ELA

EUROPEAN LEUKODYSTROPHIES ASSOCIATION

Presentation file




ELA is an association that fights against leukodystrophies

ELA is an association of parents who have been working together for 30 years to overcome leukodystrophies.

In 1992, when the Association was founded by Guy Alba with Pascal Prin and Cathy Schorderet, all that was known about leukodystrophies was that they were genetic neurodegenerative diseases and that the days of the people affected by these terrible diseases were numbered, without being able to offer a more precise prognosis.

Recognised as a public utility since 1996, it is ELA's investment and the unfailing commitment of families, researchers, partners, sponsors, supporters and donors that have made it possible to identify more than thirty forms of leukodystrophy.

In the 30 years of ELA's existence, thousands of children, adults, parents, spouses and siblings have been supported on a daily basis to face the demanding battle against the progression of the disease.

This is why ELA is relentlessly pursuing its efforts to raise awareness, mobilise, inform and collect donations from as many people as possible.

Our missions

- Supporting and helping families affected by the disease
- Supporting medical research on leukodystrophies
- Raising public and medical awareness
- Developing action at international level

Some key figures

3 to 6 children per week
are born with leukodystrophy in France (20 to 40 in Europe).

46.7 million euros invested
through 537 leukodystrophy research programmes.

14.7 million euros devoted
to support families

ELA is made up of determined, committed and supportive people, all of whom abide by the motto: "Together, stronger against leukodystrophies". This chain of solidarity allows the Association to provide families with all the attention they need and the means to ease their daily lives. At the same time, ELA gives priority to medical research and clinical trials.

Key dates for ELA

1 992
- Creation of ELA with clear objectives: to finance medical research, to support families, to raise public awareness and to develop its action at international level.
- Identification of the gene responsible for adrenoleukodystrophy.

1 994
- "I run, you sponsor, he lives": first event for schools, which will become known as "Mets tes baskets et bats la maladie" ("Wear your trainers and beat disease").
- Organisation of the first respite weekend for families at Center Parcs in Sologne.

1 996
- ELA recognised as a public utility: after only four years of existence, the ELA association has been granted the Recognition of Public Utility, undeniable proof of the quality of its work.

1 998
- Creation of ELA Belgium

2000
- Zinédine Zidane joins the ELA team: extremely touched by ELA's fight, he gets involved and brings the association to the forefront.
- Creation of ELA Switzerland

2001
- Creation of ELA Spain

2004
- Creation of the ELA Research Foundation. François-Henri Pinault, Franck Riboud, Florent Pagny and Zinédine Zidane join the Supervisory Board.
- Launch of the first ELA Creed: "Pour toi, Pour Moi" ("For you, for me") by Philippe Claudel

2005
- Launch of Stades en fête in Rennes.
- The Evian Masters alongside ELA.

2006
- Creation of ELA Luxembourg

2007
- "Les Stars se dépassent pour ELA": a charity show concept in prime time on TF1.

2009
- Publication by Prof. Patrick Aubourg and Dr. Nathalie Cartier on the first results of a gene therapy trial on adrenoleukodystrophy supported by ELA. An innovation that opens up prospects for the treatment of leukodystrophies but also for other diseases.
- Creation of ELA Italy

2011
- Launch of the first "Mets tes baskets" (Wear your trainers and beat disease) campaign at work - The operation "1 step = 1 euro cent" during the working day is an immediate and overwhelming success with employees.

2012
- ELA celebrates its 20th anniversary by gathering its young ambassadors and sponsors at Disneyland Paris and by creating ELA Indian Ocean.

2013
- Creation of ELA Germany
- Augusto Odone passes away, a pioneer parent in the fight against adrenoleukodystrophy.

2014
- ELA and MedDay launch an international clinical trial to test a molecule for the treatment of an adult form of leukodystrophy. The trial is being launched simultaneously in Germany, France and Spain.

2015
- Creation of ELA International - Based in Luxembourg, ELA International was created to federate all ELA structures (Germany, Belgium, Spain, France, Italy, Luxembourg, Switzerland, Indian Ocean) and to develop research by bringing together researchers from around the world.

2017
- The "Wear your trainers" campaign for the general public - After schools and companies, it is now the turn of the general public to get involved in the fight against leukodystrophies.

