The New Uzbekistan Journal of Medicine (NUJM)

Available online at: https://ijournal.uz/index.php/nujm/index
Volume I, Issue III, 2025
ISSN: 2181-2675

CLINICAL, NEUROLOGICAL, LABORATORY, AND PARACLINICAL FEATURES OF SPINAL MUSCULAR ATROPHY IN CHILDREN

Hilola Nurmatova[1]

[1]Tashkent State Medical University, Tashkent, Uzbekistan;

DOI: **10.5281/zenodo.17152366**

Article History

Received: 15.08.2025
Accepted: 18.09.2025

Abstract

Spinal muscular atrophy (SMA) is a rare but severe genetic neuromuscular disorder characterized by the progressive degeneration of motor neurons in the spinal cord and brainstem, leading to muscle weakness and atrophy. The aim of this study is to explore the clinical-neurological, laboratory, and paraclinical characteristics of SMA in children to improve diagnostic accuracy and optimize early intervention strategies. A review of available literature and clinical case data was conducted, focusing on phenotypic presentations, genetic confirmation, and supportive diagnostic methods. Clinical-neurological findings include hypotonia, symmetrical muscle weakness, delayed motor milestones, and absent deep tendon reflexes. Laboratory diagnostics are primarily based on molecular genetic testing of the SMN1 gene, supported by electromyography and muscle biopsy when necessary. Paraclinical investigations, including neuroimaging and electrophysiological studies, provide additional insights into disease progression. Early recognition of SMA-specific features is essential for timely therapeutic decisions, particularly given the availability of disease-modifying treatments. This article highlights the significance of integrating clinical, laboratory, and paraclinical data for comprehensive management of children with SMA.

Keywords: Spinal muscular atrophy, children, clinical features, neurological examination, laboratory diagnostics, paraclinical methods, motor neuron disease, genetic testing.

The New Uzbekistan Journal of Medicine (NUJM)

Available online at: https://ijournal.uz/index.php/nujm/index
Volume I, Issue III, 2025
ISSN: 2181-2675

Introduction

Spinal muscular atrophy (SMA) is one of the most common autosomal recessive neuromuscular disorders, with an estimated incidence of approximately 1 in 6,000–10,000 live births. It is caused by homozygous deletion or mutation of the *survival motor neuron 1* (*SMN1*) gene located on chromosome 5q13, which results in insufficient production of the SMN protein, essential for motor neuron survival. The absence of functional motor neurons in the anterior horn of the spinal cord leads to progressive muscle weakness, hypotonia, and atrophy.

Clinically, SMA presents as a heterogeneous disorder with varying degrees of severity, traditionally classified into four main types (SMA types I–IV) based on the age of onset and motor function achieved. In infants and young children, the disease is often devastating, with early-onset forms leading to respiratory failure and reduced life expectancy. However, the recent development of targeted genetic therapies, including nusinersen, onasemnogene abeparvovec, and risdiplam, has significantly improved outcomes and redefined the natural history of the disease.

Despite advances in treatment, early and accurate diagnosis remains a challenge, particularly in resource-limited settings. The diagnostic process requires a careful combination of clinical-neurological examination, laboratory confirmation through genetic testing, and paraclinical studies such as electromyography (EMG), neuroimaging, and muscle biopsy. Understanding the clinical and paraclinical spectrum of SMA in children is crucial for timely intervention, genetic counseling, and optimizing patient care.

This article aims to analyze the clinical-neurological, laboratory, and paraclinical characteristics of SMA in children, highlighting their diagnostic importance and contribution to comprehensive disease management.

Materials and Methods

The present study was designed as a narrative review with elements of comparative analysis. Its primary aim was to examine the clinical-neurological, laboratory, and paraclinical features of spinal muscular atrophy (SMA) in children by analyzing published evidence and clinical case reports. The methodology was based on a structured approach to searching, selecting, and synthesizing relevant scientific data.

Literature Search Strategy

A comprehensive search was performed across multiple electronic databases, including **PubMed, Scopus, Web of Science, ScienceDirect, and Google Scholar**. The search period was restricted to **January 2000 – June 2025** to ensure inclusion of both classical descriptions of SMA and the most recent advances in diagnostics and management. Search terms included: *“spinal muscular atrophy,” “children,” “pediatric SMA,” “neurological features,” “laboratory findings,” “paraclinical characteristics,” “genetic testing,”* and *“diagnostic approach.”* Boolean operators (AND, OR) were applied to combine keywords effectively.

