

# Humanistic, economic, and social impact of lysosomal storage disorders in Spain

EE413



**Authors.** Brignani E.<sup>1</sup>, de Castro-López M.J.<sup>2</sup>, Cruz J.<sup>3</sup>, Gonzalez-Meneses A.<sup>4</sup>, Morales M.<sup>5</sup>, Pintos G.<sup>6</sup>, Poveda J.L.<sup>7</sup>, Vilalta I.<sup>8</sup>, De Paz H.D.<sup>9</sup>

1. Asociación Catalana del Síndrome de Rett, Barcelona, Barcelona, Spain 2. Complejo hospitalariouniversitario de Santiago de Compostela, Santiago de Compostela, A Coruña, Spain 3. Asociación MPS-Lisosomales, Igualada, Barcelona, Spain 4. Hospital universitario Virgen del Rocío, Sevilla, Sevilla, Spain 5. Hospital universitario 12 de Octubre, Madrid, Madrid, Spain 6. Hospital Vall d’Hebron, Barcelona,Barcelona, Spain 7. Hospital La Fe, Valencia, Valencia, Spain 8. Hospital Sant Joan de Déu, Palma, Palma, Spain 9. Outcomes<sup>10</sup> (a ProductLife Group Company), Castellón, Spain

## Introduction.

Lysosomal storage diseases (LSDs) constitute a group of severe, disabling, and progressively worsening conditions with highly heterogeneous clinical presentations, requiring a comprehensive, multidisciplinary approach. The challenges faced by patients and their families are multifaceted, as these diseases profoundly affect their personal, familial, social, and professional lives, while also imposing a substantial financial burden. [1]. Although LSDs are classified as rare diseases, their collective impact can place significant pressure on the Spanish healthcare system (SHS). Nonetheless, the humanistic, economic, and social consequences of these conditions remain poorly understood, particularly within the Spanish context.

## Objectives.

To estimate the socio-economic impact of LSDs on the SHS, society, patients and caregivers.

## Methods.

This cross-sectional study followed five stages: 1) literature review; 2) focus groups with patients and healthcare professionals (HCPs); 3) questionnaire design; 4) survey; and 5) cost analysis. The study was guided by a Scientific Committee consisting of seven HCPs, representing various specialties (neuropaediatrics, paediatrics, orthopaedic surgery, internal medicine, psychology, and hospital pharmacy) and a patient representative. The study was approved by the Ethics Committee of Hospital Puerta de Hierro (Madrid).

The questionnaire was based on a literature review and insights from patients/caregivers and HCPs.



### Literature review

A comprehensive review of current evidence on the burden of LSDs in Spain was conducted using the international databases PubMed/MEDLINE and Google Scholar, along with clinical practice guidelines and consensus documents from leading Spanish scientific societies.

### Focus groups

Patients and caregivers were invited to participate by the Spanish MPS-Lisosomales Association providing insights into impact of the disease on their quality of life (QoL), daily activities, and the associated economic burden.

HCPs were also invited by the association to identify key aspects related to the management of LSD patients, as well as the healthcare resources required for diagnosis, treatment, and follow-up.

### Questionnaire

The study utilized two anonymous e-questionnaires: one for patients and another for caregivers. The patient questionnaire consisted of 52 questions, while the caregiver questionnaire included the 52 patient questions along with 11 additional questions specifically for caregivers, totaling 63. Both questionnaires were divided into four sections: 1) Sociodemographic characteristics, 2) Clinical variables, 3) Quality of life, and 4) Economic costs.

### Survey

Adult patients with a diagnosis of any type of LSD or their caregivers were invited to participate in the study through the web page and social media of the Spanish MPS-Lisosomales Association.

