Neuromuscular assessment form (NAF): pediatric outpatient follow-up experience from a single Brazilian center

Formulário de avaliação neuromuscular: experiência de seguimento ambulatorial de um centro Brasileiro

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ABSTRACT

Background: Neuromuscular disorders are a heterogeneous group of genetic or acquired diseases affecting the anterior horn, peripheral nerves, neuromuscular junctions, or muscles. Given their multisystemic complexity, comprehensive care and standardized evaluation tools are essential.
Objective: To describe the development, implementation, and utility of the Neuromuscular Assessment Form (NAF) as a tool for integrated clinical evaluation in patients with NMDs at a Brazilian reference center.
Methods: A descriptive observational study was conducted using retrospective data from patients followed over the past thirty years at a reference center. The NAF was designed based on literature review, national therapeutic guidelines, and consultations with multidisciplinary specialists in pediatric neurology, physiotherapy, speech therapy, and other support areas.
Results: The NAF enabled the systematic documentation of motor and multisystemic data, including cardiac, respiratory, nutritional, orthopedic, and neuropsychiatric functions. It facilitated interdisciplinary interventions and ensured continuity of care.
Conclusion: The NAF is a practical and effective tool for standardized clinical evaluation and longitudinal follow-up of patients with NMDs. It has the potential to promote equitable care and enhance research across different regions, especially in resource-limited settings. Future validation and integration into electronic health systems may further improve its applicability and impact.

Keywords: neuromuscular manifestations, neuromuscular diseases, muscular dystrophy, spinal muscular atrophy

RESUMO

Introdução: Os distúrbios neuromusculares são um grupo heterogêneo de doenças genéticas ou adquiridas que afetam o corno anterior, os nervos periféricos, as junções neuromusculares ou os músculos. Dada a sua complexidade multissistêmica, o cuidado integral e as ferramentas de avaliação padronizadas são essenciais.
Objetivo: Descrever o desenvolvimento, a implementação e a utilidade do Formulário de Avaliação Neuromuscular (FAN) como ferramenta para avaliação clínica integrada em pacientes com DNMs em um centro de referência brasileiro.
Métodos: Um estudo observacional descritivo foi conduzido utilizando dados retrospectivos de pacientes acompanhados ao longo dos últimos trinta anos em um centro de referência. O FAN foi elaborado com base em revisão de literatura, diretrizes terapêuticas nacionais e consultas com especialistas multidisciplinares em neurologia pediátrica, fisioterapia, fonoaudiologia e outras áreas de apoio.
Resultados: O FAN permitiu a documentação sistemática de dados motores e multissistêmicos, incluindo funções cardíacas, respiratórias, nutricionais, ortopédicas e neuropsiquiátricas. Facilitou intervenções interdisciplinares e garantiu a continuidade do cuidado.
Conclusão: O FAN é uma ferramenta prática e eficaz para avaliação clínica padronizada e acompanhamento longitudinal de pacientes com DNMs. Tem o potencial de promover cuidados equitativos e aprimorar a pesquisa em diferentes regiões, especialmente em ambientes com recursos limitados. A futura validação e integração em sistemas eletrônicos de saúde podem aprimorar ainda mais sua aplicabilidade e impacto.

Palavras-chave: manifestações neuromusculares, doenças neuromusculares, distrofia muscular, atrofia muscular espinal

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INTRODUCTION

Neuromuscular disorders (NMDs) are a heterogeneous group of disorders, either inherited or acquired, resulting in abnormalities that affect the anterior horn of the spinal cord, peripheral nerves, neuromuscular junction, or muscles function. They share clinical similarities, such as reduced muscle strength, reduced reflexes. Some present myotonia, pain, fatigue or trophic changes. Most lead to joint contractures and cardioventilatory dysfunction and some have neuropsychiatric comorbidities. In general, there is a progressive decline in motor/bulbar abilities and cardiorespiratory dysfunction over time. The most prevalent NMDs worldwide are Duchenne muscular dystrophy (DMD) (0.7–4.7 per 100,000), Becker muscular dystrophy (BMD) (0.07–3.65 per 100,000), and spinal muscular atrophy (SMA) (1.3–3.2 per 100,000), those three with first signs and symptoms in childhood.[1-3]

Our neuropediatric outpatient clinic was established in mid-1989 and since then, has followed more than 300 patients with neuromuscular disorders. A multidisciplinary team, including pediatric neurologists' cardiologists, pulmonologists, nutritionists, physiotherapists, speech therapists, and social workers, manages the systemic comorbidities. The current scientific literature provides a range of consensus and recommendations to guide optimal clinical care for patients with NMDs, highlighting the importance of a multidisciplinary approach and addressing systemic dysfunctions.[4-11] Given the complexity of these diseases, our team has developed the clinical and multidisciplinary evaluation here presented, the Neuromuscular Assessment Form (NAF). Although our center has a 30-year database, the NAF was created ten years ago, and has been used since then, to standardize medical data and reduce follow-up gaps in multisystem comorbidities. It is continually updated based on the latest current guidelines. The NAF is used in the routine practice of our outpatient clinic at Instituto de Puericultura e Pediatria Martagão Gesteira (IPPMG) of Universidade Federal do Rio de Janeiro (UFRJ). The main goal of this paper is to present NAF as an instrument to comprehensive and integrated care of patients with rare diseases, a guide to help other services in those patients care. This tool might help not only to improve follow-up monitoring of those patients, but also as a backbone for observational multicenter research initiatives.

