Original article

Arch Argent Pediatr. 2026;e202611045

Nutritional and metabolic profile of children with type 1 and 2 spinal muscular atrophy managed at a referral center

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ABSTRACT

Introduction. Spinal muscular atrophy (SMA) is a degenerative genetic disorder that primarily affects motor neurons. Disease-modifying therapies have altered the disease's natural course and led to new clinical phenotypes. Recent evidence suggests a systemic impact, including metabolic abnormalities.

Objective. To describe the anthropometric, nutritional, and metabolic profiles of children with SMA types 1 and 2 treated at a referral center.

Population and methods. We conducted a descriptive cross-sectional study at a tertiary care hospital between 2020 and 2023. We analyzed anthropometric measurements, lipid profile, glucose profile, and nutritional status in 17 children under 12 years of age with type 1 and 2 SMA during their first visit.

Results. A high prevalence of overweight, as well as abnormalities in lipid and glucose metabolism—including insulin resistance—was observed in a significant proportion of patients. An exploratory comparison between type 1 and type 2 SMA revealed a heterogeneous profile: some parameters, such as low HDL and high BMI, were more common in type 1 SMA, while elevated insulin levels were more common in type 2 SMA. These findings do not allow for definitive conclusions regarding differences between subtypes. The only patient treated during the presymptomatic stage had a nutritional and metabolic profile within expected parameters.

Conclusion. This study highlights the need for a multidisciplinary approach that includes nutritional and metabolic assessment in the follow-up of patients with SMA, tailored to their new phenotypes and increased life expectancy.

Keywords: *spinal muscular atrophy; metabolism; dyslipidemia; insulin resistance; nutrition.*

doi: http://dx.doi.org/10.5546/aap.2026-11045.eng

To cite: Venditti A, Busaniche J, Muntadas J, Squitín Tasende M. Nutritional and metabolic profile of children with type 1 and 2 spinal muscular atrophy managed at a referral center. *Arch Argent Pediatr.* 2026;e202611045. Online ahead of print 19-NOV-2026.

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Funding*: None.*

Conflict of interest*: None.*

Received*: 2-18-2026*
Accepted*: 8-28-2026*

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INTRODUCTION

Spinal muscular atrophy (SMA) is an inherited, progressive, degenerative disease that affects motor neurons and causes muscle weakness and atrophy. Mutations in the survival motor neuron 1 (*SMN1*) gene cause a functional deficiency of the encoded protein. Its prevalence is 1 in 10 000 live births, and without treatment, it is the leading genetic cause of infant mortality.[1,2] Traditionally, it is classified into phenotypes based on age of onset and motor milestones achieved.[3]

Recent studies show that the SMN protein is involved not only in motor function but also in the development and function of various tissues; SMA is now considered a systemic condition.[4] From a metabolic perspective, some reports suggest that severe muscle atrophy may predispose patients to fasting hypoglycemia and other catabolic states.[5] Furthermore, body composition differs from that observed in healthy children, with a predominance of overweight, increased fat mass, and decreased muscle mass, as a result of neuronal degeneration, an imbalance between energy intake and expenditure, and low physical activity.

A comprehensive approach has gained prominence in recent years. Early care guidelines have adopted a proactive approach and, together with pharmacological therapies, have improved prognosis and survival, giving rise to new clinical phenotypes.[6,7] The presence of dyslipidemia, impaired glucose metabolism, and cardiovascular risk in adults with type 3 SMA has broadened the focus beyond neurological and respiratory involvement to include metabolic, nutritional, and endocrinological aspects.[8,9] In light of this new paradigm, it is essential to generate evidence that allows for the characterization of previously neglected aspects.

This study aims to analyze and describe anthropometric characteristics and lipid and carbohydrate metabolism in children with type 1 and type 2 SMA 2 treated at the Hospital Italiano in Buenos Aires during 2020-2023.

Objective

To describe lipid and carbohydrate metabolism and nutritional status in patients with type 1 and type 2 SMA during their first outpatient visit with the neuromuscular diseases team at the Hospital Italiano de Buenos Aires.

