2025 ANNUAL ACTIVITY REPORT

LAL-D

LYSOSOMAL ACID LIPASE DEFICIENCY
PATIENT ORGANIZATION

We will continue advancing together. Every step we take as a community is an opportunity to transform lives and to build a fairer future for all families affected by LAL-D, regardless of borders

LALD.ORG

LAL-D Patient Organization (LAL-D PO)
Tax ID G66476045 | Association Registration Number: 607386
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LAL-D PO ANNUAL ACTIVITY REPORT 2025

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A year of international breakthrough, research and support for families

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1. Prologue

Lysosomal Acid Lipase Deficiency (LAL-D) is an ultra-rare, progressive genetic disorder that disrupts lipid metabolism. It results from a deficiency or complete absence of the lysosomal acid lipase enzyme, leading to the accumulation of fats in vital organs such as the liver, spleen, and cardiovascular system. This buildup can cause severe liver disease, multi-organ dysfunction, and premature arteriosclerosis. The most severe form, known as Wolman's disease, can be fatal within the first few months of life if not diagnosed and treated promptly.

Early detection is crucial. Prompt diagnosis—especially through newborn screening—enables immediate treatment, which can save lives and significantly improve patients' quality of life. Unfortunately, LAL-D is not currently included in neonatal screening programs in Spain. Addressing this gap remains a major challenge and a top priority for our association. We believe that prevention and early diagnosis are fundamental human rights that can mean the difference between life and death.

For these reasons, LAL-D PO works tirelessly to make early diagnosis and access to treatment a reality for all affected families.

2. Aims of the Association

LAL-D PO, formerly known as AELALD, is an association of patients and families committed to the following objectives:

- Raise awareness of LAL-D and other rare liver diseases among society and healthcare professionals.
- Promote early diagnosis and advocate for the inclusion of LAL-D in newborn screening programs.
- Support patients and families by providing information, resources, emotional support, and guidance on social and healthcare procedures.
- Foster scientific research and professional training by collaborating with hospitals, universities, and reference centers.
- Encourage international collaboration and networking with other associations and scientific organizations to share experiences and advance together.
- Defend the rights of patients by advocating for equity in access to diagnosis, treatment, and social support.

3. Activities and achievements 2025

3.1 Participation in Events and Congresses

- **January 14:** We participated, alongside other patient associations, in the XIII Cursa Benèfica de Malalties Minoritàries held in Badalona and organized by Javier Cabrera. This annual event fosters solidarity and raises awareness of rare diseases within the community.
- **January 17:** We attended the Rare Diseases meeting organized by Juntas Menos Raras at the University of Barcelona. The event focused on training, research, and data collection, strengthening support among families and opening new horizons in scientific research. It was an inspiring experience that reinforced the importance of working together for a more hopeful future for people with rare diseases.
- **February 14:** We took part in the II Conference for World Rare Disease Day at Parc Taulí University Hospital, under the slogan "Building a comprehensive and participatory model." We highlighted the importance of the voice of patient associations in moving towards more inclusive and effective care, and we appreciated the collaboration between professionals, researchers, and patients.
- **February 27:** We attended the Rare Diseases Conference at Vall d'Hebron Hospital, a key event for sharing knowledge and advances in the management of these pathologies. We reaffirmed our commitment to raising awareness of Lysosomal Acid Lipase Deficiency (LAL-D) and to networking with professionals and other associations.
- **February 28:** As part of World Rare Disease Day, we participated in the conference at Hospital de la Santa Creu i Sant Pau and carried out visibility campaigns, advocating for early diagnosis and neonatal screening, especially for diseases such as Wolman's Disease.
- **March 5:** We were present at the commemorative event for World Rare Disease Day in Catalonia, organized by the Department of Health. Topics included improvements in information and care coordination, the importance of the Xarxes d'Unitats d'Expertesa Clínica (XUEC), and the impact of art on the health and well-being of patients.

