Neurological Sciences
https://doi.org/10.1007/s10072-025-08266-9

ORIGINAL ARTICLE

The impact of Down syndrome on patients' caregivers: a survey of an Italian paediatric cohort

Rossana Gnasso[1] · **Ayda Tavakkolifar**[1] · **Giuseppe Esposito**[1] · **Angela Palomba**[1] · **Stefano Palermi**[1] · **Antonio Picone**[1] · **Carlo Ruosi**[1]

Received: 4 March 2025 / Accepted: 21 May 2025

Abstract

Down syndrome is a chronic multisystem condition that not only affects individuals diagnosed with the disorder but also has a profound impact on their caregivers and families. Numerous studies have explored various aspects of life and overall quality of life for both patients and their families. Our research specifically focuses on the caregiver burden scale and was conducted at the Down Syndrome Outpatient Clinic at the Policlinic Hospital of Federico II, which operates every Thursday. The study utilized a questionnaire consisting of 10 questions, with a total of 100 patients participating. These questions aimed to evaluate different dimensions of caregivers' lives, including social, economic, and psychological factors. The scoring system is based on the Burden Scale for Family Caregivers (BSCF), which ranges from 0 (minimal burden) to 3 (maximum burden). Our findings reveal that certain aspects of caregiving are more significantly affected than others. For example, the median score for physical health is 105, indicating that this area is the most impacted by Down syndrome. In contrast, the median score for psychological health stands at 68.2, suggesting it is the least affected area.

Keywords Down syndrome · Caregiver burden · Paediatric population · Survey

Introduction

Down syndrome is the most commonly known cause of intellectual disability. One of the primary risk factors is advanced maternal age, which can also lead to a higher risk of miscarriage. This disorder is classified as a genetic condition caused by trisomy 21, meaning there is an extra chromosome 21. Numerous studies involving both humans and mice have been conducted to explore the role of this chromosome in producing various manifestations of the disorder. A significant aspect of these manifestations is the imbalance between genes on chromosome 21 and those on non-21 chromosomes [1].

Chromosome 21 is responsible for producing many important protective and functional proteins. Therefore, a deficiency in chromosome 21 can lead to a variety of complications and multi-system disorders, including cancers, musculoskeletal disorders, neurological issues and cardiovascular diseases. These manifestations are commonly observed in children with Down syndrome, but the degree of impact can vary among individuals. Essentially, the duplication of genes on this chromosome results in the production of over 200 proteins, leading to diverse clinical manifestations [2, 3].

Children with Down syndrome also have a notable tendency to develop cancers, particularly a high incidence of leukemias, while showing some protection against solid tumors [4–6]. Moreover, they often exhibit short stature and reduced neuronal density, which can result in cerebral hypoplasia. These children typically present with muscular hypotonia and congenital heart defects, such as atrioventricular septal defects [7, 8]. Additionally, they may experience specific health conditions, including epilepsy, certain autoimmune diseases like hypothyroidism, obstructive sleep apnea (OSA), vision and hearing problems [8, 19].

Currently, various therapeutic plans aim to improve the life expectancy of individuals with Down syndrome. In terms of diagnosis, screening plays a crucial role in developed countries to manage high-risk pregnancies and minimize the risk of spontaneous miscarriage. Primary prenatal

✉ Rossana Gnasso
rossana.gnasso@unina.it

[1] Department of Public Health, University Federico II of Naples, Naples, Italy

screening typically involves assessing several biochemical markers in maternal blood, such as alpha-fetoprotein levels and the size of nuchal translucency observed during ultrasound examinations [9]. If screening tests return positive results, further invasive diagnostic tests, such as amniocentesis and chorionic villus sampling, are recommended [10, 11].

Management of Down syndrome is highly individualized and specialized based on each patient's disabilities. Some individuals require continuous support and extensive medical intervention from birth, while others may have the ability to live independently [12, 13].

Regarding the quality of life for these patients, they face various challenges, and their family members and caregivers play vital roles in facilitating their active participation in the community. Patients with Down syndrome may encounter cultural barriers and stigma [14], which can further limit their social integration. Importantly, the well-being of caregivers and family members is significantly impacted by their responsibilities; they often experience emotional and economic burdens [15, 16]. This study aims to examine the effects of this multisystem disease on families and identify which aspects of their lives are most affected.

Methods

Study design

The main goal of our study is to survey the overall effects of this genomic multisystem disease on caregivers and families of affected children. We aim to identify and reveal the various challenges posed by the disease, particularly which domains of life are most affected. Our survey addresses social, economic, emotional, and several other aspects of life.

