

Health-related quality of life in children with Down syndrome

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ABSTRACT

Introduction. The characteristics of patients with Down syndrome (DS) may affect their quality of life. The objective of this study was to assess the health-related quality of life (HRQoL) in patients with DS.

Population and methods. This was a cross-sectional study to assess the HRQoL with the PedsQL 4.0 questionnaire administered to patients with DS and healthy patients aged 2 to 4 years in 2020–2021 at a teaching hospital.

Results. Each study group included 51 patients. The HRQoL score in children with DS was 82.1 compared to 88 ($p = 0.003$) in the population without DS. Psychosocial health was impacted the most ($p = 0.007$), especially in terms of social and school functioning ($p = 0.0001$).

Conclusion. Children with DS aged 2 to 4 years were observed to have a lower HRQoL. The psychosocial health scale was affected the most, especially in terms of social and school functioning.

Key words: health-related quality of life; Down syndrome; chronic conditions.

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INTRODUCTION

Down syndrome (DS) is the most common known genetic cause of intellectual disability, with an estimated worldwide prevalence of 1 in 1000¹ live births and a national prevalence of 19.2 per 10 000 live births.²

Children with DS have multiple comorbidities and associated diseases, such as various congenital malformations, oncohematological diseases, hearing and/or visual impairment, and growth retardation. In addition, they have developmental disorders with cognitive and motor delays and certain social and behavioral aspects that may affect their quality of life.³ With the advances in medicine, the life expectancy of individuals with DS has increased, with an 88% survival rate at 20 years old.⁴

Health-related quality of life (HRQoL) describes the impact of health status and/or treatments received on a person's quality of life.⁵ The measurement of HRQoL is an attempt to quantify the perspectives of individuals and/or caregivers through different domains, such as physical, emotional, and social well-being, considering the interaction of personal and environmental factors.

Different instruments have been validated in Argentina to measure the HRQoL.⁶ One of them is the generic Pediatric Quality of Life Inventory (PedsQL) 4.0 scale, used to assess the HRQoL of healthy or ill children.⁷

According to published studies, patients with DS have a lower HRQoL compared to children without DS; physical and social functioning are the most affected domains. No significant differences were observed in emotional functioning when compared to the population without DS.⁸⁻¹⁰

We have not found information regarding HRQoL in children with DS in Argentina; therefore, the objective of our study was to assess the HRQoL of children with DS seen at a teaching hospital compared to that of children without DS.

POPULATION AND METHODS

A cross-sectional study was carried out in children aged 2 to 4 years with DS seen at our hospital, a tertiary care teaching hospital, where the Department of Clinical Pediatrics consists of hospitalization wards, an intensive care unit, and multiple outpatient specialty clinics. Cases were identified in the electronic medical record, and healthy patients receiving outpatient follow-up were defined as controls.

The exclusion criteria for the population with DS were cancer treatment in the past year or hospitalization in the past month. In the case of patients without DS, those with chronic diseases or requiring hospitalization during the past year were excluded.

Families were invited to participate by e-mail between November 2020 and September 2021, and 3 reminders were sent. The control group was selected by convenience in a 1:1 ratio matched by sex, and were recruited in the same way. The informed consent of all participants was obtained.

The generic version of the PedsQL 4.0 questionnaire was used for the population aged 2 to 4 years and was completed by caregivers in a self-administered manner using Google Forms. The PedsQL includes 4 domains: physical, emotional, social, school functioning, and summary scores (total, physical, and psychosocial). The scale obtained ranges from 0 to 100; the higher the score, the better the HRQoL.⁷

The variables assessed were age, sex, place of residence, siblings, number of caregivers, relationship between the respondent and the child, caregiver's level of education and employment status, and attendance to the kindergarten. The type of kindergarten and comorbidities were also recorded in the population with DS (*Supplementary material 1 and 2*).

The sample size was estimated based on the mean number of patients with and without chronic conditions, 73.3 (SD: 16.1) versus 82.1 (SD: 12.9), respectively. For a 95% confidence interval, an 80% power for a two-tailed test with a 1:1 ratio, 43 patients were required in each group.⁷

Categorical variables were described as absolute numbers and percentages. Continuous variables were reported as median and interquartile range. Categorical outcome measures were compared using the χ^2 test, whereas the populations with and without DS were compared using the Wilcoxon test. Data were analyzed using the Stata software, version 15.