METHODS

The development of this instrument occurred step by step. One of us (APQCA) as a previous member of the Treat-NMD committee (https://www.treat-nmd.org/what-we-do/global-registry-network/), became familiarized with the core dataset necessary to be implemented on a local registry. Those first variables became the initial ones to compose the present tool. To put our service in context and to determine which diseases are more prevalent in our practice, a descriptive observational study was conducted, utilizing a historical overview of clinical records from patients followed over the past thirty years at our center.[4-7,12-32]

As knowledge and care recommendations in NMDs are continually updating, literature review and consultations with experts in patient support areas such as pediatric neurology, pediatrics, motor and respiratory physiotherapy, and speech therapy were conducted to assure that NAF encompassed fundamental health data for patients follow up. The non-systematic review employed databases (PubMed, SciELO, UpToDate, Google Scholar), using the following search terms: 'Duchenne Muscular Dystrophy,' 'DMD progression and treatment,' 'DMD guidelines and management,' 'Spinal Muscular Atrophy,' 'SMA guidelines,' 'SMA 5q subtype management,' 'Neuromuscular disorders management,' 'Multisystemic approach in neuromuscular disorders,' 'Interdisciplinary care in neuromuscular disorders, 'Consensus on DMD treatment,' and 'Clinical guidelines for SMA,' within the last 15 years.

This study was approved by the Ethics and Research Committee of IPPMG on 08/21/2012, under CAAE number 01934112.0.0000.5264

RESULTS

1. Pediatric Neuromuscular Center – IPPMG/UFRJ

The Pediatric Neuromuscular Disorders Center at IPPMG-UFRJ was started over 30 years ago by pediatric neurologist Alexandra Prufer de Queiroz Campos Araujo and has been monitoring more than 300 children and adolescents since then. Currently, it operates with a multidisciplinary team consisting of physicians (pediatricians, neurologists, cardiologists, pulmonologists), nutritionists, social workers, physiotherapists, and speech therapists. Patients are referred to us from primary healthcare units or by other colleagues from Rio de Janeiro and neighboring states. Additionally, IPPMG-UFRJ offers a two-year pediatric neurology residency program, admitting eight junior physicians annually from various Brazilian states. All residents actively participate in completing the NAF during outpatient consultations. Table 1 summarizes the main neuromuscular disorders monitored at our center over the past thirty years.

The core dataset, those that were the mandatory ones for Duchenne Muscular Dystrophy and Spinal Muscular Atrophy in the first version of the Treat-NMD registry can be found in https://www.treat-nmd.org/wp-content/uploads/2023/10/DMD_core_dataset_May-2013.pdf and in https://www.treat-nmd.org/wp-content/uploads/2023/08/v0-SMA-Core-Dataset.pdf.

Table 1. Pediatric NMDs overview at IPPMG-UFRJ

Pediatric NMDs	Disease	Number of patients	Diagnostic Exams	Treatment available in Brazil
1. **Muscular Dystrophies**	Duchenne/Becker Muscular Dystrophy • Gene: *DMD* • Protein: Dystrophyn	196	• Screening test: creatine phophokinase (CPK) • Genetic-molecular test (MLPA /Sequencing) • Immunohystochemical analysis in muscle bipsy	• Clinical care according to Brazilian consensus[4,5,6] • Ataluren approved by National Medicine Agency*, but is only available by judicial decision. • Genetic therapy approved by National Medicine Agency*, but is only available by judicial decision.
	Limb-Girdle Muscular Dystrophy	9	• Screening test: creatine phophokinase (CPK) • Genetic-molecular test (MLPA /Sequencing) • Immunohystochemical analysis in muscle bipsy	• Conservative approach with periodic exams (Echocardiogram, electrocardiogram, spirometry, seric vitamin D, spine panoramic x-ray, and rehabilitation)
2. **Spinal Muscular Atrophy (SMA)**	SMA type 1 • Gene: *SMN1* • Disease Modyfier: SMN2 • Protein: SMN	23	• Newborn screening test: not available in Rio de Janeiro, only in some Brazilian states* • Genetic-molecular test (MLPA /Sequencing)	• Clinical care according to Brazilian consensus[7] • Nusinersen and Risdiplam available on Public Health System[12] • Genetic therapy approved by National Medicine Agency**, but is only available by legal decision. In 2025 approved for use up to the age of 6 months SMA type 1 on Public Health System
	SMA type 2	31	• Genetic-molecular test (MLPA /Sequencing)	• Clinical care according to Brazilian consensus[7] • Nusinersen and Risdiplam available on Public Health System[12] • Genetic therapy approved by National Medicine Agency*, but is only available by judicial decision.
	SMA type 3	22	• Genetic-molecular test (MLPA /Sequencing)	• Clinical care according to Brazilian consensus[7] • None Disease Modifying Therapy approved.
3. **Congenital Muscular Dystrophies**	LAMA2 Deficiency	4	• Genetic-molecular test (MLPA /Sequencing)	• Clinical care according to International consensus[14,15]
	Dystroglycanopathies related (αDG)	1		
4. **Congenital Myopathies**	Central core myopathy	3	• Genetic-molecular test (MLPA /Sequencing)	• Clinical care according to International recommendations[16]
5. **Myasthenic syndromes**	Autoimmune Juvenile Myasthenia gravis	8	• Autoantibodies targeting the AChR (acetylcholine receptor) • Repetitive nerve stimulation (RNS) and single-fiber electromyography • Pyridostigmine testing	• Cholinesterase inhibitors (ChE-I): Pyridostigmine • Immunosuppressive therapy: prednisolone, azathioprine, Rituximab, Cyclosporine • Clinical care according to national recommendations[17-18]
	Congenital Myasthenia	1	• Genetic-molecular test (MLPA /Sequencing)	• Clinical care according to international recommendations[19]
6. **Myotonic Syndromes**	Myotonic Dystrophy type 1	2	• Genetic-molecular test (PCR)	• Clinical care according to international recommendations[8,20]
7. **Metabolic Myopathies**	Palmitoil Carnitine Transferase II Deficiency	2	• Enzymatic assay • Genetic-molecular test (MLPA /Sequencing)	• Clinical care according to international recommendations[21]
8. **Hereditary sensitive and motor neuropathies**	Charcot Marie Tooth Disease	2	• Neurophysiologic tests • Genetic-molecular test (MLPA /Sequencing)	• Clinical care according to international recommendations[22]
9. **Inflammatory myopathies**	Dermatomyositis	2	• Immunologic tests	• Clinical care according to international recommendations[23]