2018
- ELA back on TV with the new spot in which Zidane coaches researchers - In this spot, our godfather plays a coach who motivates a team of real researchers. It is broadcast on many television channels and in cinemas.
- ELA's innovation in fundraising - With the launch of an excellent product to reach major donors in France and internationally: the crystal foot, Zidane's Crystalfoot.

2019
- www.leuconnect.com - The international platform is launched to create patient cohorts and promote clinical studies in the field of leukodystrophies by involving several countries.
- ELA takes advantage of International Rare Disease Day - ELA is making this day a key event to shed a light on leukodystrophies and to provide updates about the progress of research.

2020
- ELA develops its "Mets tes baskets" ("Wear your trainers") app. It allows connected challenges to be organised at school, in companies or as part of public operations.
- Launch of the first epidemiological studies on the Leuconnect platform.

2021
- Marketing authorisation for gene therapy for the treatment of cerebral adrenoleukodytrophy.
- First annual families/researchers virtual meeting with an international dimension.
- First edition of the "Wear your trainers and beat disease at work" International Day organised by all ELA structures.
- Exceptional auction "The Stars' Hearts beat for ELA" in partnership with Christie's.

Leukodystrophies

Leukodystrophies are genetic neurological diseases that affect the myelin (white matter) of the nervous system and lead to very severe disabilities, progressively causing the loss of all functions: sensory, motor, mental (sight, hearing, locomotion, memory, etc.). They often lead to death. Children with leukodystrophy have symptoms that can appear as early as the first year and get progressively worse as they grow. There is currently no treatment available for these rare diseases.

Myelin is the white matter of the brain and spinal cord. It envelops the nerve fibre in the manner of an electrical sheath: this is what allows the proper conduction of nerve messages.

When this sheath is damaged, the current no longer flows and nerve messages are interrupted.

"When the power goes on, life goes on!"

The nervous system is like an electric current that flows through a sheath.

For a child with a leukodystrophy, the current has difficulty flowing through the sheath.

https://www.youtube.com/watch?v=OckKLsiaLNE

Our missions

Our missions

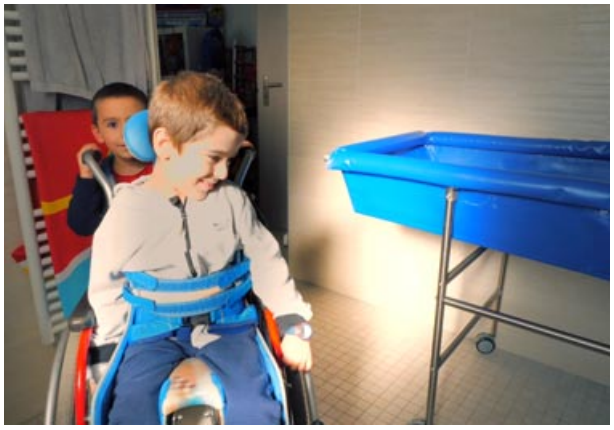
Supporting and helping families

Being confronted with leukodystrophy, a serious, progressive and disabling orphan genetic disease, is life-changing. ELA provides families with information that will help them to understand the disease, its evolution, and the help available. Its teams are at the service of families to support them and respond to their needs as best as possible according to their personal situation. The association provides administrative, social, psychological, material and financial support.

Every year, the ELA France association organises a weekend that brings families together with the aim of offering sick children and their loved ones a special moment.

Some examples of activities:

- Helping families to find personalised solutions according to their needs.
- Relieving families of daily difficulties (adaptation of housing, vehicles, purchase of equipment, organisation of adapted leisure activities, etc.).
- Financing therapeutic treatments and comfort products that are not covered.
- Acting as a liaison with various bodies for procedures related to illness and disability.
- Organising moments of exchange, respite and relaxation allowing children and families to escape from the daily grind of the disease (a family weekend is organised every year).
- Helping families to improve their child's well-being (comfort, stimulation, relief, etc.).
- Providing rigorous, up-to-date and appropriate information on leukodystrophies and their therapeutic approaches through exchanges with researchers
- Supporting families in all their applications for assistance.
- Participating financially in adaptation, acquisition and paramedical care projects related to the pathology.