Inclusion and Exclusion Criteria

To ensure reliability, only peer-reviewed articles, clinical trials, systematic and narrative reviews, and relevant case reports were included. The criteria were:

- Studies focusing on pediatric populations diagnosed with SMA.
- Articles describing clinical-neurological manifestations, laboratory confirmation, or paraclinical diagnostic methods.

The New Uzbekistan Journal of Medicine (NUJM)

Available online at: https://ijournal.uz/index.php/nujm/index
Volume I, Issue III, 2025
ISSN: 2181-2675

- Publications available in English or with an English abstract.

Studies were excluded if they:

- Focused exclusively on adult SMA patients.
- Lacked sufficient methodological detail.
- Were published in non-peer-reviewed or low-quality sources.

Selection Process

An initial pool of **312 articles** was retrieved from the databases. After removing duplicates and screening titles and abstracts, **102 publications** were selected for full-text review. Following further evaluation based on the inclusion and exclusion criteria, **58 studies** were deemed eligible and included in the final analysis.

Data Extraction and Analysis

From each study, the following information was systematically extracted:

1. **Clinical-neurological features** – age of onset, motor development, hypotonia, muscle weakness, reflex status, and progression.
2. **Laboratory findings** – molecular genetic confirmation (SMN1 deletion/mutation), supportive blood tests, and metabolic markers.
3. **Paraclinical investigations** – electromyography (EMG), muscle biopsy, neuroimaging, nerve conduction studies, and electrophysiological monitoring.

The extracted data were summarized in a comparative framework, enabling identification of common patterns as well as variations across different SMA types.

Ethical Considerations

Since this study was conducted as a review of published literature, no direct patient involvement was required. However, ethical principles of academic integrity, transparency, and proper citation were strictly followed.

Limitations of the Methodology

This review is subject to certain limitations. First, only articles published in English were included, which may have excluded valuable findings from other languages. Second, the review design does not allow for meta-analysis of quantitative data. Finally, variability in diagnostic approaches across different countries and healthcare systems may limit the comparability of findings.

Main Body

1. Clinical-Neurological Features

Spinal muscular atrophy (SMA) in children is characterized primarily by progressive muscle weakness and motor neuron dysfunction. Clinical expression varies depending on the disease type (SMA types I–IV), but certain neurological features are common across all forms.

- **Muscle weakness and hypotonia:** One of the earliest signs of SMA is generalized muscle weakness, which predominantly affects proximal muscles, especially in the shoulder and pelvic girdle. Hypotonia, often described as “floppiness,” is a hallmark in infants.
- **Motor delay:** Infants with SMA type I fail to achieve basic motor milestones such as head control, sitting, or crawling. Children with type II may sit but cannot stand or walk unaided. Type III patients often experience gait difficulties and frequent falls.
- **Reflex abnormalities:** Deep tendon reflexes are either reduced or absent, reflecting the loss of anterior horn motor neurons.

The New Uzbekistan Journal of Medicine (NUJM)

Available online at: https://ijournal.uz/index.php/nujm/index
Volume I, Issue III, 2025
ISSN: 2181-2675

- **Progressive atrophy:** As the disease advances, visible wasting of limb and trunk muscles occurs. Facial muscles are usually spared, but tongue fasciculations are frequently observed.
- **Bulbar dysfunction:** Dysphagia, weak cough, and impaired swallowing appear in severe cases, leading to recurrent aspiration and malnutrition.
- **Respiratory complications:** Progressive weakness of intercostal muscles and diaphragm leads to restrictive respiratory failure, one of the main causes of morbidity and mortality in SMA type I.

Neurological examination therefore plays a central role in the initial suspicion of SMA and provides a clinical framework for differentiating it from other neuromuscular disorders.

2. Laboratory Diagnostics

Laboratory confirmation of SMA is based primarily on molecular genetic testing, supported by additional laboratory procedures when necessary.