Source: from author's database

Legend: NMDs: neuromuscular disorders; IPPMG: Instituto de Puericultura e Pediatria Martagão Gesteira; UFRJ: Universidade Federal do Rio de Janeiro; MLPA: Multiple Ligand Probe Amplification; PCR: polymerase chain reaction; *Newborn screening test Brazilian states link: https://www.gov.br/ebserh/pt-br/comunicacao/noticias/diagnostico-precoce-e-tratamento-especializado-ajudam-a-conter-a-atrofia-muscular-espinhal; **National Medicine Agency (ANVISA).

2. Neuromuscular Assessment Form (NAF)

According to global consensus on various NMDs and experts' recommendations, neuromuscular clinical care should go beyond motor decline, and encompass cardiac, respiratory, gastrointestinal, nutritional, orthopedic, and neuropsychiatric dysfunctions. Therefore, the comprehensive and integrated care of patients with rare diseases in a university hospital and reference centers has become a huge challenge. The Neuromuscular Assessment Form (NAF) was established to standardize and organize medical data, and to minimize follow-up gaps regarding the multisystemic comorbidities.

The NAF for pediatric patients records clinical data related to diagnosis, treatment, and periodic evaluations, such as (see Table 2):

- **Diagnosis:** clinical phenotype, classification into subtypes (if applicable), genotype, prognostic markers.
- **Diagnostic tests:** biochemical screening tests, genetic-molecular tests, neurophysiological tests, and results of muscle immunohistochemical evaluation.
- **Growth data:** anthropometric data, immunizations, nutrition.
- **Neurodevelopment milestones and neurocognitive profile:** school learning and neuropsychiatric disorders.
- **Cardiorespiratory function evaluation:** signs/symptoms of hypoventilation, need for ventilatory support, cough assistance, pulmonary function tests, echocardiogram, Holter, and ambulatory blood pressure monitoring (MAPA).
- **Motor function evaluation:** best motor milestone, use of orthoses, wheelchair, rehabilitation therapies (physiotherapy, hydrotherapy, occupational therapy), and orthopedic surgeries.
- **Physical and neurologic examination:** vital signs, motor scales, and semiology of the motor system.

Table 2: NAF (Neuromuscular Assessment Form)

Identification:				**Date:**	**Age:**
Neuromuscular Disorder				**Phenotype/Subtype**:	
Prognostic Markers	**Loss of ambulation (LoA) (age): _____**	**Ventilatory support (age of start) NIV: _____ IV: _____**		**Nutritional Support (age of start)**	**Nasoenteral/ Nasogastric tube (age):_____** / **Gastrostomy (age):_____**
Diagnostic Exams	Creatine phosphokinase (CPK):			Anti-AChR:	
	Genetic Test:	Gene:	Exon:	Localization:	Genetic notation:
	Muscle biopsy:			Electroneuromyography (ENMG):	
Clinical Complaints					
Medications in use					
Immunization	23-valent pneumococcal: y ☐ n ☐	Influenza: y ☐ n ☐	Palivizumab: y ☐ n ☐	Other:	
Growth	Weight (kg): _____	Height (m): _____	Ulna length boys (H = 4,605Ulna + 1,308Age + 28,003) = _____ Ulna length girls (H = 4,459Ulna + 1,315Age + 31,485) = _____	Head Circumference (cm): _____	
	Chest Circumference on Exhalation(cm): _____	Chest Circumference on Inhalation(cm): _____	Thoracic Index: _____ (Axillary thoracic diameter / Diaphragmatic thoracic diameter)		
Gastrointestinal Function/ Nutritional Care	***Blood Levels*** Ferritin: _____ Carnitine free/total: _____ Vitamin A: _____ Vitamin D: _____ Vitamin E: _____ Calcium: _____	***Gastroesophageal Reflux***: Frequency: _____ Consistency: _____ ***Emesis***: Frequency: _____ Colour: _____		***Abdominal Pain:*** Frequency: _____ Localization: _____ ***Intestinal Function:*** Frequency: _____ Consistency: _____	

