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Intrathecal Nusinersen Therapy in a Patient With Spinal Muscular Atrophy Type 1 in Bahrain: A Case Report

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Abstract

Spinal muscular atrophy (SMA) Type 1 is a severe infantile-onset neuromuscular disorder in which independent sitting is generally not achieved without disease-modifying treatment. We describe the longitudinal course of a girl with molecularly confirmed 5q SMA and a clinical SMA Type 1 phenotype treated at the Royal Medical Services Military Hospital in Bahrain. The diagnosis was documented at one month and six days of age, and intrathecal nusinersen was initiated at 10 months and 28 days. She received ten 12-mg administrations between June 23, 2024, and November 9, 2026, alongside physiotherapy and occupational therapy. Pretreatment assessment documented fair head control, inability to roll or sit independently, and reduced limb function and weight-bearing. During approximately 28 months of treatment and rehabilitation, follow-up assessments documented improvements in head and trunk control, rolling, reaching, grasping, and weight-bearing, as well as the acquisition of independent sitting. Clinical photographs at three years and three months of age illustrated upright standing with external contact. A post-treatment Children's Hospital of Philadelphia Infant Test of Neuromuscular Disorders (CHOP-INTEND) score of 51/64 was recorded. This case documents clinically relevant motor milestones and prolonged multidisciplinary follow-up during nusinersen treatment for SMA Type 1.

Categories: Neurology, Physical Medicine & Rehabilitation, Pediatrics
Keywords: case report, disease-modifying therapy, early diagnosis, motor function, nusinersen, occupational therapy, pediatric neuromuscular disease, physiotherapy, smn1 gene, spinal muscular atrophy type 1

Introduction

Approximately one in 10,000 live births worldwide will be affected by spinal muscular atrophy (SMA), making it one of the most common inherited and lethal neuromuscular disorders of infancy [1]. The disease is categorized into five clinical subtypes (Types 0-4) based on the age at symptom onset and the highest motor milestone achieved [2]. SMA Type 1 is the predominant form, representing nearly 50-60% of all cases, and is one of the most severe forms, with high mortality [1,2]. Symptoms typically emerge during the first six months of life, when affected infants develop marked hypotonia, progressive proximal muscle weakness, poor head control, feeding difficulties, and respiratory impairment [2]. In the untreated natural history, children with SMA Type 1 do not achieve independent sitting [2]. The high mortality associated with SMA Type 1 is attributed to its early involvement of respiratory and bulbar muscles, which leads to recurrent respiratory complications, ineffective airway clearance, and swallowing dysfunction [2]. Most affected infants either die or require permanent ventilatory support before reaching two years of age if no disease-modifying therapy is given [3-5].

Historically, management of SMA focused on supportive respiratory care, physiotherapy, and nutritional management, aiming to reduce complications and improve quality of life [2]. The introduction of nusinersen (Spinraza), the first US Food and Drug Administration-approved disease-modifying therapy for SMA, represented an important advancement in treatment [6]. Nusinersen is an antisense oligonucleotide administered intrathecally that modifies splicing of the SMN2 gene, increasing production of functional survival motor neuron protein [6]. The ENDEAR randomized controlled trial demonstrated improvements in motor milestone responses and survival-related outcomes among infants with symptomatic SMA treated with nusinersen [7]. More recent real-world evidence, including a 2025 multicenter Turkish cohort, has documented longitudinal motor, respiratory, and bulbar outcomes in children with SMA Type 1 receiving nusinersen [8]. However, detailed documentation of treatment exposure, rehabilitation, and functional outcomes remains important for interpreting individual clinical courses. The present report describes the management of a child with SMA Type 1 in Bahrain, focusing on the diagnostic timeline, nusinersen administration, multidisciplinary rehabilitation, and observed motor outcomes.

Case Presentation

We describe the clinical course of a three-year-old girl with genetically confirmed 5q SMA, clinically

diagnosed as SMA Type 1, who received intrathecal nusinersen (Spinraza) alongside multidisciplinary rehabilitation at the Royal Medical Services Military Hospital in Bahrain. The diagnosis was documented at one month and six days of age, and nusinersen therapy began on June 23, 2024, at 10 months and 28 days of age. Ten treatment administrations were documented through November 9, 2026, when the patient was approximately three years and three months old.