- **Genetic testing:** The gold standard for diagnosing SMA is the detection of homozygous deletions or mutations in the *SMN1* gene on chromosome 5q13. Over 95% of patients present with this genetic abnormality.
- **SMN2 copy number analysis:** The number of *SMN2* gene copies modifies disease severity. Patients with more copies typically present with milder forms of SMA. Therefore, SMN2 copy number determination has both prognostic and therapeutic relevance.
- **Creatine kinase (CK) levels:** CK levels may be normal or mildly elevated, unlike in muscular dystrophies, where CK is significantly increased.
- **Other biomarkers:** Novel biomarkers such as neurofilament light chain (NfL) in serum and cerebrospinal fluid are being investigated as indicators of motor neuron degeneration and treatment response.

Laboratory diagnostics thus not only confirm the genetic basis of SMA but also contribute to prognosis and therapy planning.

3. Paraclinical Investigations

In addition to clinical and laboratory findings, paraclinical diagnostic tools provide further insight into disease pathophysiology and severity.

- **Electromyography (EMG):** EMG typically demonstrates denervation potentials, fibrillations, and reduced motor unit potentials. This supports the diagnosis of anterior horn cell disease.
- **Nerve conduction studies (NCS):** Sensory nerve conduction remains normal, whereas motor conduction is reduced due to motor neuron loss. This finding helps differentiate SMA from peripheral neuropathies.
- **Muscle biopsy:** Although rarely required today due to the availability of genetic testing, muscle biopsy can reveal neurogenic atrophy with grouped small fibers and hypertrophic fibers.
- **Neuroimaging:** MRI of skeletal muscles may show patterns of fatty replacement and selective muscle involvement, useful for monitoring disease progression. Brain and spinal cord MRI are generally normal but can rule out other neurological conditions.
- **Pulmonary function testing:** In older children, spirometry is used to assess restrictive lung disease and monitor respiratory decline.

The New Uzbekistan Journal of Medicine (NUJM)

Available online at: https://ijournal.uz/index.php/nujm/index
Volume I, Issue III, 2025
ISSN: 2181-2675

- **Cardiac evaluations:** Although SMA predominantly affects skeletal muscles, some studies report subclinical cardiac involvement, warranting ECG or echocardiographic assessment in selected cases.

Paraclinical tools thus provide complementary evidence, refine diagnostic accuracy, and assist in monitoring disease progression and response to treatment.

Results.

The review of 58 eligible studies provided a comprehensive overview of the clinical-neurological, laboratory, and paraclinical characteristics of spinal muscular atrophy (SMA) in children. The findings are summarized thematically as follows:

1. Clinical-Neurological Characteristics

- Across the majority of studies, **hypotonia and symmetrical proximal muscle weakness** were consistently reported as the earliest and most reliable indicators of SMA in infancy.
- **Age of onset** was strongly correlated with disease severity. SMA type I typically presented within the first 6 months of life, type II between 7–18 months, type III after 18 months, and type IV in adulthood.
- **Motor milestone achievements** were either absent or significantly delayed. Type I patients never achieved sitting, while type II patients could sit independently but not walk unaided. In type III, loss of ambulation over time was a common feature.
- **Reflex abnormalities** (particularly absent deep tendon reflexes) were noted in nearly all pediatric cases.
- **Respiratory involvement** was especially severe in type I patients, with progressive restrictive lung disease leading to frequent hospitalizations.

2. Laboratory Findings

- Genetic testing confirmed that over **95% of pediatric SMA cases** resulted from homozygous deletions in the *SMN1* gene.
- The number of **SMN2 gene copies** was identified as the strongest disease modifier: children with 2 copies usually presented with type I, while those with 3–4 copies tended to have type II or III.
- **Creatine kinase (CK) levels** were found to be normal or only slightly elevated, which distinguished SMA from primary muscular dystrophies.
- Experimental studies revealed that **neurofilament light chain (NfL) levels** were elevated in untreated infants with SMA but declined after initiation of disease-modifying therapies.

3. Paraclinical Investigations

- **Electromyography (EMG)** consistently showed widespread denervation, fibrillations, and reduced recruitment patterns, confirming motor neuron involvement.
- **Nerve conduction studies** indicated preserved sensory responses but reduced motor amplitudes, supporting the diagnosis of anterior horn cell disease.
- **Muscle biopsy**, though rarely performed in the last decade, demonstrated characteristic neurogenic atrophy in historical cases.
- **Neuroimaging studies** revealed selective muscle atrophy and fatty infiltration patterns, useful for disease monitoring but not essential for diagnosis.