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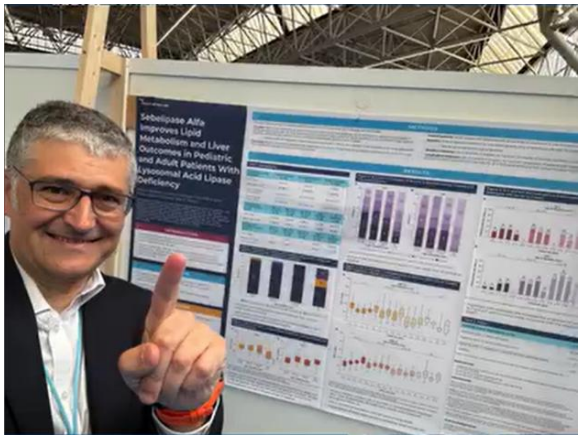
- **March 11:** We attended the Barcelona premiere of the documentary "Life in a Drop," produced by the MasVisibles association. This work highlights the importance of early detection and the expansion of newborn screening programs, showing how a simple test can save lives and improve the quality of life for many children and their families.
- **April 14:** We participated in the conference "Patient Associations in the Health System," organized by Parc Taulí together with other associations. We reflected on the role of the associative movement in the health system and shared experiences to promote more humane and coordinated care.
- **April 23:** We collaborated in the Sant Jordi solidarity initiative in Lleida, where roses were distributed to raise awareness of Lysosomal Acid Lipase Deficiency (LAL-D) and to share personal stories of overcoming adversity. We thank Tamara (@ladynemfis) for her energy and dedication.
- **May 8:** We attended the EASL Congress 2025 in Amsterdam, one of the most important international events in hepatology. We also participated in the General Assembly of the European Liver Patient's Association (ELPA), key meetings to discuss strategies, promote the defense of patients' rights, and give visibility to LAL-D. We highlighted the scientific publication on sebelipase alfa and its impact on the treatment of LAL-D.



- **June 12:** We participated in the 37th National Congress of the Spanish Society of Arteriosclerosis (SEA) in Sitges, sharing the patient experience in lipid units and highlighting the still unmet needs of those living with LAL-D. We reaffirmed the importance of listening to patients and promoting associations to advance early diagnosis and more humane care. As a result of our participation, the creation of a Lipid Working Group, called the Lipid Network, was promoted—a collaborative space that brings together patient associations and other stakeholders to work together to improve the approach to diseases related to lipid metabolism.

https://www.somospacientes.com/noticias/articulos/entrevistas/entrevista-hay-que-salir-y-construir-comunidad-mas-alla-de-nuestras-fronteras/

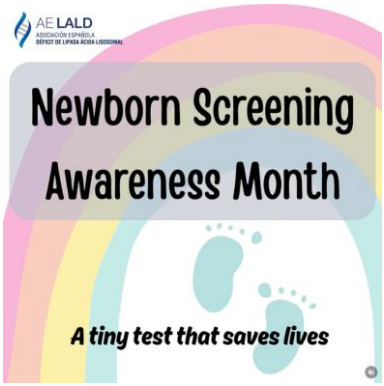
- **June 20-22.** Eduardo López represented the association at the 2nd edition of "ELPA @ Home" in Malatya (Turkey), an international meeting dedicated to LAL-D. The event addressed early diagnosis, advances in gene therapy, and the use of artificial intelligence in rare diseases. Spain was recognized for its commitment to research and diagnosis of LAL-D.

- **September 17:** We participated in the Screen4Care NBS Forum in Warsaw, exploring the future of neonatal genomic screening in Europe. The importance of ethics, patient empowerment, and international collaboration to advance the diagnosis of rare diseases was emphasized.

- **October 1:** We attended the conference "LAL-D CONNECT: A New Future for LAL-D," organized by Alexion Pharmaceuticals. We shared testimonials and updates on diagnosis, management, and treatment, and we appreciated the opportunity to give visibility to the association's work and to collaborate with professionals and patients.
- **November 18:** We participated in the Liver Rare Diseases Training Conference, organized by the Dr. Torrent-Farnell Foundation and the Malalties Minoritàries Platform, a key event to advance awareness and understanding of underdiagnosed liver diseases. Highlights included Eva's testimony on the daily challenges of living with LAL-D, Dr. Jesús Quintero's medical presentation on the importance of early diagnosis, and Eduardo López's intervention on the need for collaboration between health networks and patient associations.
- **Most important activity of the year:** On November 21 and 22, in Madrid, we held the VII Meeting of LAL-D Families and the meeting of the Committee of Experts, the central event of our association. It was a space for learning, collaboration, and mutual support, where families and professionals shared knowledge, experiences, and hope. These meetings reinforce the importance of community, research, and early diagnosis, and motivate us to continue moving forward together for the well-being of all those affected by LAL-D.