The protocol was approved by our hospital's Ethics Committee for Research Protocols (protocol number 5910, PRIISA [Platform for Electronical Registration of Health Research in Buenos Aires] file number 3725).

RESULTS

The electronic medical records of 80 patients with DS were reviewed. A total of

58 questionnaires were sent; 51 were completed, with a response rate of 87.9% (Figure 1). A total of 51 questionnaires in children without DS were completed.

The demographic characteristics of patients and caregivers are described in Table 1.

Our study found that children with DS had a worse HRQoL score compared to the population without DS in a statistically significant manner: 82.1 versus 88 ($p = 0.003$). Psychosocial health was the most affected domain ($p = 0.007$), especially in terms of social and school functioning ($p = 0.0001$) (Table 2).

DISCUSSION

This study found that children aged 2 to 4 years with DS have worse HRQoL compared to their peers without DS. The scores in the psychosocial health scale were poor; social and school functioning were affected the most.

To date, investigations on HRQoL in individuals with DS are limited. Although they were conducted using various measurement instruments, they all agreed that patients with DS have a worse HRQoL compared to patients without DS.⁹⁻¹²

In our study, the overall HRQoL score in children with DS was lower compared to the population without DS, and this is consistent with other studies using the same questionnaire.^{8,13,14} We found that our patients with DS have higher

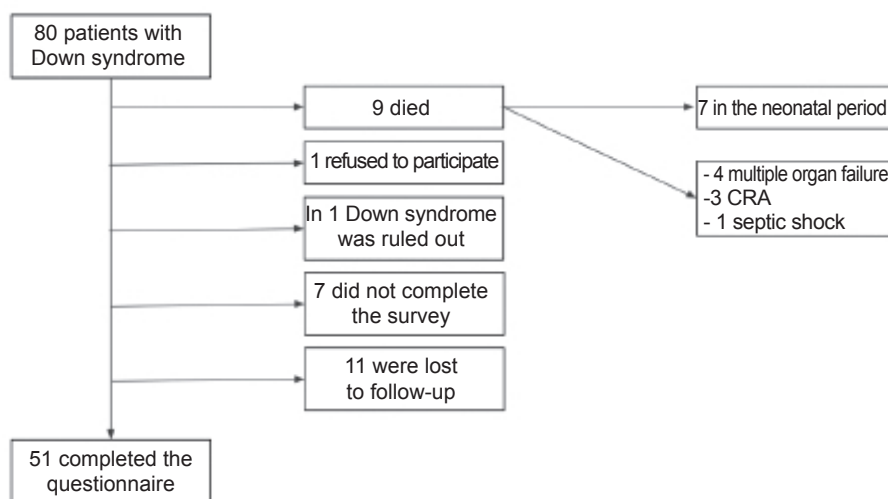
total score values compared to patients with DS from other studies published in Thailand, the United States, and Greece. This could be due to population cultural differences and expectations. Age may be another factor to take into consideration, because the rest of the studies included mostly school-aged and adolescent patients, while ours only assessed preschool patients, who might be more contained at home.

Consistent with the bibliography,^{8,13,14} we found that there were no differences in emotional functioning when comparing both study groups. Patients with DS had more difficulties in the psychosocial domain. Social and school functioning were the most affected domains, possibly due to the behavioral disorders and the intellectual disability in the population with DS. In addition, new perspectives on psychosocial disability suggest that disability is constructed by barriers imposed in a society with difficulties in understanding functional diversity. An environment that is inclusive and adapted to the particular needs of this population could contribute to their better development.

In terms of physical health, we found no statistically significant differences between both study groups, a result that differs from previous studies, where children with DS showed a higher impact on this area, probably secondary to motor retardation.^{8,13,14}

Most likely, HRQoL results may have been

FIGURE 1. Flowchart of patients with Down syndrome



CRA: cardiorespiratory arrest.