POPULATION AND METHODS

Design

Descriptive cross-sectional study. We collected data by reviewing electronic medical records.

Setting

We conducted the study at the Hospital Italiano de Buenos Aires, a tertiary-level university hospital with an Interdisciplinary Unit for Pediatric Neuromuscular Diseases.

Population

The study included patients under 12 years of age with a confirmed diagnosis of type 1 or type 2 SMA who had their first outpatient clinic visit between 2020 and 2023. We excluded patients with intercurrent conditions or those under stress, as well as those without laboratory data.

Outcome variables

1. Anthropometry

Anthropometric measurements were expressed as percentiles using curves specific to SMA: weight, height, and BMI, based on the study by De Amicis et al.[3] These references were used because the growth curves from the World Health Organization and the Argentine Society of Pediatrics were based on healthy, ambulatory children who engage in regular physical activity, whose anthropometric measurements and body composition differ from those observed in SMA patients.

2. Glucose profile[10]

- Fasting blood glucose
- Insulin levels
- Glycated hemoglobin
- HOMA (homeostasis model assessment, insulin resistance index), calculated as (blood glucose × insulin level) / 405
- OGTT (oral glucose tolerance test)

3. Lipid profile[11]

- Total cholesterol, HDL, LDL, non-HDL cholesterol
- Triglycerides

In addition, glutamic-oxaloacetic transaminase (GOT), glutamate-pyruvate transaminase (GPT), liver ultrasound, gross motor development milestones, ventilatory and nutritional support, disease-modifying therapy, and age at onset.

Operational definitions

Hypercholesterolemia was defined as total cholesterol ≥200 mg/dl; low HDL as <40 mg/dl; hypertriglyceridemia as triglycerides ≥100 mg/dl in children under 10; fasting hyperglycemia defined

as blood glucose ≥100 mg/dl; insulin resistance defined as a HOMA index >0.7 in prepubertal children or >1.4 in pubertal children; elevated insulin levels >3.5 µU/ml; and glucose intolerance defined as blood glucose levels between 140 and 199 mg/dl at 120 minutes after the oral glucose tolerance test (OGTT).[10,11]

Puberty was defined according to Tanner criteria: stage ≥2 in females (breast development) and in males (testicular volume ≥4 ml). We applied reference values for insulin levels and the HOMA index according to pubertal stage.

Fasting conditions

The protocol specified that blood samples for blood glucose, insulin, glycated hemoglobin (HbA1c), and lipid profile should be collected after a minimum of 8 hours of fasting. It was not possible to retrospectively verify compliance with this requirement in all cases.

HbA1c, OGTT, and liver ultrasound indications

HbA1c testing was ordered based on clinical judgment as part of a metabolic evaluation. OGTT was indicated for patients with impaired fasting glucose who were able to cooperate with the test. Liver ultrasound was performed as part of follow-up whenever possible during the study period.

Variables with incomplete data were analyzed using the corresponding denominator based on the number of patients for whom information was available for each variable, as detailed in the results and tables.

Sampling and sample size calculation

All patients with type 1 or type 2 SMA treated at the Hospital Italiano de Buenos Aires were included.

Statistical analysis

Categorical variables were described as absolute and relative frequencies. Continuous variables were described as mean and standard deviation or median and interquartile range, depending on the observed distribution. Statistical analysis was performed using Stata® 16 software.

Ethical considerations

The protocol was approved by the Research Protocol Ethics Committee of the Hospital Italiano de Buenos Aires (April 18, 2024)—code 12627.