3.2 Awareness and visibility campaigns

- **Social media campaigns:** We launched initiatives such as "Think LAL-D," highlighting the importance of newborn screening (NBS) and the simplicity of diagnosis using a dried blood spot. We also promoted the TEAMING campaign, encouraging the public to support the patient and family community with just €1 per month.



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- **World Rare Disease Day (28/2/25):** We participated in multiple events and conferences at leading hospitals (Vall d'Hebron, Santa Creu i Sant Pau, Parc Taulí), giving a voice to patients and advocating for early diagnosis, research, and equity in access to treatments.
- **Collaboration in Documentaries and Short Films:** We contributed to the production and dissemination of the documentary "Life in a Drop" and the short film "La Batida," both aimed at raising awareness about the importance of early detection and the daily challenges faced by people with rare diseases.
- **"Nica's Journey" Campaign:** We collaborated on this initiative promoted by FEDER and Sant Joan de Déu Hospital, with the support of Alexion. Martina, a 14-year-old patient affected by LAL-D, participated alongside 99 other children from across Spain, sharing her voice and experience to raise awareness through this educational campaign. "Nica's Journey" is a solidarity initiative that seeks to increase visibility, raise awareness, and generate funds to improve the diagnosis of rare childhood diseases. It uses traveling dolls to symbolize the difficult journey families face in search of a diagnosis, supporting research within the Uniques network.

Members of:

3.3 Patient and Family Support

- **Annual Meeting of Families and Committee of Experts:** We held the VII Meeting of LAL-D Families and the Committee of Experts in Madrid—essential spaces for sharing experiences, strengthening the community, and advancing knowledge and comprehensive care. These meetings reinforce the importance of collaboration and mutual support.

Members of:

- **Guidance and Support:** We provide information on benefits and social resources. No one should face this journey alone, which is why we accompany and empower families at every step.
- **New International Website:** We launched https://lald.org in both Spanish and English, making information and resources more accessible to families worldwide.

4. Promotion of scientific research

Throughout 2025, LAL-D PO intensified its commitment to scientific research by actively collaborating in national and international projects aimed at improving the diagnosis, treatment, and quality of life of people affected by Lysosomal Acid Lipase Deficiency. We participated in the preparation and dissemination of leading scientific publications, providing the patient perspective and facilitating access to up-to-date information for both professionals and families.

Additionally, the association allocated resources and efforts to the funding of studies, convinced that only through research can we move toward a future with greater opportunities and hope for our community. This year, we have financially supported research initiatives and promoted collaboration among centers, researchers, and patient organizations, consolidating our position as a benchmark in advancing knowledge about LAL-D.

FEETEG Team, Drs. Giraldo, Roca and Serrano

Members of:

5. International Collaborations and Alliances

- **Transformation into LAL-D PO:** After ten years of work, the association has renewed itself and expanded its international reach, changing its name and welcoming new families. There are now 17 member families in Spain and 13 in other European countries and regions.

- **Steering Committee of MetabERN:** We are part of the Steering Committee of MetabERN, the European Reference Network for metabolic diseases.
- **Participation in European Projects:** We collaborate in initiatives such as Recon4IMD, which aims to improve the diagnosis and treatment of hereditary metabolic diseases, and in international studies on the needs of caregivers of people with LAL-D.

Members of:

6. Scientific Training and Dissemination

- **Courses and Training Sessions:** We completed the course "Rare Diseases: From Diagnosis to Therapy" at the University of Barcelona, where we explored research, patient registries, and knowledge transfer. We also participated in conferences on the role of patient associations in the health system, sharing experiences and best practices.

- **Scientific Publications:** We regularly disseminate relevant publications on LAL-D and have co-authored key articles, such as "Practical Recommendations for the Diagnosis and Management of Lysosomal Acid Lipase Deficiency with a Focus on Wolman Disease" and "Childhood to Adult Transition in Youth Patients with Lysosomal Acid Lipase Deficiency: 43 Recommendations from Experts." These works provide practical guidance for diagnosis, treatment, and the transition from childhood to adult care, contributing to improved quality of life for patients.