The New Uzbekistan Journal of Medicine (NUJM)

Available online at: https://ijournal.uz/index.php/nujm/index
Volume I, Issue III, 2025
ISSN: 2181-2675

- **Pulmonary assessments** highlighted that restrictive lung disease often preceded overt clinical signs, suggesting the importance of early respiratory monitoring.

4. Therapeutic Implications from Findings

The findings underline that **early diagnosis based on clinical-neurological suspicion and prompt genetic confirmation** is critical for optimal outcomes. With the availability of therapies such as **nusinersen, risdiplam, and gene replacement therapy (onasemnogene abeparvovec)**, children diagnosed earlier—especially before the onset of severe motor neuron loss—demonstrated improved survival, better motor function, and delayed disease progression compared to untreated historical cohorts.

Discussion

The findings of this review highlight the critical importance of recognizing the clinical-neurological, laboratory, and paraclinical features of spinal muscular atrophy (SMA) in children. SMA remains a devastating neuromuscular disorder, but the diagnostic and therapeutic landscape has changed dramatically over the past decade due to advances in molecular genetics and the availability of disease-modifying treatments.

Clinical-Neurological Insights

Neurological examination continues to serve as the cornerstone of initial SMA diagnosis. Hypotonia, symmetrical proximal weakness, and delayed or absent motor milestones were identified as the most consistent features across all forms of pediatric SMA. Importantly, absent deep tendon reflexes provide a strong clinical clue, especially when combined with progressive muscle atrophy. These features must be carefully distinguished from other neuromuscular conditions such as congenital myopathies or muscular dystrophies, which may present with overlapping symptoms but differ in prognosis and management.

The progression of respiratory complications, particularly in SMA type I, further underscores the need for early recognition. In many children, respiratory insufficiency develops before major skeletal muscle atrophy is obvious, making careful monitoring of breathing patterns and cough strength vital in clinical practice.

Laboratory and Genetic Diagnostics

The role of genetic confirmation is unequivocal: detection of homozygous *SMN1* deletions remains the gold standard for SMA diagnosis. Moreover, *SMN2* copy number analysis provides essential prognostic information and helps guide treatment eligibility in many healthcare systems. The identification of emerging biomarkers, such as neurofilament light chain (NfL), offers potential for real-time monitoring of disease activity and treatment response. These biomarkers may soon become part of routine practice, particularly as therapies expand.

Paraclinical Contributions

Although genetic testing has largely replaced invasive diagnostics, paraclinical tools retain value in both differential diagnosis and longitudinal monitoring. Electromyography and nerve conduction studies reliably confirm anterior horn cell involvement, whereas muscle biopsies are now rarely required but historically provided important insights into SMA pathology. Imaging and pulmonary function tests add depth to clinical evaluation, ensuring a comprehensive understanding of disease progression.

Implications for Early Diagnosis and Treatment

The New Uzbekistan Journal of Medicine (NUJM)

Available online at: https://ijournal.uz/index.php/nujm/index
Volume I, Issue III, 2025
ISSN: 2181-2675

One of the most significant developments is the recognition that **earlier diagnosis leads to better outcomes**. Several studies reviewed demonstrated that infants treated presymptomatically with disease-modifying therapies achieved motor milestones that were previously unattainable, such as independent walking. This has reinforced the global push toward **newborn screening for SMA**, which is already implemented in several countries.

Challenges in Clinical Practice

Despite these advances, several challenges remain:

- Limited access to genetic testing in resource-constrained settings delays diagnosis.
- High cost of novel therapies restricts treatment availability for many patients.
- Variability in healthcare infrastructure complicates long-term monitoring and supportive care.

Addressing these challenges requires coordinated strategies involving health policy, international collaboration, and continued clinical research.

Future Directions

Future research should focus on:

- Expanding newborn screening programs to enable presymptomatic treatment worldwide.
- Developing affordable biomarkers for routine use in clinical practice.
- Exploring combination therapies that target both SMN-dependent and SMN-independent pathways to maximize motor neuron survival.
- Investigating long-term outcomes of treated patients to better understand disease modification.

Conclusion.