Supporting medical research

ELA supports and encourages research into leukodystrophies. The aim is to provide French and international researchers with the means to act, to better understand the mechanisms of the disease and its evolution, in particular through the use of disease assessment tools and biomarkers. For their part, clinicians carry out clinical trials to improve care, patient comfort, relieve pain and prolong life.

Medical research on leukodystrophies

Each year, through the publication of a call for proposals, ELA invites the international scientific community to submit innovative research projects in the field of leukodystrophies and myelin repair; a way to promote a better knowledge of leukodystrophies worldwide and the development of promising therapies. ELA's scientific council, composed of 15 members of 7 different nationalities, evaluates the research projects with the help of external experts and selects those that will receive funding.

To meet the expectations of patients and their families, ELA is accelerating medical research by supporting numerous projects that lead to a better understanding of the biological mechanisms involved in leukodystrophies, the identification of the many genes responsible for the disease, gene therapy trials on certain forms of leukodystrophies and pharmaceutical trials.

The Leuconnect platform

ELA has launched an Internet platform to support clinical research: Leuconnect. Designed for patients and their relatives, it allows the recruitment of a large number of patients in epidemiological studies or the implementation of clinical trials, so that these diseases are better understood and patients better managed.

The ELA families/researchers meeting

In order to provide information, ELA organises an annual colloquium dedicated to patients and their families. On this occasion, leukodystrophy specialists report to families on scientific progress in the field and answer their questions. It represents a unique opportunity for exchange between researchers and patients.

ELA brings together the best specialists from all over the world and focuses on leukodystrophies and myelin repair.

Our missions

Raising awareness of all audiences

Many events punctuate the life of the association. They raise awareness of leukodystrophies among the general public and contribute to the collection of donations for the fight against these diseases. Raising awareness through information, communication, media coverage and the organisation of events are the means by which ELA raises the funds needed to carry out its missions. ELA also initiates its own events such as the "Mets tes baskets" ("Wear your trainers") operation at school, at work and with the general public.

Wear your trainers at school

Since 1994, ELA has been running a campaign in schools called "Wear your trainers and beat disease", which starts with the ELA creed.

This action, which was approved by the Ministry of National Education, Youth and Sports in 2020, is a means of awakening citizenship and solidarity. Every year, more than 500,000 students put on their running shoes and symbolically lend their legs to ELA children who can no longer use them.

Wear your trainers at work

Since 2011, ELA has been offering the corporate world a day of solidarity and conviviality that unites employees around a cause, while making them aware of their health capital. This activity is easy to organise and takes place during a working day. The principle is simple: 1 step = 1 euro cent (or free donation) donated by the company to ELA.

The ELA association organises annual fundraising events.

Developing action at international level

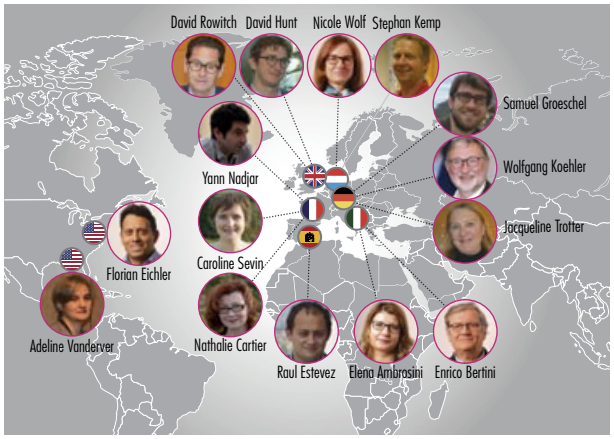
Supporting research in the field of leukodystrophies, in France and internationally, is one of ELA's priorities. To this end, ELA promotes exchanges between the different actors of the international medical and scientific world in order to accelerate research and move towards new therapeutic paths.

Accelerating the development of research

Since 1992, ELA has been working with its counterparts in various European countries and the United States and has promoted the creation of national branches. ELA now exists in Germany, Belgium, Spain, France, Italy, Japan, Luxembourg, the Indian Ocean and Switzerland.

Over time, the need to shed a light on leukodystrophies by accelerating research to an international level became crucial. In 2015, the creation of ELA International was a concrete expression of this major challenge: to increase ELA's activities throughout the world. ELA organises international scientific conferences bringing together the leading specialists in leukodystrophies.