Hermida-Ameijeiras *et al. Orphanet Journal of Rare Diseases* (2025) 20:337
https://doi.org/10.1186/s13023-025-03852-8

Orphanet Journal of Rare Diseases

RESEARCH Open Access

Childhood to adult transition in youth patients with lysosomal acid lipase deficiency: 43 recommendations from experts

Alvaro Hermida-Ameijeiras[1], Javier Blasco-Alonso[2,3], Juan Luis Carrillo-Linares[4], María Luisa González-Dieguez[5], José Pastor-Rosado[6], Montserrat Morales-Conejo[7], Marcello Bellusci[8] and María Mercadal-Hally[9*]

Abstract

Background The process of transition from pediatric to adult care is crucial, especially in rare diseases such as lysosomal acid lipase deficiency (LAL-D). Unfortunately, this process is associated with poor outcomes, and many challenges still await to be addressed. This document provides recommendations on the pediatric to adult care transition in patients with LAL-D, based on available evidence and the experience of a panel of experts, which include specialists in the management of patients with LAL-D, and representative patients of the AELALD patient organization. Additionally, the main uncertainties and/or challenges encountered by the different stakeholders during the process are defined.

Main body A total of 43 consensus recommendations were developed across 5 areas. The consensus recommendations reflect the personal opinions and experiences of the participating experts supported with evidence when available. Overall, the main uncertainties and/or challenges faced comprise the patient's mistrust in the new medical team, the insufficient information received, or the lack of time, resources and institutional support. The management of adolescents/young adults during the transition to adulthood should be a joint effort between the patient, clinical center, and parents/caregivers. The objective of the transition process should be to empower patients and progressively encourage the self-management of their disease, and therefore patients and their families should be involved in all phases of the transition. Facilitating elements, such as standardized protocols, arose as

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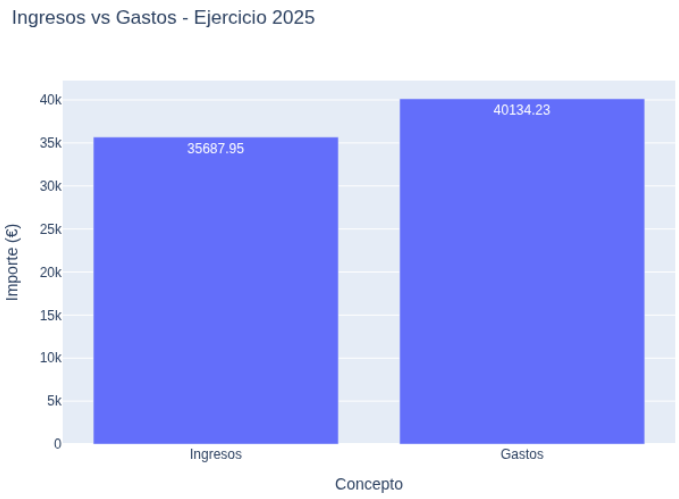
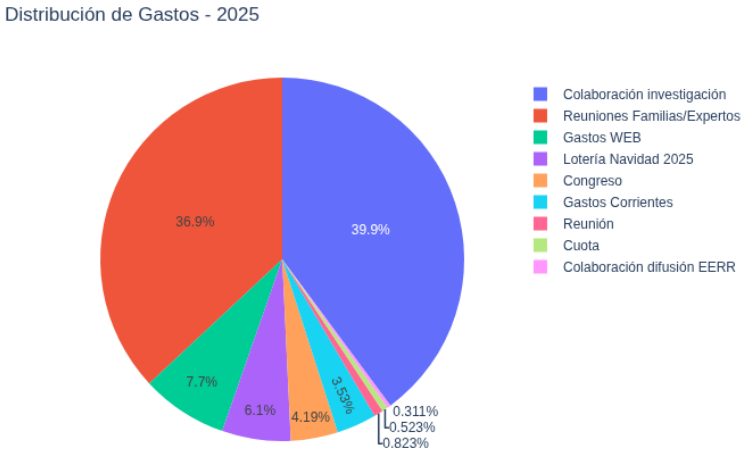
7. Financial Report

Below are the charts summarizing the financial results for the 2025 fiscal year.