The study is conducted using a questionnaire based on the Burden Scale for Family Caregivers (BSFC), specifically a shorter version of this psychometrically validated tool [17]. This questionnaire is designed for both research studies and clinical practice, serving as a global measure of the perceived burden associated with home care. It consists of 10 questions that explore key socioeconomic and emotional aspects of life, providing insight into how caregiving responsibilities affect the well-being of caregivers (Fig. 1).

The questionnaire comprehensively assesses physical strain, emotional burdens such as feelings of guilt, depression, and anxiety, along with social limitations. The scoring system allows the examiner to evaluate the degree of burden experienced by caregivers; a higher score indicates a greater burden. In some cases, different subscales may be used to investigate more specific domains. The scoring scale ranges from 0 to 3, where 3 represents the highest level of burden and 0 the lowest.

The questionnaire was administered to the families and caregivers of our patients who attend our "Down Syndrome Clinics" every Thursday. The survey will be conducted throughout 2024, with the final questionnaires completed in December 2024. This study adheres to the guidelines set forth by the local ethics committee. It is important to note that all patient information and data will be treated anonymously and they were saved on a laptop which access password is known only to the staff involved in the study.

Treatment institution

This study was conducted in the Rehabilitation Unit of the Department of Public Health at the University of Naples Federico II, Italy. The survey took place in our outpatient clinic for Down syndrome, which is held every Thursday. Two resident doctors, A.T. and G.E., along with one professor of medicine, R.G., participated in this study.

Patients

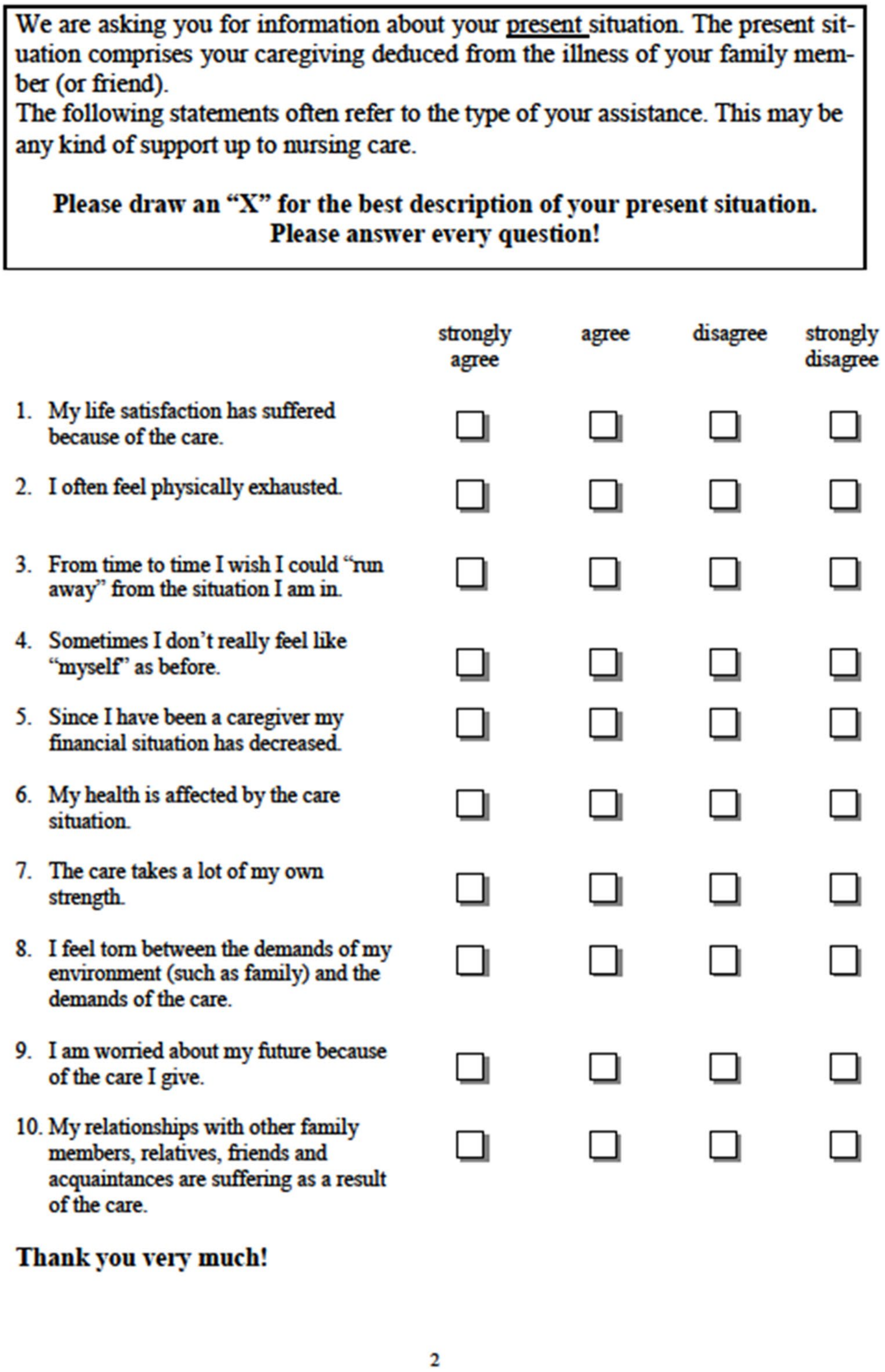
All patients presenting at our clinic are children aged from 0 to 17 years old. A total of 100 patients were involved in this survey. In our outpatient clinic, we evaluate various aspects of general musculoskeletal health, including all previous rehabilitation approaches such as physiotherapy, speech therapy, and psychomotor therapy.