<table>
<tr><td>Bulbar Function</td><td colspan="2">Feeding Route:
Oral: y ☐ n ☐
liquid ☐ pureed ☐ solid ☐
Feeding duration (minutes): _____
Maximal Mouth Opening (cm): _____
Nasogastric/Nasoenteral tube: y ☐ n ☐
Gastrostomy: y ☐ n ☐</td><td colspan="2">Dysphagia signs/symptoms:
Swallowing difficulties/? y ☐ n☐
Interruptions of feeding? y ☐ n☐
Sweating? y ☐ n☐
Choking? y ☐ n☐
Cough? y ☐ n☐
Nasal/Oral Reflux? y ☐ n☐</td><td>Expressive Communication/Language
Crying: weak☐ medium☐ strong☐
“Wet” voice: y ☐ n☐
Vocalization: y ☐ n☐
Babling of consonants: y ☐ n☐
First meaninful words: y ☐ n☐
Complete sentences: y ☐ n☐</td></tr>
<tr><td rowspan="2">Neuro-psychiatric/ Cognitive Function</td><td>School grade: _____</td><td>Learning disability:
y ☐ n ☐</td><td>Educational support: y ☐ n ☐</td><td>School start: _____
Drop out of school:_____</td></tr>
<tr><td colspan="4">Comorbidities: ID: y ☐ n ☐ ADHD: y ☐ n ☐ ASD: y ☐ n ☐ Anxiety: y ☐ n ☐ Depression y ☐ n ☐
Other:_____</td></tr>
<tr><td rowspan="2">Respiratory Function</td><td>FVC: y ☐ ___ n ☐
Value (%): _____</td><td>Nocturnal polysomnography: y ☐ n ☐
_____</td><td>Symptoms of nocturnal hypoventilation? y ☐ n ☐
_____
___</td><td>Cough Assist? y ☐ n ☐
Frequency: _____
Upper Airway suctioning
Frequency: _____</td></tr>
<tr><td>Nocturnal NIV?
y☐ (_____h/night) n ☐</td><td>Diurnal NIV?
y☐ (_____h/day) n ☐</td><td>Nocturnal IV?
y☐ (_____h/night) n ☐</td><td>Diurnal IV?
y☐ (_____h/day) n ☐</td></tr>
<tr><td>Cardiac Function</td><td>Echocardiogram: y ☐ n ☐
LVEF (%): _____</td><td>Holter: y ☐ n ☐
Arrhythmias: _____</td><td>ABPM: y ☐ n ☐
Arterial Hypertension: y ☐ n ☐</td><td>Cardiac resonance: y ☐ n ☐
Cardiac fibrosis:_____</td></tr>
<tr><td>Orthopedic Care</td><td colspan="2">Ankle Foot Orthosis (AFO):
nocturnal ☐ duration: _____ diurnal ☐ duration: _____
Knee Foot Orthosis (KAFO):
nocturnal ☐ duration: _____ diurnal ☐ duration: _____
Wrist/Hand Orthesis: y ☐ n ☐
_____
Thoracic Brace: y ☐ n ☐
_____
Parapodium: y ☐ n ☐
_____</td><td colspan="2">Spinal Panoramic Radiograph:
y ☐ n ☐ Fergusson angle: _____ Cobb angle: _____
Microfractures of vertebral bodies: y ☐ n ☐
Bone densitometry hip or femur: y ☐ n ☐ Z score: _____
Bone fractures? y ☐ When: _____ n ☐
Orthopedic surgery y ☐ n ☐</td></tr>
</table>