RESULTS

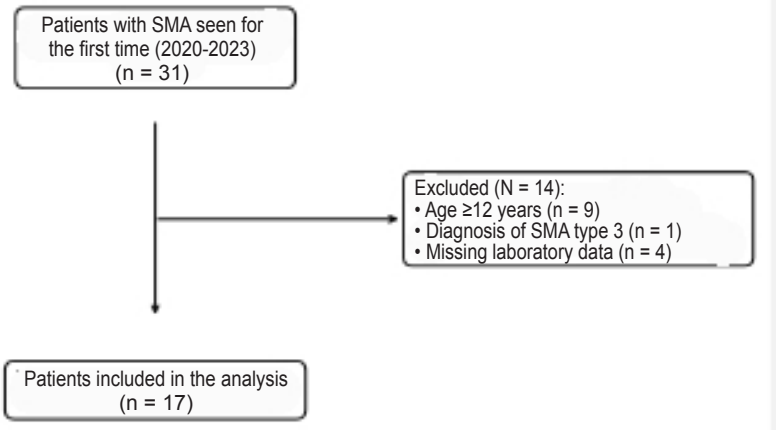
Participants

During the study period, 31 patients were seen for the first time; 9 were excluded because of age, 4 because laboratory results were unavailable, and 1 because of type 3 SMA. A total of 17 patients were analyzed (*Figure 1*). *Table 1* describes their characteristics.

Key findings

The median age at the first visit was 25 months (IQR 12-48), and the median age at the start of drug treatment was 9 months (IQR 6-19). One patient was diagnosed presymptomatically and received treatment at that stage. The female-to-male ratio was 1:1.4.

Figure 1. Patient selection flowchart



SMA: spinal muscular atrophy.

Table 1. Baseline characteristics of patients at the time of their first visit to the Hospital Italiano de Buenos Aires

Baseline characteristics	
Sex	
Female/male	7/10
Age in months at the time of the first visit, median (IQR)	25 (12-48)
Age in months at the start of drug treatment, median (IQR)	9 (6-19)
Type of SMA	
Type 1/type 2	12/5
Place of origin, n (%)	
AMBA	12 (71)
Rest of the country	5 (29)
Treatment received, n (%)	
None	4 (23)
Nusinersen	12 (71)
Risdiplam	1 (6)
Highest motor milestone achieved, n (%)	
Not sitting	11 (65)
Seated	6 (35)
Walking	1 (6)
Motor scales	
CHOP INTEND,* median (IQR)	29 (21-48)
HFMSE,** median (IQR)	21 (10-27)
Ventilatory support, n (%)	11 (65)
Invasive ventilation	8 (47)
Noninvasive ventilation	11 (65)
No support	6 (36)
Nutritional support, n (%)	11 (64)
NG-tube	3 (18)
Gastrostomy	8 (47)
No support	6 (36)

AMBA: Spanish acronym of Buenos Aires Metropolitan Area; SMA: spinal muscular atrophy; CHOP INTEND: Children's Hospital of Philadelphia Infant Test of Neuromuscular Disorders; HFMSE: Hammersmith Functional Motor Scale Expanded; SNG: nasogastric tube; IQR: interquartile range.
**n = 9; ** n = 4.*

BMI was above the 90th percentil in 41% of patients and none fell below the 25th percentile (*Figure 2*).

Table 2 shows the lipid and glucose profile results. Three patients (18%) had hypercholesterolemia; 4 (23.5%) had low HDL; 2 (12%) had elevated LDL; and 6 (35%) had hypertriglyceridemia.

Three patients (18%) had impaired fasting glucose; none had hypoglycemia. The median insulin level was 3.7 µU/ml (IQR 2.2-9.2), and 9 of the 13 patients evaluated (69%) had an elevated HOMA index.[12]

We performed liver ultrasounds on 16 patients, and steatosis was detected in 2. *Table 3* compares the main laboratory parameters between type 1 and type 2 SMA.