We conduct a physical examination primarily focused on the patient's spinal column to assess the degree of scoliosis, kyphosis, and other related conditions. Additionally, we evaluate the patient's gait, muscular tone, and overall physical development. We also assess the patient's ability to perform daily activities and their verbal communication skills.

Results

The questionnaire in Fig. 1 is used for the survey in order to see the effect of Down syndrome on different aspects of life. The aforementioned questionnaire is composed of 10 questions and each question tries to challenge one specific domain of life. The main mentioned domains in the questionnaire are psychological, physical wellbeing and health, economic and social.

As we see in Table 1, each question has 4 answers going from completely right to "it not right". The scaling system of our questionnaire is based on the BSFC scale. The scale goes from 0 which means" it's not right "to a score of 3 which is it's completely right.



We are asking you for information about your present situation. The present situation comprises your caregiving deduced from the illness of your family member (or friend).
The following statements often refer to the type of your assistance. This may be any kind of support up to nursing care.

Please draw an "X" for the best description of your present situation.
Please answer every question!

	strongly agree	agree	disagree	strongly disagree
1. My life satisfaction has suffered because of the care.	☐	☐	☐	☐
2. I often feel physically exhausted.	☐	☐	☐	☐
3. From time to time I wish I could "run away" from the situation I am in.	☐	☐	☐	☐
4. Sometimes I don't really feel like "myself" as before.	☐	☐	☐	☐
5. Since I have been a caregiver my financial situation has decreased.	☐	☐	☐	☐
6. My health is affected by the care situation.	☐	☐	☐	☐
7. The care takes a lot of my own strength.	☐	☐	☐	☐
8. I feel torn between the demands of my environment (such as family) and the demands of the care.	☐	☐	☐	☐
9. I am worried about my future because of the care I give.	☐	☐	☐	☐
10. My relationships with other family members, relatives, friends and acquaintances are suffering as a result of the care.	☐	☐	☐	☐

Thank you very much!

2

Fig. 1 Shorter version of Burden Scale for Family Caregivers (BSFC) used in the survey [17]

A total of 100 patients participated in our study, which lasted for one year. Data collection took place from December 2023 to December 2024. It's important to note that all the patients included in our survey were pediatric patients. The participation of caregivers in this study was entirely voluntary, and none of the invited family members refused to partecipate. Therefore, we asked their families and caregivers to complete the questionnaire, while ensuring that all information remained confidential.

In the table below we will see a statistical analysis and therefore the final score of each question (Table 2):

Table 1 Shows the scoring system of the BSFC scale. As it is demonstrated, we can see that the scale is composed of 4 scores which go from 0 to 3

Answer	Score
It is completely right	3
Its almost true	2
Not much true	1
Completely wrong	0

Table 2 Title of each question in english along with their corresponding scores

Title of question In English	Score
Q1: from the time at which I have to be the caregiver of my family, my joy of living is decreased	42
Q2: most of the times I feel I am exhausted	133
Q3: sometimes I feel like I want to take myself out of this situation	41
Q4: Sometimes I don't feel like myself	59
Q5:my standard of living is decreased because of caregiving to my family	69
Q6: because of this assistance and caregiving my health state is decreased	54
Q7: this caregiving and assistance costs me lots of strength	128
Q8: I feel like I am divided in 2 person, one for assisting and caregiving and another for my existence	111
Q9: because of this assistance I am worried about my future	88
Q10: my relationships with other family members and friends are suffering because of this assistance	100