She is the daughter of consanguineous Jordanian parents. An older sibling died at six months of age following muscle weakness, reduced activity, and respiratory difficulties; the cause of death was not established, and no genetic testing was performed. Neither parent had undergone genetic testing, and no other relatives were reported to have a similar condition. Her mother has diet-controlled type 2 diabetes mellitus.

The patient was delivered by elective lower-segment cesarean section at 38+3 weeks' gestation due to previous cesarean deliveries. Birth weight was 3.225 kg, with Apgar scores of 9 and 9 at one and five minutes, respectively. A detailed fetal anomaly scan performed at 36+5 weeks' gestation was normal. Immediately after birth, the infant was active and alert, with good muscle tone, normal spontaneous symmetrical movements of all four limbs, no facial asymmetry, a good and vigorous sucking reflex, and intact primitive neonatal reflexes. The anterior fontanelle was level, and there were no signs of hypotonia, muscle weakness, tongue fasciculations, or respiratory distress. Cardiovascular examination revealed normal heart sounds with no murmurs, while abdominal examination was soft with no palpable masses or organomegaly. She was admitted to the neonatal intensive care unit for observation because of the significant family history of a sibling who had died from suspected SMA, with particular monitoring for the development of hypotonia, muscle weakness, or poor sucking reflex. However, her hospital stay was unremarkable, and she remained clinically stable throughout before being discharged home.

Because of the significant family history of a sibling who died following an illness suspected to represent SMA, the patient underwent early genetic evaluation. A clinical diagnosis of SMA was documented at one month and six days of age. At approximately two months of age, neurological assessment documented reduced spontaneous movements, peripheral hypotonia, tongue fasciculations, areflexia, and generalized muscle atrophy, consistent with the clinical phenotype of SMA Type 1. The differential diagnosis included congenital myopathies, congenital muscular dystrophies, and other hereditary neuromuscular disorders. Molecular analysis was performed using multiplex ligation-dependent probe amplification (MLPA; P021-B1 assay, MRC Holland). The available laboratory report, issued on September 28, 2023, demonstrated a homozygous deletion of exons 7 and 8 of the SMN1 gene, a homozygous deletion of exon 5 of the neuronal apoptosis inhibitory protein gene, and two copies of the SMN2 gene. The molecular findings confirmed 5q SMA, while the SMA Type 1 classification was based on the clinical presentation, age of symptom onset, and pretreatment motor phenotype rather than SMN2 copy number alone [9].

Before initiation of intrathecal nusinersen therapy, physiotherapy assessment demonstrated substantial motor impairment consistent with the clinical diagnosis of SMA Type 1. The patient had fair head control, was unable to roll or sit independently, and demonstrated reduced upper- and lower-limb strength with impaired trunk and hip control. Rolling required prompting or assistance, and lower-limb weight-bearing ability was limited. Functional use of the upper limbs was reduced, and feeding activities required assistance. Full passive range of motion was preserved in all extremities. At the first nusinersen admission on June 23, 2024, the patient was alert and vitally stable, with hypotonia on examination and clear breath sounds bilaterally. She was feeding well on formula and solid foods. Her weight was 8.9 kg, length was 74 cm, and head circumference was 44 cm. Outpatient complete blood count, biochemical investigations, and coagulation tests were reported as normal. These findings supplement the descriptive pretreatment motor baseline.

The patient received regular physiotherapy and occupational therapy alongside intrathecal nusinersen. Rehabilitation focused on head and trunk control, rolling, upper- and lower-limb function, mobility, weight-bearing ability, and preservation of joint range of motion to minimize musculoskeletal complications. Serial physiotherapy assessments documented changes in motor performance and functional abilities. Caregiver education and home exercise instructions supported ongoing rehabilitation between visits.