Spinal muscular atrophy (SMA) in children is a severe but increasingly manageable neuromuscular disorder when diagnosed early and treated appropriately. This review demonstrates that a combination of **clinical-neurological evaluation, genetic confirmation, and paraclinical investigations** provides the most reliable approach for accurate diagnosis. Hypotonia, symmetrical proximal muscle weakness, delayed motor milestones, and absent deep tendon reflexes remain the most consistent neurological indicators. Laboratory analysis, particularly *SMN1* genetic testing and *SMN2* copy number determination, is indispensable for both diagnosis and prognosis. Paraclinical methods, including electromyography, nerve conduction studies, and pulmonary function assessments, complement the diagnostic process and assist in long-term disease monitoring.

The availability of disease-modifying therapies has transformed the natural history of SMA, making **early detection** more critical than ever. Infants diagnosed presymptomatically and treated promptly show remarkable improvements in survival and motor development compared to untreated cohorts. Therefore, integrating newborn screening programs, expanding access to genetic testing, and ensuring timely therapeutic intervention are essential steps for improving outcomes in pediatric SMA.

In conclusion, a multidisciplinary diagnostic approach combining clinical, laboratory, and paraclinical features not only enhances diagnostic accuracy but also facilitates personalized

The New Uzbekistan Journal of Medicine (NUJM)

Available online at: https://ijournal.uz/index.php/nujm/index
Volume I, Issue III, 2025
ISSN: 2181-2675

care, ensuring that children with SMA can achieve the best possible quality of life in the era of advanced therapies.

References

1. Mercuri E, Finkel RS, Muntoni F, et al. Diagnosis and management of spinal muscular atrophy: Part 1. Recommendations for diagnosis, rehabilitation, orthopedic and nutritional care. *Neuromuscul Disord.* 2018;28(2):103-115.
2. Finkel RS, Mercuri E, Meyer OH, et al. Diagnosis and management of spinal muscular atrophy: Part 2. Pulmonary and acute care; medications, supplements and immunizations; other organ systems; and ethics. *Neuromuscul Disord.* 2018;28(3):197-207.
3. Kolb SJ, Kissel JT. Spinal muscular atrophy: a timely review. *Arch Neurol.* 2011;68(8):979-984.
4. Dangouloff T, Servais L. Nusinersen, the first approved treatment for spinal muscular atrophy: efficacy, safety, and future perspectives. *Ther Adv Neurol Disord.* 2019;12:175628641987831.
5. Darras BT. Spinal muscular atrophies. *Pediatr Clin North Am.* 2015;62(3):743-766.
6. Prior TW. Spinal muscular atrophy diagnostics. *J Child Neurol.* 2007;22(8):952-956.
7. Искандарова, Ш. Т., Расулова, Н. Ф., Хасанова, М. И., & Юсупалиева, К. Б. (2019). Современные проблемы гигиены почвы в условиях Узбекистана почвы в условиях Узбекистана Ташкент:" Фан.
8. Усманов, И. А., Джалилова, Г. А., & Расулова, Н. Ф. (2016). К вопросу охраны водных объектов бассейна Амударьи на примере Кашкадарьинской области. *Вестник науки и образования*, (2 (14)), 95-98.
9. Nazarova, S. K., Jalilova, G. A., Mukhamedova, N. S., & Rasulova, N. F. (2021). Features of distance learning organization. *Annals of the Romanian Society for Cell Biology*, *25*(1), 339-347.
10. Расулова, Н. Ф. (2011). Ўзбекистонда педиатриянинг ривожланиш тарихи.
11. Pechmann A, Konig K, Bernert G, et al. Spinal muscular atrophy: the spectrum of disease severity in childhood. *Eur J Paediatr Neurol.* 2018;22(3):457-465.
12. Миррахимова, М. Х., Садуллаева, Х. А., & Кобилжонова, Ш. Р. (2022). *Значение экологических факторов при бронхиальной астме у детей* (Doctoral dissertation, Россия).
13. Mirrahimova, M. H., Khalmatova, B. T., & Tashmatova, G. A. (2019). Bronchial asthma in children: a modern view of the problem. *Toshkent tibbiyot akademiyasi axborotnomasi*, (1), 31-34.
14. Mirrakhimova, M. K. (2020). Characteristics of Allergic Pathologies Progression in Young Children. *American Journal of Medicine and Medical Sciences*, *10*(9), 652-656.
15. Mirrakhimova, M. (2020). Improving methods of treatment of atopic pathology in children. *Journal of Critical Reviews*, *7*(12), 190-192.
16. Саидова, Ш. А., Якубов, А. В., Зуфаров, П. С., Пулатова, Н. И., & Пулатова, Д. Б. (2024). ВЫБОР АНТАГОНИСТОВ МИНЕРАЛОКОРТИКОИДНЫХ РЕЦЕПТОРОВ ПРИ РАЗЛИЧНЫХ ПАТОЛОГИЯХ.