<table>
<tr><td></td><td colspan="3">Manual Wheelchair: y ☐ n ☐
_____
Electric Wheelchair: y ☐ n ☐
_____</td><td></td></tr>
<tr><td rowspan="2">Rehabilitation</td><td>Respiratory Physiotherapy:
y ☐ (_____/week) n ☐</td><td>Motor Physiotherapy:
y ☐ (_____/week) n ☐</td><td>Speech therapy:
y ☐ (_____/week) n ☐</td><td>Aquatic Physiotherapy:
y ☐ (_____/week) n ☐</td></tr>
<tr><td>Occupational therapy:
y ☐ (_____/week) n ☐</td><td>Psychology:
y ☐ (_____/week) n ☐</td><td>Educational Psychologist:
y ☐ (_____/week) n ☐</td><td>Other:</td></tr>
<tr><td>Best Motor Function</td><td>☐ Cervical Support
☐ Roll
☐ Sit with Support
☐ Sit without Support</td><td>☐ Crawl
☐ Stand with Support
☐ Stand without Support</td><td>☐ Walk with Support
☐ Walk without Support
☐ Walk >10 meters without help
☐ Run</td><td>☐ Raise Hands above Head
☐Raise Hands on the Head
☐ Bring Hands to Mouth
☐ Use Hands/Move Fingers</td></tr>
<tr><td rowspan="11">Physical and Neurologic Examination</td><td colspan="2">Read/Write: y ☐ n ☐</td><td colspan="2">Addiction/Subtraction? y ☐ n ☐</td><td colspan="2">Multiplication/Division? y ☐ n ☐</td></tr>
<tr><td colspan="3">Mini-Mental State Examination:</td><td colspan="3">WISC-IV:</td></tr>
<tr><td>SpO_2:</td><td>Respiratory rate:</td><td colspan="2">Heart rate:</td><td colspan="2">Blood arterial pressure:</td></tr>
<tr><td rowspan="2">Respiratory Count</td><td>Lying down:</td><td rowspan="2">Time to rise from the floor (TRF):
_____</td><td colspan="3">10 meters Walking Test:</td></tr>
<tr><td>Sitting:</td><td colspan="3">10 meters Running Test:</td></tr>
<tr><td rowspan="2">Axial</td><td colspan="2">Neck Extensor:</td><td>Trapezius</td><td colspan="2"></td></tr>
<tr><td colspan="2">Neck Flexor:</td><td>Abdominals:</td><td colspan="2"></td></tr>
<tr><td rowspan="6">Upper Limbs</td><td>Deltoid</td><td></td><td rowspan="6">Lower Limbs</td><td>Iliopsoas</td><td></td></tr>
<tr><td>Triceps</td><td></td><td>Gluteus</td><td></td></tr>
<tr><td>Biceps</td><td></td><td>Quadriceps</td><td></td></tr>
<tr><td>Wrist Extensor</td><td></td><td>Biceps</td><td></td></tr>
<tr><td>Wrist Flexor</td><td></td><td>Dorsiflexion</td><td></td></tr>
<tr><td>Finger Extensor</td><td></td><td>Plantar flexion</td><td></td></tr>
<tr><td>Finger Flexor</td><td></td><td></td><td>Ankle angle</td><td></td></tr>
<tr><td colspan="6">Joint Contractures: Shoulder ☐ Elbow☐ Wrist☐ Fingers ☐ Hips ☐ Knees ☐ Ankle☐</td></tr>
<tr><td>Motor Scales</td><td colspan="5">CHOP INTEND: _____ HINE: _____ HFMSE: _____ RULM: _____NSAA: _____
PUL: _____ MFM:_____ Other scale:</td></tr>
<tr><td></td><td colspan="5">Night time sleep hours:</td></tr>
</table>

Additional Information	Time spent on electronic media:
	Addicional tests:
Recommenda-tions	

Source: Nardes F. authorship

Legends: ID: intellectual disability; ASD: autism spectrum disorder; ADHD: attention deficit and hyperactive disorder; FVC: forced vital capacity; LVEF: left ventricular ejection fraction; NIV: Non-invasive ventilation; IV: invasive ventilation; ABPM: arterial blood pressure monitoring; SpO_2: oxygen saturation; Anti-AChR: antibodies against receptor of acetylcholine; CHOP INTEND: Children's Hospital of Philadelphia Infant Test of Neuromuscular Disorders HINE: Hammersmith Infant Neurological Examination; HFMSE: Hammersmith Functional Motor Scale – Expanded RULM: Revised Upper Limb Module NSAA: North Star Ambulatory Assessment PUL: Performance of Upper Limb MFM: Motor Function Measure

3. Research opportunities:

The initial dataset and its further development have laid the backbone for academic and clinical research, resulting in important publications to the field (see table 3).

Table 3. Scientific Research and publications - List of Research Projects and Proponent/Sponsors (from https://plataformabrasil.saude.gov.br)