The patient commenced intrathecal nusinersen (Spinraza) therapy on June 23, 2024, at 10 months and 28 days of age, receiving 12 mg (5 mL) per administration according to the prescribed low-dose regimen [6]. Ten intrathecal administrations were verified between June 23, 2024, and November 9, 2026, comprising four loading doses and six maintenance doses. The loading doses were administered on June 23, July 8, July 28, and August 22, 2024, at intervals of 15, 20, and 25 days, respectively. Maintenance doses were administered on December 22, 2024; April 22, 2025; August 24, 2025; December 29, 2025; May 4, 2026; and November 9, 2026. The maintenance intervals were 122, 121, 124, 127, 126, and 189 days, with the final interval longer than the prescribed four-month schedule. The overall documented treatment period extended for approximately 28 months. Intrathecal administration was performed under procedural sedation, with ketamine use documented during the first administration. The first procedure was recorded as uneventful, and the discharge summaries for all 10 treatment admissions documented discharge home in stable condition. At the second administration on July 8, 2024, the patient was documented as maintaining oxygen

saturation on room air. No immediate procedural complications were reported in the reviewed discharge summaries.

Following initiation of intrathecal nusinersen and continuation of multidisciplinary rehabilitation, serial physiotherapy assessments described improvements in motor and functional abilities. Head control improved from fair to good, with full cervical range of motion and bilateral head turning. Rolling ability improved, although occasional prompting remained necessary. The patient subsequently achieved independent sitting without support, a motor milestone that had not been attained before treatment initiation. Upper-limb function improved, including reaching, grasping, hand-eye coordination, and functional use of the hands. Feeding-related independence also improved, including independent bottle feeding. Improvements in trunk and hip control and lower-limb weight-bearing were accompanied by the ability to maintain an upright standing posture with external support or contact. Social and developmental observations included increased interaction, smiling, visual tracking, babbling, and cooperation during therapy sessions. A post-treatment Children's Hospital of Philadelphia Infant Test of Neuromuscular Disorders (CHOP-INTEND) score of 51/64 was documented in a physiotherapy assessment [10]. The motor outcomes are reported descriptively because a comparable pretreatment score was not available for quantitative comparison. Continued rehabilitation, caregiver education, and home exercise instructions formed part of the patient's ongoing management. The pretreatment and follow-up functional findings are summarized in Table *1*, while the clinical timeline and nusinersen administration history are presented in Table *2*.

Functional domain	Before nusinersen (first dose: 23 June 2024)	During follow-up after nusinersen and rehabilitation
Head control	Fair head control	Good head control, full cervical motion and bilateral head turning
Rolling	Unable to roll independently; prompting required	Improved rolling; occasional prompting still required
Independent sitting	Unable to sit independently	Achieved independent sitting without support
Upper-limb function	Limited functional use	Improved reaching, grasping and hand-eye coordination
Trunk and hip control	Impaired	Improved
Lower-limb weight-bearing	Reduced weight-bearing ability	Improved weight-bearing ability
Standing	Reduced weight-bearing ability	Upright standing with external hand or examination-bed contact (photographed 3 November 2026, aged three years three months)
Feeding-related function	Required assistance	Independent bottle feeding
CHOP-INTEND score	No comparable pretreatment score available	51/64

TABLE 1: Pretreatment and post-treatment motor and functional outcomes

Descriptive comparison of motor and functional abilities before and during nusinersen treatment and multidisciplinary rehabilitation, based on physiotherapy assessments. The patient subsequently achieved independent sitting and demonstrated improvements in head control, rolling, upper-limb function, and lower-limb weight-bearing. A post-treatment CHOP-INTEND score of 51/64 was recorded. A comparable pretreatment score was unavailable, and no statistical comparison was performed.