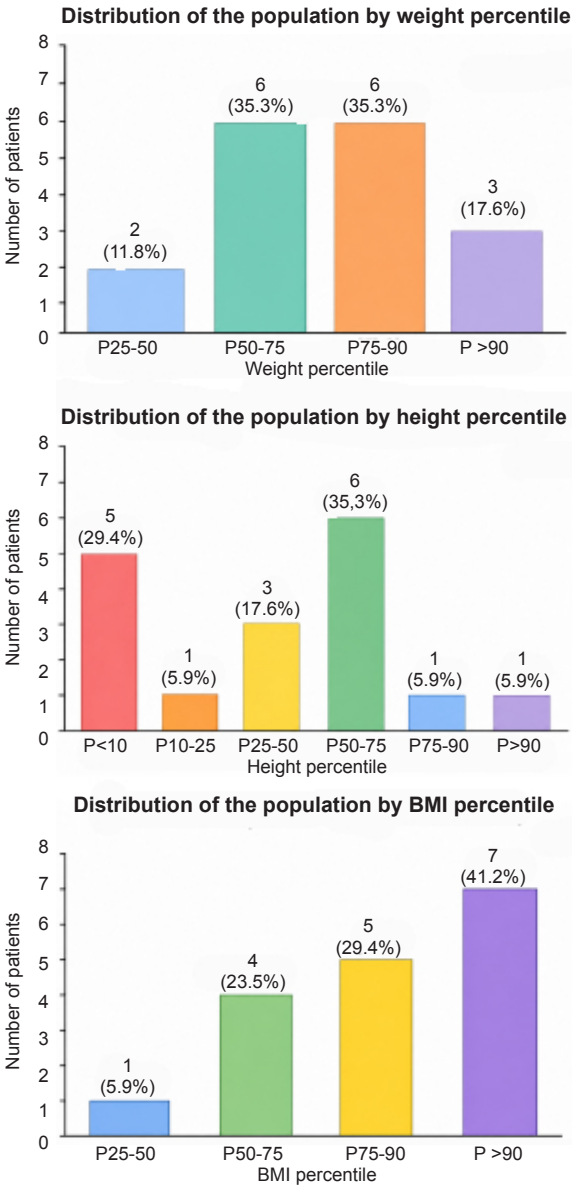
DISCUSSION

Anthropometry

Seven patients were classified as overweight according to BMI tables specific to SMA.[3] The guidelines suggest classifying sedentary individuals with a BMI >25th percentile on tables for the general population as overweight; in our study, only 6 patients were sedentary, and just 2 were of normal weight according to these criteria, highlighting the discrepancy between reference standards.[12-14]

Body composition is essential for accurately assessing nutritional status in SMA, but clinical practice rarely provides direct evaluation methods. One patient was evaluated using scintigraphy (65% fat mass), which revealed a BMI >90th percentile and carbohydrate intolerance but a normal lipid profile, highlighting the discrepancy between anthropometric indices and metabolism.

Figure 2. Anthropometric measurements of the patients (n = 17)



BMI: body mass index.

Table 2. Lipid and glucose profile of patients

Parameter	Median (IQR)	Expected normal values	n (%) out of range
Total cholesterol (mg/dl)	162 (147-177)	<200	3 (18)
HDL (mg/dl)	45 (35-57)	≥40	4 (23.5)
LDL (mg/dl)	63 (63-113)	<130	2 (12)
Non-HDL cholesterol (mg/dl)	113 (92.5-135.5)	<120	6 (35)
Triglycerides (mg/dl)	98 (70-122)	<100*	6 (35)
Blood glucose (mg/dl)	88.5 (85.5-94.5)	<100	3 (17.6)
Insulin (µU/ml)	3.7 (2.2-9.2)	<3.5 prepubertal	9 (53)
HOMA index	0.96 (0.51-2.16)	<0.7 prepubertal / <1.4 pubertal	9 (53)
HbA1c (%)	–	<5.7	0
OGTT at 120 min (mg/dl)	–	<140	2/2 between 140-199

**For children under 10 years of age, according to the Sociedad rgentina de Pediatría.*
HOMA: Homeostasis Model Assessment; HbA1c: glycated hemoglobin; OGTT: oral glucose tolerance test; IQR: interquartile range.

Table 3. Exploratory comparison of metabolic parameters according to the spinal muscular atrophy type

Parameter	Type 1 SMA (n = 12)	Type 2 SMA (n = 5)
Age (median, months)	34.5	38.8
HDL <40 mg/dl	5/12	1/5
Insulin >3.5 µU/ml	6/12	3/5
HOMA >0.7/1.4	7/12	2/5
BMI >p90	6/12	1/5

SMA: spinal muscular atrophy; HOMA: Homeostasis Model Assessment; BMI: body mass index; p: percentile.

