

Health-related quality of life in children and adolescents with myelomeningocele: A cross-sectional study

Clara Meyer¹ , Lucía Pérez¹⁻² , Marcelina Carretero² , Santiago Portillo³ , Cristina Cortines¹ 

ABSTRACT

Introduction. Myelomeningocele (MMC) is the most severe form of neural tube defects, characterized by physical, cognitive, and social sequelae. Despite advances in treatment, its impact on health-related quality of life (HRQoL) remains significant. In Argentina, no studies have been found that assess HRQoL in this population using locally validated tools.

Objective. To assess lung function in children and adolescents with MMC and compare it with that of a healthy control group matched for age and sex.

Population and methods. An observational, analytical, cross-sectional study was conducted between December 2023 and December 2024. Patients aged 2 to 18 years were included at a tertiary care hospital. The PedsQL™ 4.0 generic version (proxy-report) questionnaire, validated in Argentina, was used to assess physical, emotional, social, and academic aspects in both sick and healthy children.

Results. The study included 68 children with MMC and 68 controls (median age: 9 years). The total PedsQL score was significantly lower in the MMC group compared with the controls (median 65.8 vs. 82.2; $p < 0.001$). The greatest differences were observed in the physical domain (median 56.3 vs. 96.9; $p < 0.001$); emotional functioning showed a smaller difference (median 70 vs. 80; $p < 0.01$). No differences in the HRQoL were observed between patients with or without prenatal surgery, ventriculoperitoneal shunt, or intermittent clean catheterization.

Conclusion. The study found that children with MMC have significantly lower HRQoL scores compared to their healthy peers.

Keywords: meningomyelocele; health-related quality of life; pediatrics; PedsQL.

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¹ Pediatric Clinic, Department of Pediatrics; ² Internal Medicine Research Unit, Department of Internal Medicine, Department of Pediatrics; ³ Pediatric Neurosurgery Unit, Department of Pediatrics; Hospital Italiano de Buenos Aires, Autonomous City of Buenos Aires, Argentina.

Correspondence to Clara Meyer: clara.meyer@hospitalitaliano.org.ar

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INTRODUCTION

Myelomeningocele (MMC), or spina bifida, is the most common congenital defect of the central nervous system, characterized by incomplete closure of the neural tube.¹ It is associated with hydrocephalus, paralysis, neurogenic bladder, and even death. The extent of the condition depends on the location and severity of the defect.^{2,3}

With advances in treatment, survival rates improved significantly.^{4,5} In Argentina, since the implementation of the law mandating the fortification of flour with folic acid, the prevalence has decreased by 60%,⁶ standing at 0.43 per 1000 live births according to the Argentine National Network for Congenital Anomalies (RENAC-Ar, by its Spanish acronym) in 2024.

Increased life expectancy brings new challenges in the quest to improve quality of life (QoL). The latter is defined as “an individual’s perception of their place in life within the context of the culture and values in which they live.”^{1,2,7} Health-related quality of life (HRQoL) subjectively assesses how health, healthcare, and health prevention and promotion efforts influence a person’s ability to function adequately and achieve overall well-being, encompassing physical, social, and psychological-cognitive dimensions.^{7,8}

There are numerous tools available for assessing HRQoL in chronic conditions. In Argentina, the Pediatric Quality of Life Inventory™ (PedsQL™) has been validated; it assesses the physical, emotional, social, and academic aspects of both sick and healthy children.⁹

In 2011, the MOMS (Management of Myelomeningocele Study) demonstrated that prenatal surgery is more effective than postnatal surgery in reducing sequelae and improving HRQoL.¹⁰ However, chronic complications can negatively affect HRQoL.¹¹ Recent studies show that patients with MMC have a lower HRQoL than the general population, with a particular impact on the physical and psychosocial domains.^{2,11,12}

At the hospital, the Myelomeningocele Clinic provides comprehensive care for patients with myelomeningocele. It is staffed by pediatricians, neurosurgeons, urologists, and orthopedic surgeons who work together in the same facility. Follow-up care coordinated by trained interdisciplinary teams is essential for the treatment of these patients and is associated with better long-term quality of life.¹³

In conclusion, given the scarcity of local data on HRQoL in pediatric patients with MMC and

the lack of regional studies that include matched controls,^{2,14} this study aims to evaluate QoL in a population treated at a tertiary care hospital.

OBJECTIVES

The main objective of the study was to evaluate the HRQoL in children and adolescents with MMC and to compare it with that of a group of healthy children and adolescents treated at the same institution.

As a secondary objective, we set out to describe the clinical and demographic characteristics of the population with MMC included in the study.

