor Roles Taxonomy)

Luana Breda: contributed to conceptualization, resources, methodology, project administration, and supervision.
Genner Mateus Secco: contributed to formal analysis, methodology, investigation, and writing (original draft).
Gabriel Sousa Andrade: contributed to writing (original draft, review, and editing).

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