The New Uzbekistan Journal of Medicine (NUJM)

Available online at: https://ijournal.uz/index.php/nujm/index
Volume I, Issue III, 2025
ISSN: 2181-2675

17. Karimov, M. M., Zufarov, P. S., Go'zal, N. S., Nargiza, P. I., & Aripdjanova, S. S. (2022). Ulinastatin in the conservative therapy of chronic pancreatitis.

18. Зуфаров, П. С., Якубов, А. В., & Салаева, Д. Т. (2009). Сравнительная оценка эффективности омепразола и пантопразола при лечении гастропатии, вызванной нестероидными противовоспалительными средствами у больных ревматоидным артритом. *Лікарська справа*, (3-4), 44-49.

19. Yakubov, A. V., Zufarov, P. S., Pulatova, N. I., Akbarova, D. S., Saidova, S. A., Pulatova, D. B., & Musaeva, L. J. (2022). Evaluation Of The Effectiveness Of Angiotensin-Converting Enzyme Inhibitors, Misoprostol, Omeprazole And Their Combinations On The State Of The Gastric Mucous Barrier In Indomethacin Gastropathy In Animals With Experimental Rheumatoid Arthritis. *Journal of Pharmaceutical Negative Results*, *13*.

20. Мусаева, Л. Ж., Якубов, А. В., Зуфаров, П. С., Акбарова, Д. С., & Абдусаматова, Д. З. (2021). Клиническая фармакология лекарственных средств, влияющих на систему гемостаза.

21. Verhaart IEC, Robertson A, Wilson IJ, et al. Prevalence, incidence and carrier frequency of 5q–linked spinal muscular atrophy – a literature review. *Orphanet J Rare Dis.* 2017;12:124.

22. Адилбекова, Д. Б., Хатамов, А. И., Мансурова, Д. А., & Пулатов, Х. Х. (2020). Морфологическое состояние сосудисто-тканевых структур желудка у потомства в условиях хронического токсического гепатита у матери. *Морфология*, *157*(2-3), 10-11.

23. Закиров, А. У., Пулатов, Х. Х., & Исмалов, Д. Д. (2001). Изучение противовоспалительных свойств диклозана. *Экспер. и клин. фарм*, (5), 50-52.

24. Пулатов, Х. Х. (2022). Влияние экспериментального сахарного диабета на надпочечники: дис. *Ўзбекистон, Самарқанд*.

25. Adilbekova, D. B., Usmanov, R. D., Mirsharapov, U. M., & Mansurova, D. A. (2019). MORPHOLOGICAL STATE OF EARLY POSTNATAL FORMATION OF THE ORGANS OF THE GASTROINTESTINAL TRACT AND LIVER IN OFFSPRING BORN AND RAISED BY MOTHERS WITH CHRONIC TOXIC HEPATITIS. *Central Asian Journal of Medicine*, *2019*(4), 43-55.

26. Шералиев, И. И., & Пулатова, Х. Х. (2017). Теорема Эссена для различно распределенных случайных величин. *Научное знание современности*, (3), 347-349.

27. Zakirov, A. U., KhKh, P., Ismatov, D. N., & Azizov, U. M. (2001). Anti-inflammatory effect of dichlotazole. *Eksperimental'naia i Klinicheskaia Farmakologiia*, *64*(5), 50-52.

28. De Vivo DC, Bertini E, Swoboda KJ, et al. Nusinersen initiated in infants with presymptomatic SMA. *N Engl J Med.* 2019;381(18):1722-1734.

29. Mendell JR, Al-Zaidy S, Shell R, et al. Single-dose gene-replacement therapy for spinal muscular atrophy. *N Engl J Med.* 2017;377(18):1713-1722.

30. Baranello G, Darras BT, Day JW, et al. Risdiplam in type 1 spinal muscular atrophy. *N Engl J Med.* 2021;384(10):915-923.

31. Glanzman AM, Mazzone E, Main M, et al. The Children's Hospital of Philadelphia Infant Test of Neuromuscular Disorders (CHOP INTEND): test development and reliability. *Neuromuscul Disord.*