In addition, HRQoL was examined within the group of patients with MMC, stratified by the presence or absence of prenatal surgery, the need for a ventriculoperitoneal shunt (VPS), and the use of clean intermittent catheterization (CIC). These interventions were selected because they constitute key pillars of the clinical management of CM, with potential impact on motor, cognitive, and urological function, and, consequently, on the patient’s perception of HRQoL.^{5,9,15}

POPULATION AND METHODS

An observational, analytical, and cross-sectional study was conducted between December 2023 and December 2024. Patients aged 2 to 18 years from the Myelomeningocele Clinic were included as cases.

The control group consisted of healthy children who attended well-child visits at the institution’s outpatient pediatric clinics. For each child with MMC, a healthy child of the same sex and age (with a maximum age difference of 6 months) was selected until all paired visits were completed.

Selection criteria

Patients with neural tube defects other than MMC (lipomeningocele, meningocele, occult spina bifida) were excluded, as were those whose parents were unable to complete the questionnaire or declined to participate. In the control group, children with chronic illnesses or acute conditions at the time of evaluation were excluded.

Procedure and data collection

Contact with the MMC group was phased: first, communication was established via email; then, by phone; and finally, in person during scheduled appointments. The follow-ups were conducted sequentially until all pairs were completed.

The study was reviewed and approved by the Research Protocol Ethics Committee of the Hospital Italiano de Buenos Aires (protocol number 6863; file number 10962 in the Buenos Aires Computerized Health Research Registry [PRIISA, by its Spanish acronym]). Since this was an observational, risk-free study based on anonymous surveys and involving no intervention or modification of health care, the Committee approved the waiver of written informed consent, in accordance with international ethical guidelines (CIOMS).

Participation was voluntary; parents or guardians were informed of the study's objectives and were free to decide whether to participate, with no consequences for medical care. No identifying data were collected, nor was any sensitive information recorded that would allow for the identification of participants.

MEASUREMENT INSTRUMENT: PedsQL Generic version

The generic version of the PedsQL 4.0 questionnaire was used, with proxy reporting—that is, completed by parents or caregivers—to assess physical, emotional, social, and school-related dimensions. Responses are converted to a 0–100 Likert scale, with higher scores indicating better HRQoL.

The following variables of interest were collected: age, sex, place of residence, number of siblings, parents' educational level, special education teacher, age at diagnosis, MMC height, prenatal surgery, placement of a VPS, Chiari II malformation, concerns regarding sexuality, urological, neurosurgical, and orthopedic problems, and cognitive function.

Additional question

A self-designed question was included to explore parents' overall perception of their children's quality of life, which they were asked to rate as poor, fair, good, very good, or excellent.

Calculation of sample size

The total proxy-reported PedsQL score in patients with chronic diseases and healthy individuals was 73.4 (SD 16.1) and 82.2 (SD 13), respectively, according to the validation study in the Argentine population.¹⁶ For a 95% confidence interval (CI), 80% power, and a two-tailed test with a 1:1 ratio, it was necessary to include at least 44 patients in each group. However, during the study period, all patients who met the eligibility criteria

were included consecutively.

Since the validation literature reports results in terms of means and standard deviations, the sample size was calculated accordingly. Subsequently, based on the observed distribution of the PedsQL variables, the analysis was performed using nonparametric tests.

Statistical analysis

Categorical variables were described using absolute counts and percentages; continuous variables were described by their distribution, assessed using the Shapiro-Wilk test and histograms. Given the PedsQL distribution, nonparametric tests were used: the Wilcoxon test to compare HRQoL between patients with MMC and controls, and quantile regression (quantile 0.5) to estimate differences in the median across its domains. Results were expressed as absolute differences with a 95% confidence interval (95%CI). Bonferroni correction was applied for multiple comparisons. The analysis was performed using Stata version 16.