Publications	Research Project	Proponent/Sponsor
	Neuromuscular Registry	IPPMG/UFRJ
	Spinal Muscular Atrophy 5q types II and III followed in the public health system Registry: an observational cross-sectional study	ROCHE
	Spinal Muscular Atrophy Brazilian panorama	IPPMG/UFRJ
	Risdiplam studies: BN40703 (Rainbowfish), RO7034067 (Firefish and Sunfish)	ROCHE
	PTC 124 phase 3: PTC124_GD-041-DMD and PTC124_GD-020-DMD and extension study	PTC pharmaceutic
	Natural history of DMD on routine care 4658-407	IPPMG/UFRJ
	Retrospective cohort for natural history of SMA type I	IPPMG/UFRJ
	Ataluren experience in Brazilian DMD-nm	IPPMG/UFRJ
	Multicentric SMA registry	IPPMG/UFRJ
	GSK 2402968 for DMD phase 3 and extension	GSK pharmaceutic
Nardes F, Araújo APQC, Russi S, Henriques SFB. **Similar disease progression in nonsense Duchenne muscular dystrophy boys as general natural history: single Brazilian center 15 years registry view**. Eur J Paediatr Neurol. 2024;38:66-73		IPPMG/UFRJ
Becker MM, Nardes F, Dangouloff T, Servais L, Araújo APQC, Gurgel-Giannetti J. **Why should a 5q spinal muscular atrophy neonatal screening program be started?** Arq Neuropsiquiatr. 2024;82(10):e20240123.		IPPMG/UFRJ
Alves BKAMF, Araújo APQC, Nardes F, Ribeiro MG.**Type-1 spinal muscular atrophy cohort before and after disease-modifying therapies**. Arq Neuropsiquiatr. 2024;82(11):e20240211		IPPMG/UFRJ
Alexandra Prufer de Queiroz Campos Araujo, Jonas Alex Morales Saute, Clarisse Pereira Dias Drumond Fortes, Marcondes Cavalcante França Jr, Jaqueline Almeida Pereira, Marcos Ferreira Rebel, Flavia Nardes dos Santos et al. **Update of the Brazilian consensus recommendations on Duchenne muscular dystrophy. Arq Neuropsiquiatr. 2023;81(3):e20230012.**		IPPMG/ UFRJ; UFRS; Unicamp; HC/ USP; FMABC; Rede Sarah; AACD; HCPA; UFMG; Univille; UECE; UFPR; FMRP/ USP.
Alexandra P. Q. C. Araujo, Flavia Nardes, Clarisse P. D. D. Fortes,Jaqueline A. Pereira, Marcos F. Rebel et al. **Brazilian consensus on Duchenne muscular dystrophy. Part 2: rehabilitation and systemic care**. Arq Neuropsiquiatr. 2018;76(7):481-489.		UFRJ; UNISUAM; FHEMIG/ HIJPII/ HJK; APDM; ABDIM; AACD
Nardes F, Araújo APQC, Ribeiro MG. **Mental retardation in Duchenne muscular dystrophy**. J Pediatr (Rio J). 2012;88(1):16-21.		IPPMG/UFRJ
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Araújo, Alexandra; Fortes, Clarisse; Nardes, Flavia; Jorge, Eduardo; Raskin, Salmo.**The role of the pediatrician in suspected neuromuscular diseases in childhood.** REV. RESIDÊNCIA PEDIÁTRICA, v. 13, p. 1-11, 2023.		
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Universidade Federal do Rio Grande do Sul (UFRS); Universidade Estadual de Campinas (Unicamp); Hospital de Clínicas da Universidade de São Paulo (HC/ USP); Centro Universitário Faculdade de Medicina do ABC (FMABC); Rede SARAH de Hospitais de Reabilitação - Associação das Pioneiras Sociais; Associação de Assistência à Criança Deficiente (AACD); Hospital de Clínicas de Porto Alegre (HCPA); Universidade Federal de Minas Gerais (UFMG); Universidade de Joinville (Univille); Universidade Estadual do Ceará (UECE); Universidade Federal do Paraná (UFPR); Faculdade de Medicina de Ribeirão Preto da Universidade de São Paulo (FMRP/ USP); Centro Universitário Augusto Motta (UNISUAM); Fundação Hospitalar do Estado De Minas Gerais (FHEMIG); Hospital Infantil João Paulo II (HIJPII); Hospital Julia Kubitschek (HJK); Associação Para Desenvolvimento Social dos Municípios do Estado de Mato Grosso (APDM); Associação Brasileira de Distrofia Muscular (ABDIM)

DISCUSSION

The NAF has enabled us to organize clinical data over the last ten years, improving better neuromuscular assistance to patients and providing valuable natural historical information for scientific purposes. The form was structured into eleven systemic sections of anamnesis (medications/immunizations, growth, neurocognitive, nutritional, bulbar, respiratory, cardiac, orthopedic, rehabilitation and best motor function). It facilitates a comprehensive assessment of health-related complaints and allows longitudinal monitoring of disease progression. Graduate students as well as residents can improve their skills by following this tool on patient out-clinic consultations.

The first step in approaching a child with a suspected neuromuscular disorder includes detailed history and physical examination. Some relevant information from history and clinical signs can be described in table 4 below. All these signs, symptoms and neurodevelopmental milestones are registered in the first clinical encounter with the overall collection of history and neurological physical examinations.