Graph 1: Revenue vs Total Expenses



Graph 2: Distribution of Expenses



As shown in the charts, total income for the year amounted to €35,687.95, while total expenses reached €40,134.23, resulting in a final balance of –€4,446.28. The majority of expenditures were dedicated to research activities and support for families.

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8. Conclusion

The year 2025 has been crucial for LAL-D PO, marked by intense activity, growth The year 2025 has been pivotal for LAL-D PO, marked by intense activity, growth, and consolidation. This period represents a significant step toward an international approach: we have welcomed new families and have been able to support not only Spanish families, but also families from across Europe and other regions, strengthening our network and global response capacity.

We have made progress in visibility, training, research, and support for families, always maintaining early diagnosis and equity in access to healthcare as central priorities. The internationalization of our work has enabled us to share experiences, resources, and hope beyond our borders, ensuring that the voice of the LAL-D community is heard in all spheres.

We remain committed to ensuring that no LAL-D patient is left without diagnosis or support, and that every family—regardless of their place of origin—finds in our association a space for guidance, information, and advocacy for their rights.

9. Annexes (Photos, Links, Testimonials)

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https://youtu.be/4JYVFsdOS50?si=Fbg5jt90YLXC1WhH

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Links of interest:

- VII Meeting of LAL-D Families: https://lald.org/2025/11/25/727/
- LAL-D CONNECT: a new future for LAL-D: https://lald.org/2025/10/02/lal-d-connect-un-nuevo-futuro-para-lal-d/
- Where Would You Like Your Baby to Be Born?: https://lald.org/2025/10/02/donde-te-gustaria-que-naciera-tu-bebe/
- Interview with Eduardo López by Somos Pacientes: https://lald.org/2025/10/02/entrevista-que-somos-pacientes-hizo-a-eduardolopezsantamaria/
- AELALD Grows and Transforms: https://lald.org/2025/11/04/aelald-is-growing-and-transforming/
- Conference on Rare Liver Diseases: https://lald.org/2025/11/21/jornada-de-enfermedades-hepaticas-minoritarias/
- Participate in the International Study for LAL-D Caregivers: https://lald.org/2025/12/02/participa-en-el-estudio-internacional-para-cuidadores-de-pacientes-con-lal-d/

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TESTIMONIALS

Testimony from Martina's Family

Our daughter Martina was diagnosed with Lysosomal Acid Lipase Deficiency (LAL-D) when she was three years old, but the first signs that something was wrong appeared much earlier. Although we sensed that something was not quite right, physically she looked like a healthy girl, which led doctors not to suspect any serious illness.

For months, our concerns were dismissed, until a pediatrician—to whom we will always be grateful—decided to request a blood test. The results were alarming: her cholesterol was extremely high. The tests were repeated several times, and the results kept getting worse. In addition to cholesterol, her transaminase levels also began to rise, indicating possible liver damage.

From there, a long and distressing diagnostic journey began, lasting 18 months. Finally, what we feared was confirmed: Martina had a devastating rare disease, Lysosomal Acid Lipase Deficiency (LAL-D), which was little known at the time. Fortunately, we arrived just in time for her to participate in a clinical trial for enzyme replacement therapy, which was about to close recruitment.

The tests carried out then revealed the true extent of the damage: extremely high cholesterol, hepatomegaly, grade 3 liver fibrosis... Her health was in serious danger and, without access to treatment, the prognosis would have been devastating.

Today, ten years later, Martina leads a completely normal life. Every 15 days she receives her intravenous treatment at the hospital, her blood tests are within normal ranges, and she can do the same activities as any other girl her age. Thanks to the National Health System covering this treatment, she has been able to maintain an excellent quality of life.

As parents, we are convinced that early diagnosis is essential. Detecting the disease early can prevent irreversible damage and give children a chance to grow up healthy. That is why we believe it is urgent and necessary to incorporate neonatal screening for LAL-D in Spain. No family should go through what we experienced, when a simple test at birth could change the future for these children.

Testimony authorized by Martina's family (LAL-D PO, 2025)

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