Table 3 Impact of down syndrome on four key aspects of life based on the statistical median

Domains that are most affected from down syndrome	Median number
Psychological	68,2
Physical health	105
Economical	69
Social	100

Based on our statistical analysis, the questions with the highest scores are as follows: Question 2 (score: 133), Question 7 (score: 128), Question 8 (score: 111), and Question 10 (score: 100). It is important to note that the lowest scores are associated with Question 1 and Question 3, which received scores of 42 and 41, respectively.

The questionnaire examines four main domains: psychological, physical health, social, and economic factors. Questions 1, 3, 4, 8, and 9 pertain to psychological issues. Questions 2, 6, and 7 are related to physical health, while Question 5 highlights the economic effects of Down syndrome. Finally, Question 10 addresses social impacts.

At the end of our analysis, we calculated the statistical median for each domain, which is presented in the table below (Table 3):

Based on the caregiver burden scale and the median values for each aspect of life, we find that the highest median score is 105, while the lowest is 68.2, which pertains to the psychological domain. This indicates that the domain most affected is physical health, while the least affected domain is psychological health.

Discussion and conclusion

Numerous studies conducted in various countries have investigated the quality of life of patients with Down syndrome, focusing on different aspects of their lives. However, very few studies have examined the socioeconomic and emotional impact of Down syndrome on families and caregivers. One study conducted by Gashmard et al. in 2020, highlighted the challenges families faced in accepting the reality of the diagnosis, particularly in the early stages. These families often experienced significant mental and physical difficulties. Many mothers felt compelled to leave their careers to fully care for their children, leading to considerable economic strain on the family [18, 19].

Barros et al. conducted research in Portugal in 2017, showed that the caregivers of children/adolescents affected by Down syndrome reported a higher burden compared to the caregivers of able-bodied children/adolescents [20].

Additionally, another study (Kuyken et al.) revealed that the psychological well-being of caregivers was severely affected, indicating that this is one of the most critical areas in need of support [21].

As with this survey conducted on the population affected by Down's syndrome, the similar study carried out by Tarantino et al., regarding the pediatric population with cerebral palsy (CP), showed that CP significantly worsen the psychophysical sphere of patients and the care burden of their families, exposing them to awful stress [22].

Our study analyzed the various impacts of this chronic genetic condition on the lives of caregivers and families of children with Down syndrome. Specifically, we evaluated the psychological aspect of life using questions 1, 3, 4, 8, and 9, which yielded a median score of 68.2 for this area.

We used questions 2, 6, and 7 for the second domain, physical health, resulting in a median score of 105. This suggests that physical health is more adversely affected than psychological health. The economic impact was assessed through question 5, which resulted in a median score of 69. Finally, we evaluated the social domain using question 10, yielding a median score of 100.

In conclusion, from these findings, the most affected aspect of life due to Down syndrome is physical health. Our study also indicates that psychological aspects are less impacted compared to other domains. Caregivers and families of patients with this chronic genetic condition face significant challenges across financial, emotional, social, psychological and physical dimensions. Down syndrome is a multisystemic condition that not only seriously hits the neuromotor abilities of subjects affected by disease, but also has profound impact on the quality of life for both patients and their family. A limitation of our study is that it only concerns family members of pediatric patients but it should be emphasized that, to our knowledge, this is the first Italian survey focused on the effect of Down Syndrome on psychophysical status of patients' caregivers.

Author contributions All authors contributed to the study conception and design. Material preparation, data collection, and analysis were performed by Rossana Gnasso. The first draft of the manuscript was written by Rossana Gnasso, Ayda Tavakkolifar and Giuseppe Esposito, and all authors commented on previous versions of the manuscript. All authors read and approved the final manuscript.

Funding Open access funding provided by Università degli Studi di Napoli Federico II within the CRUI-CARE Agreement.

Data availability All data generated or analyzed during this study are included in this published article.

Declarations

Ethical approval The questionnaires and the methodology for this study followed the guidelines given by the local ethics committee. All information from patients and their caregivers were treated anonymously, and all data were saved on a laptop where the access password was given just to the authors involved in the study.

Informed consent Verbal informed consent was obtained prior to the interview.

Consent for publication Verbal consent for publication was obtained prior to the interview.

Conflict of interest The authors declare no competing interests.