RESULTS

Baseline characteristics

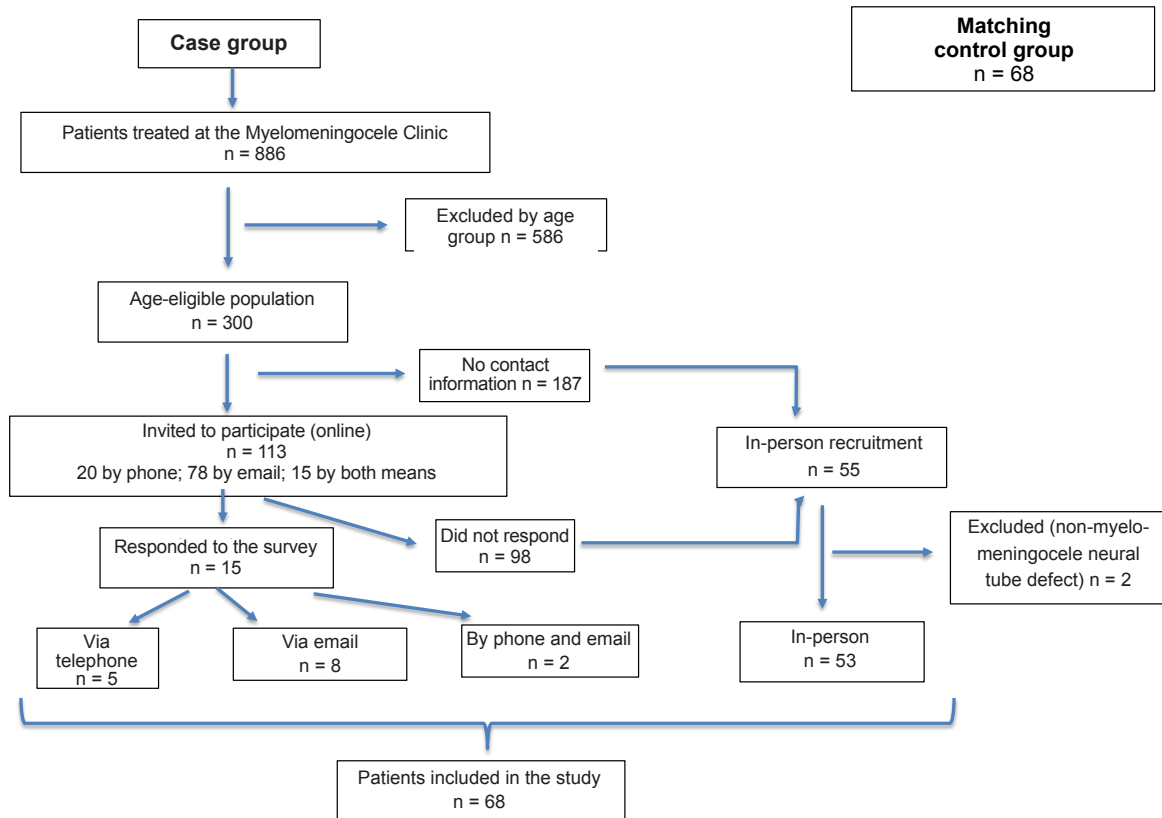
From a historical cohort of 886 patients treated at the Myelomeningocele Clinic, 300 met the age criteria (2 to 18 years) and constituted the eligible population. Of these, 113 had contact information and were invited to participate using non-mutually exclusive strategies (telephone and/or email). Fifteen responses were received. Due to the low initial response rate, 55 additional patients were enrolled through consecutive in-person recruitment during scheduled appointments. After excluding 2 patients with a diagnosis other than myelomeningocele, the final sample consisted of 68 patients with MMC. Sixty-eight healthy controls matched for age and sex were included (*Figure 1*).

The median age of the sample was 9 years (IQR 6–13); 51% were male; 52% of patients with MMC resided in provinces in the country's interior. The complete demographic characteristics of both groups are presented in *Table 1*.

In 57 cases, the diagnosis was made prenatally; the second trimester was the most common time of detection. The clinical characteristics of patients with MMC are presented in *Table 2*.

Eight patients (8/68) underwent prenatal surgery; 3 (3/8) had VPS, compared with 44 patients (44/60) who underwent postnatal

FIGURE 1. Flowchart



surgery. None of the patients who underwent prenatal surgery used a wheelchair, and all had normal positioning of the posterior fossa structures, with no evidence of Chiari II malformation.

Regarding the management of hydrocephalus, 20 of the 47 patients with a VPS experienced no device-related complications. In the remaining cases, the most common complications were malfunction and infection. Seventeen patients with MMC experienced seizures, and within this subgroup, 14 had a VPS implant. Regarding sexuality, 40 of the 68 parents expressed concern about their children's sexuality and reproduction. Additionally, 19 parents reported that their child had expressed concerns related to sexuality; of these cases, 12 involved patients over the age of 12.

Key findings

The total PedsQL score in MMC patients was 65.8 (95%CI 61.1–69.4), with physical health and psychosocial health scores of 56.3 (95%CI 48.3–64.2) and 72 (95%CI 66.5–77.5), respectively. In

all areas, they had lower scores than the control group ($p < 0.001$), with the greatest difference in physical health and the smallest in emotional functioning. The PedsQL scale results for MMC patients and the control group are shown in *Table 3*.