TABLE 1. Demographic characteristics of children with Down syndrome and control children without Down syndrome

	Children without DS (n = 51)	Children with DS (n = 51)	p value
Sex, n (%)			
Female	29 (56.9)	30 (58.8)	0.8
Male	22 (43.1)	21 (41.2)	
Age, median (IQR)	3.1 (2.5–3.9)	3.9 (2.8–4.2)	0.01
Place of residence, n (%)			
CABA	37 (72.5)	18 (35.3)	< 0.0001
Greater Buenos Aires	14 (27.5)	15 (29.4)	
Other provinces	-	18 (35.3)	
Siblings, n (%)	27 (52.9)	38 (74.5)	0.02
Number of siblings, n (%)			
1	20 (74.1)	22 (57.9)	0.4
2	5 (18.5)	12 (32.4)	
3 or more	2 (7.4)	4 (10.8)	
Number of caregivers, n (%)			
1	3 (5.9)	6 (11.8)	0.4
2	30 (58.8)	31 (60.8)	
3 or more	18 (35.3)	14 (27.5)	
Relationship to caregiver, n (%)			
Mother	44 (86.3)	45 (88.2)	0.5
Father	6 (11.8)	5 (9.8)	
Grandparent	-	1 (2)	
Other type of caregiver	1 (2)	-	
Maximum level of education completed by caregiver, n (%)			
Incomplete secondary education	-	3 (5.9)	0.2
Complete secondary education	2 (3.9)	2 (3.9)	
Incomplete tertiary education	2 (3.9)	1 (1.9)	
Complete tertiary education	5 (9.8)	8 (15.7)	
Incomplete university education	6 (11.8)	11 (21.5)	
Complete university education	36 (70.6)	26 (50.9)	
Caregiver's employment status, n (%)			
Full-time employment	33 (64.7)	16 (31.4)	0.02
Part-time employment	11 (21.6)	22 (43.1)	
Temporary employment	3 (5.9)	1 (1.9)	
Unemployed	1 (1.9)	2 (3.9)	
Unpaid care work (household and family)	1 (1.9)	5 (9.8)	
Retired	-	1 (1.9)	
Other	2 (3.9)	4 (7.8)	
Attendance to kindergarten, n (%)	44 (86.3)	40 (78.4)	0.3
Type of kindergarten, n (%)			
Regular	-	21 (41.1)	
Regular with special education aide	-	16 (31.3)	
Special education	-	1 (1.9)	
Informal education	-	2 (3.9)	

QR: interquartile range.

DS: Down syndrome.

n: number.

CABA: the City of Buenos Aires.

Table 2. Health-related quality of life in children with and without Down syndrome according to the PedsQL questionnaire

	Without Down syndrome (n = 51)	With Down syndrome (n = 51)	*p value
Total	88 (82.1–94)	82.1 (75–91.6)	0.003
Physical health	87.5 (81.2–93.7)	84.4 (71.8–93.7)	0.08
Psychosocial health	88.4 (82.7–94.2)	82.7 (73.07–92.3)	0.007
Emotional functioning	80 (70–90)	85 (75–90)	0.47
Social functioning	100 (90–100)	85 (65–95)	0.0001
School functioning	100 (91.6–100)	91.6 (75–91.6)	0.0001

Health-related quality of life scores are expressed as median and interquartile range.

*Wilcoxon test.

n: number.

influenced by the epidemiological situation secondary to the COVID-19 pandemic in both study populations, due to the lockdown, the suspension of in-person classes, and the barriers resulting from online activities.¹⁵ It is possible that physical health in children with DS was favored by family stimulation during the lockdown.

In recent years, the life expectancy of individuals with DS has increased; it is essential to have HRQoL serve as another indicator to assess health in this population.⁵ It is critical to know the aspects that influence the quality of life to contribute to and promote hospital health policies, and to train health care providers to improve the needs of children with DS. In addition, a profound debate on the scope of integration of individuals with functional diversity is still pending in our society.

Our study has several limitations: it was a single-center study with a limited number of participants, conducted using a generic electronic questionnaire based on the perspective of parents; the controls were not matched by origin or sex. There could have been a selection bias, as caregivers who did not complete the questionnaire could have had a negative perception of HRQoL.

However, we believe that our study provides valuable information useful to describe the HRQoL in children with DS in Argentina.

CONCLUSION

Children with DS aged 2 to 4 years were observed to have a lower HRQoL. The psychosocial health scale was affected the most, especially in terms of social and school functioning. ■

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Supplementary material available at: https://www.sap.org.ar/docs/publicaciones/archivosarg/2023/2756_CB_Fernandez-Scotto_Anexo.pdf

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