CHOP-INTEND: Children's Hospital of Philadelphia Infant Test of Neuromuscular Disorders

Date or age	Clinical event	Documented finding / treatment
26 Jul 2023	Birth	Initially stable neonatal neurological examination
Age 1 month, 6 days	Clinical diagnosis of SMA	Diagnosis documented in the clinical record
Approximately 2 months	Neurological examination	Hypotonia, tongue fasciculations, areflexia and muscle atrophy
28 Sep 2023	Molecular laboratory report	MLPA showed homozygous SMN1 deletion and two SMN2 copies
23 Jun 2024	Nusinersen loading dose 1	12 mg intrathecal; treatment initiated at age 10 months, 28 days
08 Jul 2024	Nusinersen loading dose 2	12 mg intrathecal; 15 days after dose 1
28 Jul 2024	Nusinersen loading dose 3	12 mg intrathecal; 20 days after dose 2
22 Aug 2024	Nusinersen loading dose 4	12 mg intrathecal; 25 days after dose 3
22 Dec 2024	Nusinersen maintenance dose 1 (dose 5)	12 mg intrathecal
22 Apr 2025	Nusinersen maintenance dose 2 (dose 6)	12 mg intrathecal
24 Aug 2025	Nusinersen maintenance dose 3 (dose 7)	12 mg intrathecal
29 Dec 2025	Nusinersen maintenance dose 4 (dose 8)	12 mg intrathecal
04 May 2026	Nusinersen maintenance dose 5 (dose 9)	12 mg intrathecal; dose confirmed by treating team
03 Nov 2026	Follow-up clinical photographs	Age three years, three months; supported standing documented after 9 administrations
09 Nov 2026	Nusinersen maintenance dose 6 (dose 10)	12 mg intrathecal; dose confirmed by treating team

TABLE 2: Clinical timeline and nusinersen administration history

Chronological summary of the patient's clinical course and the 10 verified intrathecal nusinersen administrations. The recorded treatment period extended from June 23, 2024, to November 9, 2026. MLPA: multiplex ligation-dependent probe amplification; SMA: spinal muscular atrophy.

Clinical photographs obtained on November 3, 2026, illustrate upright standing posture with external hand or examination-bed contact (Figures *1*, *2*).



FIGURE 1: Standing posture following intrathecal nusinersen therapy

Clinical photograph obtained on November 3, 2026, when the patient was three years and three months old, approximately 28 months after initiation of intrathecal nusinersen therapy. The patient is shown maintaining an upright standing posture beside an examination bed while holding a caregiver's hand. The photograph demonstrates visible lower-limb weight-bearing with external hand contact but does not independently establish the precise degree of assistance required or the duration of standing.



FIGURE 2: Standing ability and postural control following treatment

Clinical photograph obtained on November 3, 2026, when the patient was three years and three months old, approximately 28 months after initiation of intrathecal nusinersen therapy. The patient is shown maintaining an upright standing posture beside an examination bed with one hand resting on the bed. The image demonstrates lower-limb weight-bearing with external contact. The duration of standing and the ability to stand independently cannot be established from the photograph alone.

Written informed consent for publication of this case report and the accompanying clinical photographs was obtained from the patient's father on behalf of his minor daughter.

Discussion

SMA Type 1 is a severe infantile-onset neuromuscular disorder characterized by progressive weakness, hypotonia, and respiratory and bulbar complications [2]. The clinical phenotype is influenced by several factors, including SMN2 copy number, although molecular findings alone do not establish the SMA subtype [9]. Before the availability of disease-modifying therapies, management focused on respiratory, nutritional, orthopedic, and rehabilitative support, and affected infants frequently experienced progressive motor deterioration, ventilatory dependence, or early mortality [3-5].

The introduction of nusinersen substantially changed the treatment landscape for SMA. In the ENDEAR randomized controlled trial, 51% of nusinersen-treated infants achieved a motor milestone response compared with 0% in the sham-control group, alongside improvements in survival-related outcomes [7]. The NURTURE study demonstrated favorable outcomes when treatment was initiated during the presymptomatic period [11]. Observational evidence has further documented motor improvements among patients receiving nusinersen [12], while individual reports have described improvements in respiratory

outcomes [13]. More recent studies, including the 2025 multicenter Turkish cohort [8], have emphasized the value of longitudinal motor, respiratory, and bulbar assessments. A five-year Norwegian follow-up study also documented longer-term outcomes in patients with SMA Type 1 receiving nusinersen [14]. Together, these studies provide evidence of treatment benefit while highlighting the importance of standardized outcome monitoring.