Table 4: Key clinical features in pediatric neuromuscular disorders

History	Symptoms	Physical Examination
Prenatal	***Motor symptoms***	***At birth***
• Pregnancy complications?	• Muscle weakness: difficulty getting up from floor (Gower's sign), climbing stairs, upper limb reaching above head, inability to keep up with peers;	• Hypotonia
• Decreased fetal movements?	• Gait abnormality: toe walking; waddling gait; frequent falls, slowed walking/running	• No spontaneous movements/decreased movements/oligodramnia
• Polyhydramnios?	• Endurance: fatigability, diurnal variation	• Orthopedic deformities (arthrogryposis)
Posnatal	• Disconfort: cramps, stiffness, pain	• Respiratory failure
• Delivery complications?	***Sensory symptoms***	• Bulbar failure
• Preterm delivery?	• Numbness, paresthesia	***From birth to 2 years***
• Hypotonia on initial presentation?	***Respiratory symptoms***	• Hypotonia
• Extremity deformities at birth such as arthrogryposis?	• Breathing difficulties	• Weakness (postural changes, functional difficulties such as Gower's sign)
• Respiratory difficulties?	• Shortness of breath	• Muscle Atrophy or pseudohypertrophy
• Feeding difficulties?	• Dyspnea on exertion	• Cranial nerve involvement (limited tracking)
Past Medical History	• Nocturnal hypoventilation	• Progressive extremity deformities (high arched feet)
• Frequent respiratory illnesses,	***Bulbar symptoms***	• Delayed motor milestones (sitting, crawling, walking)
• Urinary or bowel changes such as constipation?	• Feeding difficulties	***After 2 years***
• Delayed motor milestones? Regression?	• Dysphagia,	• Weakness (main manifestation)
• Any cognitive or language delay?	• Failure to thrive	• Frequent falls/regression in motor milestones
Familial History	***Cardiac/Autonomic symptoms***	• Functional difficulties (Gower's sign)
• Other family members with similar concerns?	• Shortness of breath	• Progressive extremity deformities (acquired toe-walking, high arched feet)
• Consanguinity?	• Dizziness,	• Muscle atrophy/pseudohypertrophy
• Premature death?	• Syncope	
	• Chest pain complaints with exertion	
	• Tachycardia, hypo/hypertension, hypothermia	

Source[2, 26-31] Lee, 2018; McDonald, 2012; Darras BJJ et al, 2015; Mary P et al, 2018; Araújo et al. 2023.

In our clinical routine, pediatric and juvenile patients presenting with hypotonia, delayed motor development, or weakness should first undergo a comprehensive neurological examination to establish a topographic diagnosis. Following this, serum creatine phosphokinase (CPK) levels are measured. For hypotonic infants, we simultaneously test serum CPK, alpha-glucosidase activity, and a genetic panel (MLPA/Sequencing) for neuromuscular disorders (NMDs). If conditions such as SMA, Pompe disease, congenital muscular dystrophies, and congenital muscular myopathies are excluded, further investigations are conducted, including PCR testing for Myotonic Dystrophy, neurophysiological tests, or muscle

biopsy.[28] Regarding prognostic markers, late diagnosis, the need for ventilatory support (due to early-onset dyspnea), gastrostomy (for dysphagia), and loss of ambulation may predict disease progression and mortality in neuromuscular disorders (NMDs).[33,34]

Children with early-onset NMDs had significantly higher prevalence of dysphagia, gastroesophageal reflux, vomiting, underweight, as well as higher frequency of dietetic consultations, high energy diet, swallowing assessment and tube-feeding, compared to later onset NMDs.[35] West et al. created growth curves specifically for DMD, which demonstrated that male DMD patients were shorter than unaffected boys and tended to be overweight.[36] As the disease progresses, boys with DMD experience malnutrition due to an imbalance between increased energy needs (due respiratory failure) and negative energy intake (caused by dysphagia, delayed gastric emptying, constipation and prolonged mealtimes).[37] Specific growth charts for SMA are not currently available. It may be helpful to monitor growth trends rather than monitor weight in SMA type 1 patients. In contrast, children with SMA type III are prone to overeating and obesity from physical inactivity and due to lower basal metabolic rates. In general, guidelines recommend calcium and vitamin D intake for bone health.[38] Dysphagia can appear early during NMDs, leading to complications such as malnutrition, dehydration, aspiration pneumonia or difficulty in managing secretions. In SMA patients, maximal mouth opening (MMO) has long been considered a significant indicator of the impairment of the bulbar cranial nerve nuclei. A good MMO is associated with a better outcome for dysphagia and choking. The decrease in MMO is associated with fatty degeneration of the lateral pterygoid muscle, which negatively impacts the anterior sliding movement (condylar sliding) during mouth opening. For the treatment stage, adaptation strategies (diet, food, and posture) or feeding tubes were the most reported approaches.[39-43]

Children with Duchenne muscular dystrophy (DMD) exhibit cognitive impairments in approximately one-third of cases, especially in verbal comprehension, working memory, and processing speed, due to the absence of brain-expressed dystrophin isoforms like Dp427 and Dp71DMD. In spinal muscular atrophy (SMA), cognition is generally preserved in types 2 and 3, though subtle deficits in attention and executive function may emerge in more severely affected patients; in SMA type 1, despite expressive language impairment due to motor dysfunction, cognitive function especially non-verbal reasoning can be intact and assessed through alternative tools like eye tracking. In contrast, myotonic dystrophy type 1 (DM1) presents widespread cognitive dysfunction affecting memory, attention, executive function, and visuospatial skills, with severity correlating with disease form and CTG repeat length; congenital and childhood-onset DM1 are most severely affected. DM1 reveals diffuse white matter abnormalities, ventricular enlargement, and frontotemporal atrophy, reflecting a mixed neurodevelopmental and neurodegenerative mechanism. This neurocognitive spectrum across neuromuscular disorders calls for tailored cognitive assessment and support strategies.[44-50]