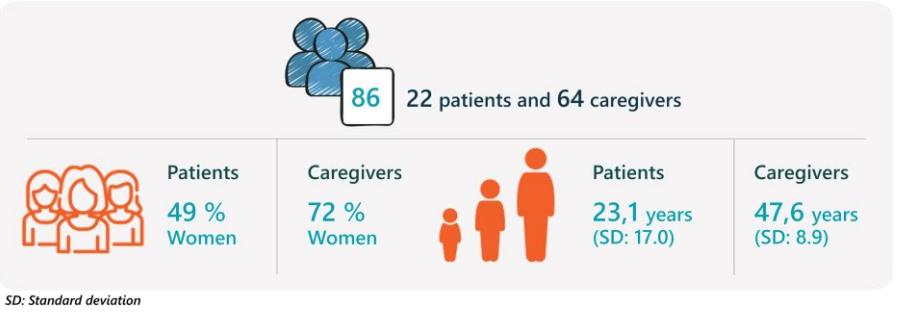
### Cost analysis/estimation

Direct costs for LSDs were estimated from healthcare utilization (medical visits, hospitalizations, tests, treatments, and formal care) using unitary costs (euros, 2024) from the eHealth database [2] and the Council of Pharmaceutical Associations [3], following Royal Decree-Law 8/2010 [4]. Costs were calculated annually, adjusting for long-term resources (e.g., surgery, devices, home/vehicle adaptations) by dividing values over the considered period. Specific treatment costs were based on the unitary cost and dosage for a 70 kg patient, averaging when multiple treatments were available. Laboratory test costs were estimated for the five most prevalent LSDs (Sanfilippo, Fabry, Hurler, Morquio, and Hunter) based on HCPs’ estimated annual resource utilization and patient percentage. Indirect costs from questionnaires included lost working hours (patient and caregiver) and informal care, with productivity losses calculated using the Spanish Statistical Office’s cost per hour [5]. Family-incurred costs covered patient-paid formal care, treatments, rehabilitation, non-SHS specialist visits, medical equipment, and home adaptations.

## Results.

### Sociodemographic characteristics

A total of 86 respondents (22 patients and 64 caregivers) completed the questionnaire, representing 86 patient-family units. The mean (SD) age of patients and caregivers was 23.1 (17.0) and 47.6 (8.9) years, respectively, with 48.8% of patients and 71.9% of caregivers being women. 28.1% of caregivers were unemployed due to their caregiving role, a rate higher than the national average in Spain (14%) [6].



Unemployment was higher among women than men (34.8% vs 11.1%). Additionally, 44.8% of employed caregivers reduced their working hours.

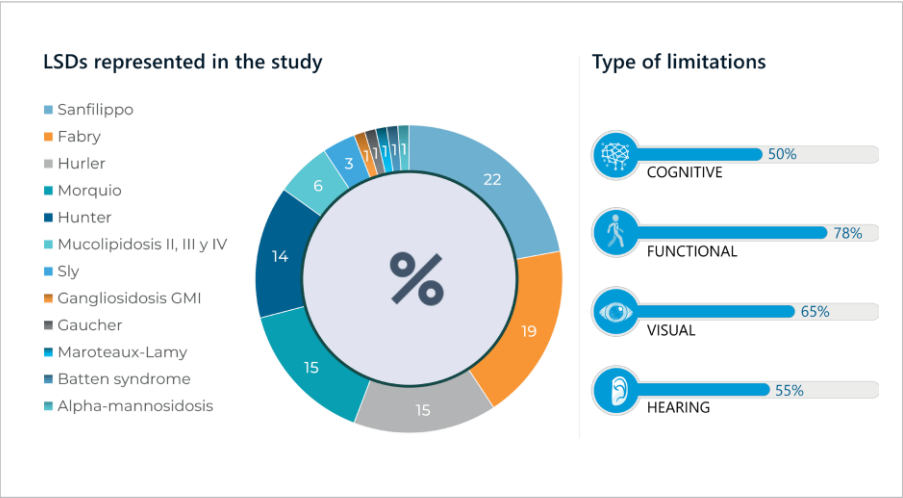
Table 1. Working and academic status of the participants in the study		
Working/academic status	Patients n (%)	Caregivers n (%)
Unemployed due to the LSD	6 (7.0)	18 (28.1)
Unemployed due to other reasons	1 (1.2)	1 (1.6)
Retired due to the LSD	14 (16.3)	2 (3.1)
Retired due to other reasons	1 (1.2)	4 (6.3)
Employed	10 (11.6)	35 (54.7)
Self-employed	3 (3.5)	3 (4.7)
Student	41 (47.7)	1 (1.6)
Not schooled	10 (11.6)	-

### Clinical characteristics

Sanfilippo (22.1%) and Fabry (18.6%) were the most represented LSDs followed by Hurler (15.1%), Morquio (15.1%), Hunter (13.9%).