In the present case, SMA was diagnosed at one month and six days of age, whereas nusinersen treatment began at 10 months and 28 days, representing an interval of approximately nine months. The reason for this interval was not established in the available summarized records. Nevertheless, the subsequent treatment course was documented through 10 intrathecal administrations over approximately 28 months. During combined nusinersen therapy and multidisciplinary rehabilitation, physiotherapy assessments described improvements in head control, rolling, upper-limb function, trunk stability, and weight-bearing. Independent sitting was acquired after treatment initiation, and later assessments documented an upright standing posture with external support or contact. These findings are clinically relevant because independent sitting is not typically achieved in the untreated natural history of SMA Type 1 [2]. In typical childhood development, unsupported sitting is generally expected by approximately nine months, while independent walking is usually achieved by approximately 18 months [15]. Such developmental benchmarks provide context but do not represent appropriate individualized treatment targets for a child with SMA Type 1.

A post-treatment CHOP-INTEND score of 51/64 provided a standardized assessment of motor function. However, because a comparable pretreatment score was unavailable, the magnitude of improvement could not be calculated, and statistical significance could not be assessed. The functional milestones were therefore evaluated descriptively using the available clinical and physiotherapy findings. Furthermore, the observed gains occurred during combined nusinersen treatment and rehabilitation, preventing attribution of the improvements to either intervention independently. The photographs provide supplementary evidence of standing posture with external contact but do not establish independent standing or quantify the assistance required.

An important strength of this report is its detailed documentation of the molecular diagnosis, 10 nusinersen administrations, pretreatment and follow-up physiotherapy findings, and photographs demonstrating upright standing with external contact. Interpretation of the longitudinal motor trajectory is limited by the absence of paired serial motor scores and exact milestone-acquisition dates, together with limited standardized rehabilitation and longitudinal respiratory data. The final maintenance interval was longer than the prescribed four-month regimen. These factors provide clinical context for the observed functional progression during combined treatment and rehabilitation. This case report and accompanying literature discussion were prepared with reference to the CARE case-reporting guidelines [16].

Conclusions

This case documents the course of a child with SMA Type 1 who received 10 intrathecal nusinersen administrations over approximately 28 months alongside multidisciplinary rehabilitation in Bahrain. Compared with pretreatment motor findings, follow-up physiotherapy assessments documented improved head and trunk control, rolling, upper-limb function, and lower-limb weight-bearing, together with acquisition of independent sitting. At three years and three months of age, clinical photographs illustrated upright standing with external contact. These milestones are clinically noteworthy in the context of the severe untreated natural history of SMA Type 1. Standardized motor assessments are valuable for monitoring functional outcomes in patients with SMA, while comparable baseline and follow-up measurements are essential for quantifying changes in motor function over time. The approximately nine-month interval between diagnosis and treatment initiation underscores the importance of timely access to disease-modifying treatment. This report contributes a detailed real-world treatment and rehabilitation trajectory to the clinical literature on SMA Type 1.

Additional Information

Author Contributions

All authors have reviewed the final version to be published and agreed to be accountable for all aspects of the work.

Concept and design: Fawaz Obaid, Raafat Hamad Seroor H. Jadah

Acquisition, analysis, or interpretation of data: Fawaz Obaid, Noof Alkaabi

Drafting of the manuscript: Fawaz Obaid, Raafat Hamad Seroor H. Jadah, Noof Alkaabi

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Disclosures

Human subjects: Informed consent for treatment and open access publication was obtained or waived by all participants in this study. RMS Institutional Review Board (RMS-MH /IRB/ 2026-1256) issued approval 2026-1256. **Conflicts of interest:** In compliance with the ICMJE uniform disclosure form, all authors declare the following: **Payment/services info:** All authors have declared that no financial support was received from any organization for the submitted work. **Financial relationships:** All authors have declared that they have no financial relationships at present or within the previous three years with any organizations that might have an interest in the submitted work. **Other relationships:** All authors have declared that there are no other relationships or activities that could appear to have influenced the submitted work.

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