Lipid metabolism

The median total cholesterol level was 162 mg/dl, with 3 patients >200 mg/dl; only 1 was overweight. Although no specific thresholds exist for SMA, this may suggest increased dyslipidemia risk, consistent with studies on neuromuscular diseases.[1,2,15]

The median HDL level was 45 mg/dl, and 23.5% had levels <40 mg/dl, possibly associated with low physical activity and altered body composition. The median LDL level was 63 mg/dl, with 12% >130 mg/dl, suggesting LDL was not significantly elevated.

Triglyceride levels had a median of 98 mg/dl, with 35% of patients having elevated levels. Previous studies have described hypertriglyceridemia and abnormalities in fatty acid oxidation in children with SMA.[1,15-17] The median non-HDL cholesterol level was 113 mg/dl, slightly below the desirable range for the pediatric population. This parameter reflects the burden of atherogenic lipoproteins and predicts long-term cardiovascular risk, especially in non-fasting samples. Wu et al. (2023) showed that childhood levels of this parameter predict cardiovascular events in adulthood more accurately than LDL-C, even when LDL-C levels are normal. This supports the importance of this parameter and the early monitoring of atherogenic risk in children with neuromuscular diseases.[18]

In addition, preliminary evidence suggests that alterations in the gut microbiome in patients with SMA may be linked to lipid metabolism disorders, opening new avenues for research and treatment strategies.[19]

Carbohydrate metabolism

According to the literature, SMA may predispose patients to disturbances in glucose homeostasis due to the factors mentioned above.[16,20]

Furthermore, recent studies reinforce the notion that metabolic abnormalities in SMA are part of a primary systemic phenomenon and not merely a result of immobility or altered body composition. Structural pancreatic abnormalities have been documented, including increased α cells and decreased β cells, which could explain dysfunction in glucose homeostasis even in the early or presymptomatic stages of the disease.[21]

Three of the patients evaluated had hyperglycemia. No episodes of hypoglycemia were recorded, a finding described particularly in patients with type 1 SMA.[5,22] However, the nature of the study does not allow us to confirm that the samples were obtained after the recommended fasting period. Furthermore, since these evaluations were conducted during periods of clinical stability, the likelihood of coexisting stressors—the primary triggers of hypoglycemia in these patients (e.g., surgery, fever, or infections)—is reduced (*Table 2*).

Amon patients with a calculated HOMA index 52% had values higher than expected for their age. Only 2 had OGTT, which showed abnormalities.

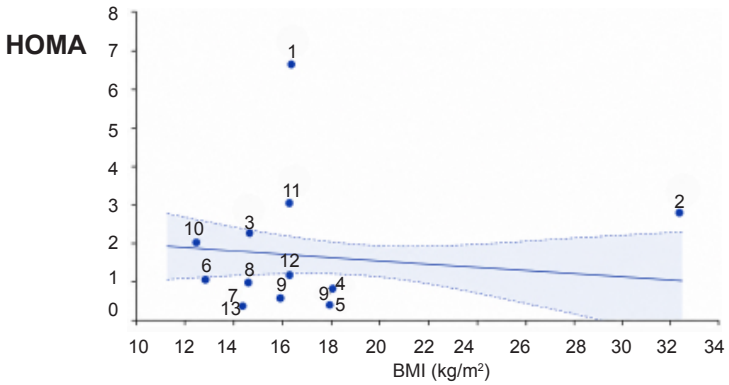
Although no formal statistical comparisons were performed, patients with a BMI above the 90th percentile according to specific curves tended to have higher HOMA indices (*Figure 3*).

Notably, the oldest patient in the cohort (8 years old, type 2 SMA), who began treatment at age 4, had the highest insulin levels (24.5 µU/ml) and HOMA index in the series. Although this isolated observation does not allow for establishing causal relationships, it suggests that metabolic abnormalities may become more pronounced with advancing age and longer cumulative disease duration. This finding underscores the importance of longitudinal metabolic monitoring in this population.