Each year, these events provide an opportunity to review the latest discoveries in the field and to promote exchanges and collaborations between researchers and clinicians.



Launching calls for proposals

Each year, ELA publishes a call for proposals to the international scientific community.
For example, the themes selected for the 2019 call for tenders were:

- Clinical tria / clinical trial preparation, including development of long-term outcome indicators, biomarker characterisation, imaging protocols, pharmacodynamic / pharmacokinetic studies.
- Pre-clinical studies specifically testing therapies (gene, cell, enzyme or pharmacological therapies).
- Development of cell models (e.g. patient-derived iPSCs).
- Study of the mechanisms responsible for the disease, in order to identify new therapeutic approaches.

The members of the ELA Scientific Council meet twice a year, in March on the occasion of the family/researchers meeting and in October. The applications are evaluated and the projects to be funded and the renewal agreements selected.

In 2021

25 new research projects were evaluated, 5 projects were selected (i.e. 20% of the applications, compared to 35.3% in 2020). The overall budget allocated to research amounted to EUR 1,105,557:

- 5 new projects for an amount of EUR 356,546
- 7 renewal projects for an amount of EUR 627,840
- 1 exceptional clinical project for an amount of EUR 121,171

What are our projects to support families and medical research?

Family support project

Multi-sensory spaces

Objectives	• Stimulate the development of the ill person. • Maintain autonomy, attention, memory. • Promote well-being.
Budget	**EUR 12,000**
Duration	-
Description	Each form of leukodystrophy is affected differently, and so is the resulting disability. Gradually, the affected person, whether a child, adolescent or adult, may lose all or part of their independence and see their ability to move, see, hear, communicate or eat diminish. Specially designed to be physically and cognitively accessible, generating positive emotions and pleasant sensations, the multi-sensory room stimulates all the senses: hearing, smell, sight, taste and touch. It is composed of different materials (vibrating cushions, water mattresses, vibrating platform, bubble column, projectors, optical fibres, etc.) which will enable the patient, accompanied by a paramedical professional and/or one of his or her carers, to stimulate his or her limbs and recover sensations which have sometimes been lost.
Impacts	• For the user: Physical and psychological relaxation / Feeling of calmness allowing attention to stimuli / Reduction of anxiety / Reduction of invasive disorders / Improvement of communication (verbal or non-verbal, relationship with others and interactions). • Pour ELA: Based on its belief in the benefits of this type of equipment, ELA enables particularly isolated patients, who can no longer be cared for in a centre, to benefit from such spaces in their own homes, by equipping their homes with the necessary equipment.

Family testimony video - "Des efoiles dans les yeux de Loris" ("Stars in Loris' eyes")
https://youtu.be/XFfmvqpDPs0

Family support project

Respite weekend for ELA families

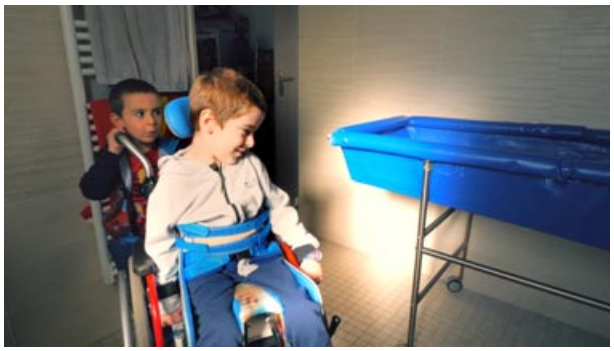
Objectives	Bringing together the families of ELA for a weekend to allow them to get together, relax, renew ties, come out of isolation and recharge their batteries to face the year of facing the disease on a daily basis.
Budget	**Total cost of the project: EUR 245,000 (accommodation / catering / activities, etc.)** **Average cost per family: EUR 1500**
Duration	3 days in August
Description	Living with a person affected by leukodystrophy on a daily basis means taking care of them 24 hours a day. Those close to the affected person are caregivers (parents, brothers, sisters, spouses, etc.). They provide daily physical and moral support, which tires them and can lead to a form of isolation. During a weekend, families benefit from a pleasant setting, leisure facilities adapted for their children, and activities that allow them to share moments of well-being and conviviality. Dedicated activities, workshops and entertainment are organised to help children and families relax. They also benefit from information meetings and discussion groups.
Impacts	180 ELA families, i.e. 900 people, take part in the respite weekend for moments of happiness and escape for children and families whose lives have been deeply affected by the disease, and for meaningful opportunities for exchange for the families, for meeting and mutual support between families.