In the subgroups, no statistically significant differences in total scores were observed. Patients who underwent prenatal surgery had a median of 72.6 (95%CI 63.6–81.7), compared with 63 in the group that did not undergo surgery (95%CI 58–68.1). Regarding VPS, those who did not require it had a median of 60.9 (95%CI 49.6–72.2), and those who did had a median of 67.4 (95%CI 61.8–73). Finally, the use of CIC showed medians of 68.5 (95%CI 57.5–79.4) for the group without catheterization and 63 (95%CI 57.8–68.3) for the group with CIC. *Table 4* presents the HRQoL in these populations.

In the follow-up question, there were statistically significant differences ($p = 0.0095$). In the control group, responses were concentrated in the categories excellent (44%), very good (45%), and good (11%). In contrast, in the MMC group,

TABLE 1. Demographic characteristics of the study population

Characteristics	Total n = 136 % (n)	Healthy N = 68 % (n)	MMC N = 68 [% (n)]	p-value †
Age (years)				
2-4	13 (18)	13 (9)	13 (9)	
5-7	24 (34)	25 (17)	25 (17)	
8-12	32 (44)	32 (22)	32 (22)	NA
13-18	30 (40)	30 (20)	30 (20)	
Male	51 (70)	51 (35)	51 (35)	NA
Place of residence				
CABA	34 (46)	37 (25)	31 (21)	
Province of Buenos Aires	32 (44)	48 (33)	16 (11)	<0.01‡
Other provinces	32 (43)	12 (8)	52 (35)	
Foreigners	2 (3)	3 (2)	1 (1)	
Attendance at an educational institution	99 (125) [§]	100 (58)	99 (67)	NS
Inclusive education teacher	28 (36) [§]	15 (9) [¶]	40 (27)	<0.05
Education level of caregivers				
Elementary	3 (4) [§]	0 [¶]	6 (4)	<0.05 [¶]
Secondary	18 (23) [§]	10 (6) [¶]	25 (17)	
Higher education/college	79 (99) [§]	90 (52) [¶]	69 (47)	

† Chi-square or Fisher's exact test.

‡ Differences between groups were assessed between "CABA vs. others" and "Province of Buenos Aires vs. others," using Fisher's exact test with Bonferroni correction.

§ n = 126

¶ n = 58

|| Differences between groups were observed between the tertiary/university level and the primary and secondary levels, using Fisher's exact test with Bonferroni correction.

MMC: myelomeningocele; CABA: Autonomous City of Buenos Aires (by its Spanish acronym); NA: not applicable, since patients in both groups were matched for age and sex; NS: not significant.

the perception of excellent quality of life dropped to 23%, while 52% rated their health as very good, 16% as good, and 9% as fair; this last category was exclusive to the MMC group.

DISCUSSION

MMC is a congenital defect resulting from incomplete closure of the neural tube. Its complications can affect the quality of life of children and their families.⁵

Patients with MMC had lower HRQoL scores than controls across all dimensions, with the greatest impact observed in the physical domain and the least in the emotional domain—findings consistent with previous studies.³ According to Bakaniene *et al.*,¹² this discrepancy stems from processes of adaptive expectations and the recalibration of internal standards; because the condition is congenital, the patient develops their identity within their functional framework, facilitating early psychological adjustment. This phenomenon indicates that emotional well-being depends on psychosocial factors and

the supportive environment rather than on the severity of motor impairment.^{12,13}

The MOMS (Management of Myelomeningocele Study) compared prenatal versus postnatal correction, showing that intrauterine repair reduces the incidence of Chiari malformation type II and decreases the need for VPS repair (40% vs. 82% in those operated postnatally) and improves motor function and independent walking, with results similar to those of our study.¹⁰

Although previous studies suggest that prenatal surgery and CIC improve quality of life,^{15,17-20} our study did not find any significant differences, likely due to the small number of patients and the young age of the prenatal surgery group (n = 8), as this is a recently introduced procedure at our institution.

The lack of statistical significance should be interpreted with caution, given the risk of type II error and the exploratory analysis's low power. However, the trend toward higher scores in the prenatal group (72.6 vs. 63) is consistent with the literature and suggests clinical benefits that

TABLE 2. Clinical characteristics of patients with myelomeningocele (n = 68)

Characteristics	N
Time of diagnosis	
First trimester	3
Second trimester	33
Third trimester	20
At birth	10
After birth	1
Location	
Dorsolumbar	1
Lumbar	19
Lumbosacral	45
Sacral	3
Urination	
Toilet training	4
Diapers	14
CIC	50
Bowel movements	
Adequate control	23
Diapers	20
Evacuation enemas	19
Peristeen™	5
Polyethylene glycol	1
Walking	
No walking aid	10
Walker	3
Orthosis	10
Mixed	22
Canadian crutch	6
Wheelchair	17
Prenatal surgery	8
VPS	3
Arnold-Chiari malformation	0
Postnatal surgery	60
VPS	44
Arnold-Chiari malformation	47

MMC: myelomeningocele, VPS: ventriculoperitoneal shunt, CIC: clean intermittent catheterization.

warrant confirmation in future studies with larger sample sizes.