The mean (SD) age at diagnosis was 9.9 (14.6) years, with a mean (SD) time from symptom onset to diagnosis of 4.3 (8.9) years.

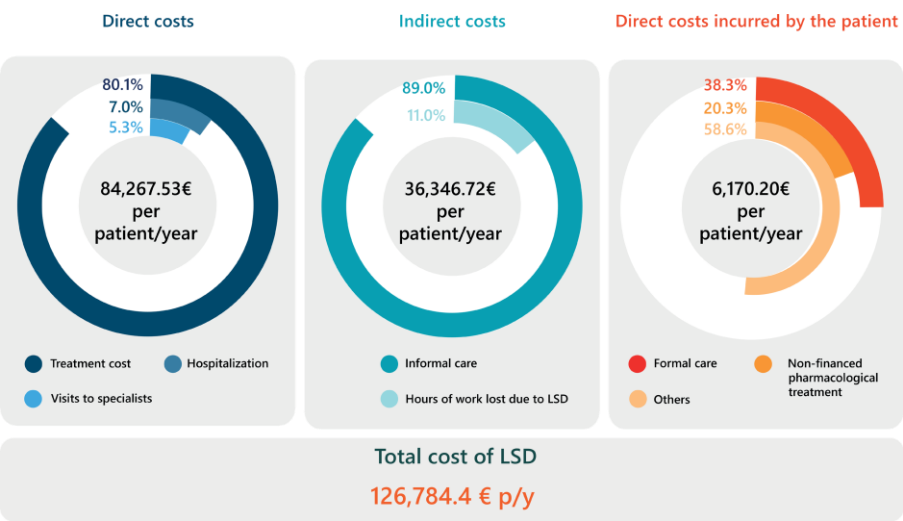
Most patients have some type of cognitive, functional, visual, or hearing limitation (as shown in Figure). Additionally, this limitation is severe to profound in 31.4% for cognitive, 29.1% for functional, 12.8% for hearing, and 8.1% for visual limitations, respectively.



### Economic costs

The resources with the least coverage by the SHS for LSD patients are psychologists (10.9% coverage, with a total of 5.0 visits per year), physiotherapists (30.8% coverage, with a total of 28.5 visits per year), speech-language therapists (51.0% coverage, with a total of 13.1 visits per year).

The total cost of LSD per patient-year (p/y) was 126,784.4 €, comprising direct costs to SHS (66.5%), indirect costs (28.7%), and direct costs incurred by the patient (4.9%). The mean direct cost was 84,267.53€ p/y, primarily due to treatment expenses (80.1%). Indirect cost was 36,346.72€ p/y, mainly corresponding to the informal care (89.0%). Direct costs incurred by the patient were 6,170.20€ p/y, primarily due to formal care (38.3%) and non-financed pharmacological treatment (20.3%).



## Conclusion.

LSDs create a significant economic burden on the SHS and affected families. Most indirect costs are due to informal care, highlighting the impact on caregivers. Furthermore, affected families experience a loss of employment opportunities and an increase in out-of-pocket costs. Thus, addressing the challenges of LSDs requires integrated strategies that consider both economic and social needs.

### REFERENCES

- Péntek M, Gulácsi L, Brodsky V, Baji P, Boncz I, Pogány G, et al. Social/economic costs and health-related quality of life of mucopolysaccharidosis patients and their caregivers in Europe. Eur J Health Econ. 2016;17 Suppl 1:89-98.
- Gisbert R, Brosa M. M. Base de datos de costes sanitarios y ratios coste-efectividad españoles: eSalud Barcelona: Oblikue Consulting, S.L.; 2007.
- BotPlus web. Consejo General de colegios de Farmacéuticos,Abril 2024.
- Ministerio de Sanidad. Listado de medicamentos afectados por las deducciones del Real Decreto-Ley 8/2010 2021.
- Instituto Nacional de Estadística. Resultados nacionales y por comunidades autónomas. Distribución salarial. Medias y percentiles por sexo y CCAA. 2021.
- Pérez Palazón P. Estimación de tasas de paro por provincia y sexo. 2021.