"Les familles à Center Parcs" ("Families at Center Parcs") video
https://youtu.be/i59oRTroYJg

Family support project

Direct aid to families

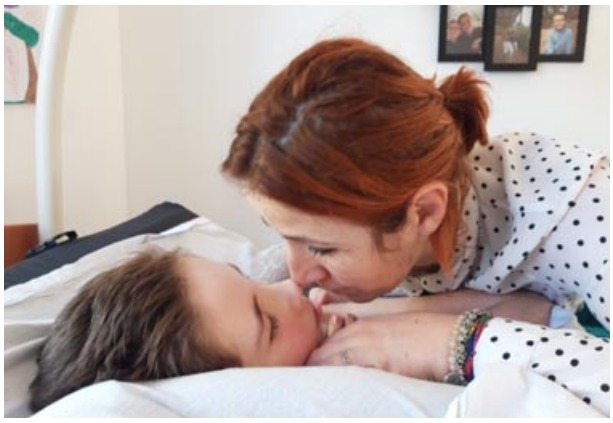
Objectives	• Participating in the acquisition of technical aids, the adaptation of living spaces or vehicles necessary to compensate for the disability. • Relieving the daily burden of families with tailor-made facilities.
Budget	**The investment for a family varies from EUR 5000 to 15,000.**
Duration	-
Description	ELA offers families administrative, psychological, technical and financial support. Without taking the place of public funders, ELA helps families to build financing plans and co-finances the part of the projects for which they remain responsible for financing: • manual or electric wheelchairs, • home improvements (widened hallway, stair lift, access ramp, adapted bathroom, etc.), • vehicle adaptation, transport assistance, • support in the event of death, • costs of accompanying persons during a hospital stay away from home.
Impacts	After administrative support and preparation of the file, ELA makes up the difference and thus improves the living conditions of families and sick children. Each year, ELA devotes around EUR 200,000 to direct aid to families.



Research support project

Pre-clinical study for patients with adrenoleukodystrophy

Objectives	Delaying, improving or even preventing brain inflammation in CALD patients.
Budget	**EUR 66,315**
Duration	1 year
Description	This research project concerns X-linked adrenoleukodystrophy (X-ALD) which is the most common leukodystrophy. The majority of patients develop fatal inflammatory cerebral demyelination, known as cerebral ALD. Haematopoietic stem cell transplantation is the only intervention available for CALD patients with gene therapy. The project partners: Dr. Johannes Berger - Centre for Brain Research, Medical University of Vienna, Austria
Impacts	This study will allow the evaluation of the anti-inflammatory potential of known drugs for ALD patients and will open the way to treatment efficacy trials.



Research support project

Clinical trial on CACH syndrome

Objectives	Studying the effect of guanabenz, an anti-oxidant molecule, on children with CACH syndrome.
Budget	**EUR 2 million, of which EUR 500,000 is financed by ELA**
Duration	4 years
Description	Analysis of the safety and efficacy of guanabenz in delaying or halting disease progression in children with early CACH syndrome. The trial plans to include 40 patients under 10 years of age. The project partners: Dr Marjo van der Knaap, MD. PhD. Neuropaediatrician. Professor of Paediatric Neurology at the University of Amsterdam VUMC - Amsterdam, the Netherlands.
Impacts	To benefit all children suffering from an early form of the disease. Expected results range from improved quality of life for patients to the ability to regain mobility. This is the first proposed treatment trial for these patients.

"Un essai clinique prometteur sur le syndrome CACH" ("A promising clinical trial on CACH syndrome") video
https://youtu.be/uF2x4Nk_1rQ

Research support project

Substrate reduction therapy for Krabbe disease

Objectives	This work should evaluate a candidate therapeutic approach for possible development in children.
Budget	**EUR 200,000**
Duration	2 years
Description	Krabbe disease is a very rare neurodegenerative disease in which the GALC enzyme does not function. No cure is currently available, although the disease is often fatal in the first 2 years of life. The project will observe the effect of a new small molecule designed to reduce psychosine that accumulates to high levels in patients' brains. The project partners: Dr. Ernesto Bongarzone,University of Illinois, Chicago, USA.
Impacts	It is hypothesised that the treatment will improve psychosine metabolism and reduce disease burden and neurological impairment.