Hepatic steatosis

Hepatic steatosis was identified by ultrasound

Figure 3. Relationship between the HOMA index and body mass index



HOMA: Homeostasis Model Assessment; BMI: body mass index.

in 2 of 16 patients (12.5%). Both patients had dyslipidemia or insulin resistance. Although the sample size is small and ultrasound has limited sensitivity for detecting mild steatosis, these findings are relevant and consistent with recent literature.

The study carried out by Deguise et al. (2019) demonstrated a higher predisposition to the development of nonalcoholic fatty liver disease.[15] In that cohort, 37.5% of patients had at least one abnormal lipid profile, and a similar percentage showed steatosis in autopsy specimens. This prevalence exceeds that reported in the general pediatric population (0.7% in children aged 2 to 4 years) and is similar to that observed in obese children. Recent evidence also demonstrates that the liver exhibits early metabolic dysfunction secondary to SMA deficiency in hepatocytes, even before the onset of neuromuscular signs.[23]

Type 1 AME 1 vs. type 2 AME

We compared subtypes for descriptive purposes only; given the small sample size and lack of hypothesis testing, the percentage differences do not allow us to infer statistical associations or determine the direction or magnitude of differences between subtypes. With this caveat, patients with type 1 SMA had higher proportions of low HDL (42% vs. 20%), an altered HOMA index (58% vs. 40%), and BMI above the 90th percentile (50% vs. 20%) than those with type 2 SMA. However, the type 2 group had a higher proportion of patients with elevated insulin levels (60% vs. 50%), indicating a heterogeneous profile with no consistent trend.

These findings should inform hypotheses for future studies with larger sample sizes.

Impact of pharmacological treatment

A single patient received presymptomatic treatment (anecdotal observation). This patient was the only one who was ambulatory and had normal anthropometric and metabolic parameters, suggesting that early treatment could improve the metabolic profile through increased mobility and improved body composition.

Recent evidence shows that SMN deficiency causes primary pancreatic and hepatic dysfunction even before symptoms appear.[21,23] Therefore, early SMN replacement through gene therapy could exert broad systemic effects, mitigating metabolic, hepatic, and endocrine abnormalities beyond neuromuscular impairment. This finding suggests a potential systemic impact of these therapies that requires confirmation in large prospective studies.[24]

Limitations

The study has inherent limitations: a small sample size for a rare disease and a single-center cohort (Hospital Italiano de Buenos Aires).

One point worth considering is the choice of SMA-specific growth curves. Although this decision could be questioned, we opted for them because patients with SMA have such unique growth trajectories and body patterns that standard curves (WHO and Sociedad Argentina de Pediatría)—developed for a healthy, ambulatory, and physically active population—result in significantly inaccurate classifications, particularly an overestimation of low weight. However, we recognize that even SMA-specific growth charts have limitations inherent to this heterogeneous population.

In addition, direct body composition measurements were unavailable; we could not verify fasting status or dietary patterns, and data on HbA1c and OGTT were limited (3 and 2 patients, respectively). The small number of presymptomatic patients prevented comparisons between treatment subgroups.

Despite these limitations, the findings provide valuable information on a little-explored aspect and lay the groundwork for future research.

CONCLUSION

This study is one of the first descriptions in Argentina of the nutritional and metabolic profile of children with type 1 and type 2 SMA in the era of disease-modifying therapies. In our cohort, we identified a high proportion of children who were overweight according to specific growth charts (41%) and insulin resistant (53%). We found dyslipidemia, primarily low HDL (23.5%) and hypertriglyceridemia (35%), as well as hepatic steatosis (12.5%). These findings support the notion that SMA is a systemic disease with significant metabolic involvement beyond the classic neuromuscular phenotype.

Despite the study's design and sample size limitations, these results underscore the need to incorporate systematic metabolic evaluation into comprehensive, multidisciplinary care, especially given increased survival and changes in the clinical phenotype. Prospective studies with larger cohorts and longer follow-up periods will help establish the long-term clinical significance of these changes and assess the systemic impact of disease-modifying therapies. ■

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