Children with neuromuscular disorders (NMDs) experience a progressive decline in respiratory function due to weakness in inspiratory, expiratory, and bulbar muscles. This leads to impaired airway clearance, sleep-disordered breathing, and ultimately diurnal ventilatory failure. Evaluation includes clinical assessment of breathing patterns (e.g., paradoxical thoracoabdominal movement), pulmonary function tests such as FVC, maximal inspiratory and expiratory pressures (MIP/MEP), and peak cough flow (PCF). A PCF <270 L/min or MEP <60 cmH_2O indicates ineffective cough in older children. Treatment involves non-invasive ventilation (NIV) to support gas exchange during sleep and later while awake, mechanical cough assistance (e.g., insufflation-exsufflation), and secretion mobilization techniques like high-frequency chest wall oscillation or intrapulmonary percussive ventilation. These interventions improve survival and reduce hospitalizations.[51-54]

Cardiac dysfunction is a major cause of morbidity and mortality in NMDs, particularly in Duchenne/Becker muscular dystrophy, myotonic dystrophy (DM), limb-girdle muscular dystrophies (LGMD), and Emery-Dreifuss muscular dystrophy (EDMD). These conditions frequently lead to progressive dilated cardiomyopathy and arrhythmias due to the replacement of myocytes with fibrofatty tissue, conduction system degeneration, and myocardial fibrosis. Early cardiac involvement can be asymptomatic, hence routine monitoring with ECG, echocardiography, and cardiac MRI is essential from early ages depending on the condition. Treatment includes standard heart failure therapies (ACE inhibitors, beta-blockers, mineralocorticoid receptor antagonists), with increasing use of implantable cardioverter-defibrillators and pacemakers in arrhythmia-prone patients.[55-59]

Rehabilitation in neuromuscular disorders should be individualized and adapted to disease stage and type. For Duchenne muscular dystrophy (DMD), motor assessments are recommended every 4 to 6 months, and physical therapy should include joint mobility exercises, stretching, and light to moderate isometric resistance training. Ambulatory patients may benefit from night-time ankle-foot orthoses and daily mobility activities, while eccentric exercises should be avoided. In spinal muscular atrophy (SMA), rehabilitation involves routine stretching (minimum 3–5 times/week), use of orthoses, supported standing, and power mobility aids, tailored according to functional classification (non-sitters, sitters, walkers) and regularly reviewed every 6 months. A systematic review supports aerobic exercises of light to moderate intensity, 3 times/week for 30 minutes, for improving mobility and participation in slowly progressive neuromuscular disorders such as myotonic dystrophy and Charcot-Marie-Tooth disease. For most NMDs, therapy should prioritize activities

of daily living and social participation, with interdisciplinary support and goal-focused planning. [4-7, 60-61]

Several standardized motor functional scales are employed in the assessment of NMDs, each with distinct purposes aligned to patient age, disease phenotype, and ambulatory status. In DMD, key scales include the North Star Ambulatory Assessment (NSAA), which quantifies ambulatory function; timed function tests (e.g., 10-meter walk/run, time to rise), predictive of gait loss; and the Performance of Upper Limb (PUL) scale, useful for non-ambulant individuals to assess upper limb progression. For SMA, common tools include CHOP-INTEND for type 1 infants, HFMSE for sitters and ambulators, and the Revised Upper Limb Module (RULM) for both ambulant and non-ambulant patients. The Motor Function Measure (MFM) is applicable across DMD and SMA, capturing both ambulatory and non-ambulatory disease progression. These scales guide clinical monitoring, predict functional decline, and evaluate treatment efficacy, especially in the context of evolving disease-modifying therapies.[61-65]

CONCLUSION

The Neuromuscular Assessment Form (NAF) developed and routinely applied at IPPMG/UFRJ is perceived by our team to be a valuable tool for standardizing clinical evaluations, tracking disease progression, and guiding interdisciplinary interventions. By integrating multisystemic data, NAF supports personalized and anticipatory care plans that align with current international standards. In resource-limited settings, such a framework is critical to ensure continuity of care and optimize outcomes for patients with rare and complex disorders. In helps as a check list of items to be explored in clinical follow-up. Furthermore, the NAF enables the systematic collection of clinical data on the natural history of neuromuscular diseases in childhood, providing a robust and essential framework for the comparative evaluation of emerging disease-modifying therapies in clinical research. This structure thereby supports and enhances the center's capacity to generate high-quality scientific output.

The main challenges associated with the NAF involve the continuous training and supervision of junior residents to ensure accurate performance of timed tests and physical examination, as well as the precise documentation of clinical information to minimize data gaps. Additionally, over the past ten years, the NAF has required periodic updates with new variables, in line with advancements in scientific knowledge.

Future efforts should focus on validating the NAF across different populations and exploring its integration into electronic health records to enhance clinical decision-making and research capacity. In our huge country, disparities in access to specialized care for neuromuscular disorders persist, with only a few referral centers located in the North, South, and Southeast regions. The NAF has the potential to serve as a standardized clinical tool for systematic evaluation and uniform data collection across regions, promoting equity in care and research. Our initial dataset has helped our team to mentor post-graduates and to be involved in multicenter clinical trials since 2011.

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