Research support project

Understanding the new choline transport leukodystrophy

Objectives	This project aims to develop innovative therapeutic solutions.
Budget	**EUR 165,000**
Duration	2 years
Description	Impaired choline transport leads to a newly identified leukoencephalopathy for which there is no cure. The project aims to characterise a model of the disease and to unravel the pathology and pathological mechanisms of this leukodystrophy. The project partners: Pedro Brites, Institute of Health Research and Innovation, University of Porto, Porto, Portugal.
Impacts	The aim of the project is to determine how the deregulation of choline transport affects brain development.

Some testimonies from ELA families

Francis Subtil, Léonie's father, tells us about ELA's support for families

❝In 2019, we moved as Leonie's needs changed according to her illness. We needed new facilities and equipment. The ELA team helped us with the administrative procedures, provided us with support in the preparation of aid files to benefit from adapted equipment and supported us financially because the equipment for vehicles or the house is very expensive.❞

Crystelle Cottart, Ewen's mother, relies on medical research

❝I have high hopes for the research. I hope there will be a study and perhaps ultimately a treatment, something that could stop the evolution of the disease in mothers with leukodystrophy. They need so much to be able to continue the fight against their child's illness. It is essential, it is necessary❞

Gaëlle, mother of Benjamin, a boy affected by leukodystrophy, feels less alone thanks to ELA

❝We have been part of the association since 2012, following the diagnosis of our son Benjamin's disease. ELA provides us with daily support for MDPH files, particularly in terms of administration. It's nice not to feel alone when preparing the aid files. ELA is a great help to us!❞

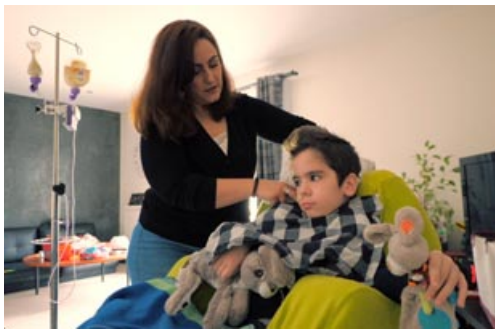
Karine, mother of Margaux, and her father, are fighting a constant battle

❝Margaux is taken to the IME every day by taxi. She receives appropriate care and activities. These are moments of contact and interaction that are important to her and provide her with relief on a daily basis. Her leukodystrophy is a daily struggle for us, and a constant fight against the progression of the disease. What is important for us is to enjoy as much as possible all these moments shared with our two daughters. So we move around whenever we can: with our family in a camper van (to have everything at hand), on "respite weekends" at Center Parcs with ELA, and by the sea in the summer. In short, we make memories.❞

Joris was able to benefit from a clinical trial

❝Joris was a very healthy little boy until he was 12 months old, never sick, very alert. Then we observed a slowing down in the developments he was supposed to acquire: he was in no hurry to start walking and talking. At the age of two, he was diagnosed with metachromatic leukodystrophy. Luckily, Joris met all the criteria for inclusion in a clinical trial, which he has been following for seven years, so he has a treatment that he tolerates well, and which allows him to continue to evolve, slowly.❞

Overnight, Noah and Nathan stopped walking



❝The ELA association helps us on a daily basis, through psychological support, meetings between parents and financial and administrative assistance.

The house had to be rearranged, walls had to be knocked down to make it easier to move around with wheelchairs, a bathtub had to be installed in the bathroom... There is no cure for Nathan and Noah yet. So we have to give them the best life possible.❞

The Association adapts to our needs

❝We needed information about this rare disease, the association provided it. It was very supportive from the beginning. Its strength is that it adapts to our needs in a personalised way, as the difficulties differ greatly from one young person to another. Improving daily life also means getting in touch with other families affected by the disease and sharing discussions, activities, laughter and sometimes tears.

The respite weekend each year at Center Parcs, which brings together an average of more than 900 members, is a highlight for the whole family, and particularly for Julien, his older brother: "We look after Alexis together, as a family - it means a lot.❞

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