Furthermore, sexuality in the context of disability presents challenges that require a professional approach: although sexual desire is often preserved, sexual activity is reduced, and the perception of problems is greater.^{21,22} This significance is reflected in our study, in which more than half of the parents expressed concern about sexuality or fertility, and one-third perceived this same concern in their children. The significance of our findings is exacerbated in the local context in Argentina. Specifically, the fact that 52% of patients with MMC live in provinces in the country's interior highlights regional barriers to access to tertiary care centers.

This study provides the first local evidence on this underrepresented population, using a tool validated in the country. These results underscore the need to implement health policies that promote an interdisciplinary approach and effective decentralization of care.

The study has limitations, beginning with the absence of multivariate models to explore intragroup factors and the exclusive reliance on proxy reports, which may differ from the child's self-perception of emotional well-being. Furthermore, the in-person data collection (following a low response rate via mail) excluded patients with irregular follow-up, introducing potential selection bias that may affect the study's representativeness. Additionally, no

TABLE 3. Comparison of health-related quality of life assessed using the PedsQL questionnaire in children with myelomeningocele and in healthy controls

	Healthy n = 68	MMC n = 68	Difference in medians (95%CI)	p-value*
Total score	87 (83.4–90.5)	65.8 (61.1–69.4)	-21.7 (-27.3 to -16.2)	<0.001
Physical health	96.9 (92.1–100)	56.3 (48.3–64.2)	-40.6 (-47.6 to -33.6)	<0.001
Psychosocial health	85 (81.2–88.8)	72 (66.5–77.5)	-13 (-18.2 to -7.8)	<0.001
Emotional functioning	80 (73.6–86.4)	70 (63.6–76.4)	-10 (-17.9 to -2.1)	<0.01
Social functioning	95 (91.2–98.8)	65 (59.9–70.1)	-30 (-34.5 to -25.5)	<0.001
Academic functioning	85 (79.9–90.1)	70 (62.4–77.6)	-15 (-24 to -6)	<0.001

*Wilcoxon test. MMC: myelomeningocele.

Values are expressed as median and 95% confidence interval (95%CI).

TABLE 4. Health-related quality of life (PedsQL) in children with myelomeningocele by clinical subgroups (exploratory analysis)

	Prenatal surgery			VPS			CIC		
	No n = 60	Yes n = 8	p-value	No n = 21	Yes n = 47	p-value	No n = 18	Yes n = 50	p-value*
Total score	63 (58-68.1)	72.6 (63.6-81.7)	0.13	60.9 (49.6-72.2)	67.4 (61.8-73)	0.75	68.5 (57.5-79.4)	63 (57.8-68.3)	0.34
Physical health	56.3 (48.1-64.4)	62.5 (45-80)	0.2	59.4 (41.6-77.2)	56.3 (47.7-64.8)	0.4	59.4 (42.6-76.1)	56.3 (47.8-64.7)	0.48
Psychosocial	71.7 (65.1-78.3)	75 (66.3-83.7)	0.47	68.7 (55.8-81.6)	74 (65.9-82.1)	0.51	76 (63.2-88.8)	71 (63-79)	0.21

*Wilcoxon test.

VVPV: ventriculoperitoneal shunt, CIC: clean intermittent catheterization. Values are expressed as median and 95% confidence interval (95%CI).

socio-geographic differences were controlled for; these act as potential confounders related to access to healthcare, highlighting the need for decentralization policies. Finally, the small sample size of the prenatal surgery subgroup limits the statistical power to draw firm conclusions in this regard.

Among the study's strengths, we note that a healthy control group was included and evaluated during the same period, which allowed us to assess the impact of MMC on HRQoL. A standardized and validated instrument was used to ensure greater reliability and comparability of the results. Furthermore, the study provides evidence from an underrepresented population and can inform care strategies tailored to their needs. Finally, we were able to include more patients than estimated in the sample size calculation, enabling more accurate estimates.

CONCLUSION

The study found that children with MMC have significantly lower QoL compared to their healthy peers across multiple dimensions. This finding highlights the physical, emotional, and social burden associated with this condition, even among patients under active clinical follow-up. ■

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