AEMO

ASSOCIATION CHILDHOOD
& ORPHAN DISEASES

2023 brochure

A word from the Vice President

After several years spent as President of the Association Childhood and Orphan Diseases, I would like first of all to express my deepest thanks to all the donors who support the cause we defend on a daily basis. Your generosity and your loyalty to our Association represent an immense help without which nothing would be possible. A big thank you to you who allow us to work for the well-being of children and families and to give us the means to continue our fight against our struggle with this serious public health challenge.

Rare diseases confront affected persons and their families with numerous challenges that affect all areas of life because they are often chronic and disabling. They generate a strong social disadvantage because in addition to the direct costs, these diseases are also the source of indirect costs. Living with a rare disease means having to deal with a marked-out path: the obstacle course.

The heterogeneity and strangeness of these diseases lead all patients to the same conclusion: a rare disease means finding oneself "outside the boxes" of medical, scientific, social and administrative institutions. Living with a rare disease means facing, with perseverance, challenges and difficulties in all areas of life. Courage to them!

Before concluding, I would like to thank all the members of the committee for their involvement, as well as for the confidence they have shown. My gratitude also goes to all the people who work within the Association to accomplish their arduous task of informing the public of the daily struggle that families go through and to carry out our annual campaign to collect donations.

It is to make this journey as smooth as possible for the patients and their families that our Association works day after day.

Olivier Meyer, Vice President

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« Wherever there is poison, nature intended us to find the counter-poison: where the disease is, we can find the cure. » Alexandre Dumas

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What is AEMO?

Our Association was created in 2004 with the aim of providing broad information on the problems generated by rare and orphan diseases.

Even though rare diseases have their own specificities, they cause the same suffering.

The Association "Enfance et Maladies Orphelines" is the only one in Switzerland that takes into account all the rare or orphan diseases listed by the WHO.

Our statutes (available on our website) stipulate that we raise funds to support any initiative or action related to the field of rare or orphan diseases, i.e.:

- To help children and young adults in difficulty, domiciled in Switzerland, whatever their ethnicity and their religion
- Helping other associations with similar goals
- Supporting research and information

We work with a team trained to inform the general public about the consequences of rare diseases, to make them aware of the problems that arise from being affected by such a disease and to collect donations. **It is important to know that without the donor, nothing would be possible.**

In parallel to the work of information and collection of donations, we manage the files of the families that we support financially. For this purpose, we have an ethics committee that visits the families wherever they are in Switzerland in order to identify as closely as possible the specific needs of the child and to define the family's situation, both moral and financial. We support the families either by a **monthly aid** unlimited in time or by **punctual aids** of an often rather important amount. Once a year, we also make an additional payment to the neediest families to help them through the end-of-year celebrations. All our grants are non-refundable.

The ethics committee obviously follows the files very closely in order to follow the evolution of the family and adapt the financial aid if necessary.

Our assistance is intended to lighten the family's overall budget by taking charge of expenses or care that are not covered by insurance companies which, on their own, sometimes provide insufficient benefits, such as dental expenses, institute fees, the purchase of therapeutic equipment, travel expenses, etc. We also pay for leisure activities such as dancing, swimming and horseback riding, which are often considered as a breath of fresh air for children. **We position ourselves as a relay for the insurance companies**. This is why these patients need external financial support like the one we provide.

The families that we support have announced themselves either by contacting the association spontaneously, or through other associations that pass on our contact details, or during our fundraising appeals during which the families contacted announce themselves as parents of a child affected by a rare disease.

Arrival of the Tuffière challenge race on September 5.

The AEMO team

Management Committee

Bhira Meyer
President

Olivier Meyer
Vice President

Francesca Seegy
Member

Silvana Gonzalez
Member

Jacques Guhl
Member

Dre Loredana D'Amato Sizonenko
Member, Orphanet Switzerland coordinator

Ethics Committee

Sylviane Moreillon-Puhovic

Manon Weibel Rosello

What is a rare or orphan disease?

Even if the diseases are rare, the patients are numerous.

It affects less than one person out of 2,000. A disease is said to be orphan when there is no effective treatment. These diseases require multidisciplinary efforts, as all medical specialties are involved.

These diseases can be of various origins: metabolic, neuromuscular, infectious. **In 80% of cases, they are of genetic origin** and express an error in the writing of a gene. Rare diseases have a great variability of expression in the same pathology with different evolutions from one person to another. But generally speaking, they are most often serious, disabling and chronic, affecting physical, intellectual, behavioral or sensory capacities, sometimes with life-threatening consequences.