References

1. Nagaoka SI, Hassold TJ (2012) Hunt PA human aneuploidy: mechanisms and new insights into an age-old problem. Nat Rev Genet 13:493–504
2. Roizen NJ, Patterson D (2003) Down's syndrome. Lancet 361:1281–1289
3. Liu Y et al (2017) Systematic proteome and proteostasis profiling in human trisomy 21 fibroblast cells. Nat. Commun 8, 1212; First quantitative study of the trisomy 21 proteome, using fibroblasts from individuals with DS
4. Hasle H, Clemmensen IH, Mikkelsen M (2000) Risks of leukaemia and solid tumours in individuals with Down's syndrome. Lancet 355:165–169
5. Yang Q, Rasmussen SA, Friedma JM (2002) Mortality associated with Down's syndrome in the USA from 1983 to 1997: a population-based study. Lancet 359:1019–1025. https://doi.org/10.1016/S0140-6736(02)08092-3
6. Zwaan MC, Reinhardt DR, Hizler J, Vyas P (2008) Acute leukemias in children with down syndrome. Pediatr Clin North Am 55:53–70. https://doi.org/10.1016/j.pcl.2007.11.001
7. Vis JC, Duffels MGJ, Winter MM et al (2009) Down syndrome: a cardiovascular perspective. J Intellect Disabil Res.; 53:419–425. https://doi.org/10.1111/j.1365-2788.2009.01158.x
8. Volman MJ, Visser JJ, Lensvelt-Mulders GJ (2007) Functional status in 5 to 7-year-old children with down syndrome in relation to motor ability and performance mental ability. Disabil Rehabil 29:25–31. https://doi.org/10.1080/09638280600947617
9. Cuckle H (2016) Maymon R development of prenatal screening–a historical overview. Semin Perinatol 40:12–22
10. Savva GM, Morris JK, Mutton DE (2006) Alberman E maternal age-specific fetal loss rates in down syndrome pregnancies. Prenat Diagn 26:499–504
11. Bianchi DW, Crombleholme TM, D'Alton ME, Malone FD (2010) Fetology: diagnosis and management of the fetal patient Ch. 131. McGraw-Hill Medical
12. Alexander M et al (2016) Morbidity and medication in a large population of individuals with down syndrome compared to the general population. Dev Med Child Neurol 58:246–254
13. Bull MJ (2011) Committee on genetics, health supervision for children with down syndrome. Pediatrics 128:393–406
14. Skotko BG, Levine SP, Macklin EA (2016) Goldstein RD family perspectives about down syndrome. Am J Med Genet A 170:930–941
15. Xanthopoulos MS et al (2017) Caregiver-reported quality of life in youth with down syndrome. J Pediatr 189:98–104
16. Diaz KM (2020) Physical inactivity among parents of children with and without down syndrome: the National health interview survey. J Intellect Disabil Res 64:38–44
17. Pendergrass A, Malnis C, Graf U, Engel S, Graessel E (2018) Screening for caregivers at risk: Extended validation of the short version of the Burden Scale for Family Caregivers (BSFC-s) with a valid classification system for caregivers caring for an older person at home. BMC Health Serv Res.;18(1):229. Published 2018 Apr 2. https://doi.org/10.1186/s12913-018-3047-4
18. Alesi M, Pepi A (2017) Physical activity engagement in young people with down syndrome: investigating parental beliefs. J Appl Res Intellect Disabilities: JARID 30:71–83
19. Gashmard R, Ahmadi F, Kermanshahi SMK (2020) Coping strategies adopted by Iranian families of children with down syndrome: A qualitative study. Medicine, 99, e20753
20. Barros ALO, Barros AO, Barros GLDM, Santos MTBR (2017) Burden of caregivers of children and adolescents with down syndrome. Cien Saude Colet 22:3625–3634
21. World Health Organisation Quality of Life Assessment Group. (WHOQOL) (1994) The development of the WHO quality of life assessment instrument (the WHOQOL) in Quality of life assessment: International perspectives. eds. J. Orley and W. Kuyken (Germany: Springer-Verlag), 41–60. World Health Organization & Unicef. (2012). Early childhood development and disability: A discussion paper. Geneva: WHO
22. Tarantino D, Gnasso R, Migliore F, Iommazzo I, Sirico F, Corrado B (2021) The effects of COVID-19 pandemic countermeasures on patients receiving botulinum toxin therapy and on their caregivers: a study from an Italian cohort. Neurol Sci 42(8):3071–3077. https://doi.org/10.1007/s10072-021-05282-3