Today, there are some 7,000 of them and five new pathologies are described every week in the medical literature. Taken as a whole, they affect a significant number of patients.

It is estimated that 30 million people are affected by rare diseases in Europe, i.e., 6 to 8% of the population. By extrapolation, and in the absence of registers, **it is estimated that approximately 500,000 people in Switzerland are affected.**

On the other hand, one in four people know someone with a rare disease. These figures put the notion of rarity into perspective. A rare disease can appear at any age, but in more than half of the cases, the symptoms appear at birth or during childhood.

Estéban, suffering from an undiagnosed disease.

Three examples of orphan diseases

Complete Mitochondrial Disease 5

Mitochondrial disease develops at the level of the mitochondria. Consisting of an outer membrane and an inner membrane formed by ridges that increase its surface, mitochondria are organelles located inside the cell that are approximately one micrometer in length.

They play an essential role in cellular respiration and in the production of energy, which is essential for the functions of each cell in the body. Some people will have benign symptoms, such as difficulties in memorizing or a susceptibility to fatigue. Other people will have much more serious manifestations, and this from birth. The main symptoms are heart problems, epilepsy, deafness, autism, vascular accident or mitochondrial myopathy, etc.

Wolf-Hirschhorn syndrome

Wolf-Hirschhorn syndrome is a chromosomal disorder that combines a characteristic face with drooping corners of the mouth, intrauterine growth retardation followed by postnatal growth retardation, frequent seizures, mental developmental delay, feeding difficulties, deafness and heart defects. Most affected children will never walk or talk.

The prevalence is one in 50,000 births and affects two girls to one boy.

Ehlers-Danlos syndrome

Ehlers-Danlos syndrome is a heterogeneous group of connective tissue diseases, characterized by an abnormality of collagen synthesis and leading to skin hyperextensibility, joint hypermobility, vascular fragility and variable involvement of other viscera.

What are the consequences of the rarity of these diseases?

These diseases have specific needs in terms of financial and moral support.

According to a study, **health expenses for people with rare diseases are 3 to 4 times higher** than for other individuals.

It is important to know **that in addition to the direct medical costs**, these diseases are the source of indirect costs such as the abandonment of professional work (often for the mother), the hours of activities related to the care of the sick person, the adaptation of the home, the cost of transportation to hospitals, the costs related to special food in some cases, sterilization therapeutic material, etc...

These health costs, which are not always covered by official organizations, mean that patients have to cover these additional costs themselves. However, this relatively large part of the costs that is borne by families should be covered on an individual basis. We must also take into account the moral and psychological burden that is added to the various problems of daily life. Faced with these many difficulties, many parents divorce and family life breaks down. Often, the mother has to face alone the expenses related to the disease, and this without having any professional income.

Another cause of suffering for families is the obstacle course to obtain a diagnosis, which is the most important issue for them. The delay in making a diagnosis between the first symptoms and the appropriate diagnosis sometimes leads to unacceptable risks and can be the cause of the transmission of the disease to other future children because of the lack of information given to the couple who remain in the dark.

Putting a name to a disease is a vital necessity for the patient. Knowing what to fight against is the hope of taking charge of the disease, improving treatment and even stabilizing the evolution of the disease. It is the hope of having a life project.

In the life of the child and the family, three distinct periods can be distinguished: life before the announcement, the moment of the announcement and those following the announcement.

The period before the announcement is one of doubt, fear, questioning and waiting. It is a long period of inactivity.

The very moment of the announcement is a moment when life stops and everything changes. It is a real psychological trauma for the family. When the disease is announced, it is also announced that there is no real curative treatment. Patients go through different stages: revolt, withdrawal, guilt, shame, fear.

The period following the announcement is dramatic in all cases. The patient and his or her family know but are not able to understand all the consequences. Moreover, not everything has been said or wanted to be heard.

It is a time of activity made up of a search for information followed by hope and despair
.

"It is often difficult for them to speak, but it is also often difficult for them to remain silent."

Our association has purchased equipment adapted to Lucie to practice skiing

This is why **misdiagnosis** or late diagnosis can lead to consequences such as loss of time, unnecessary, inappropriate or even dangerous treatments.

Because of the involvement of various organs and the consequences on other functions, people with the disease should benefit from a multidisciplinary management. All these factors imply that psychological support should be set up to avoid families in the midst of distress not knowing where to turn, not knowing any infrastructure to advise them or direct them to competent professionals. It should be noted that, in recent years, the National Coordination of Rare Diseases (Kosek) was created. The latter is a coordination platform that aims to improve the care of patients with rare diseases. To this end, Kosek carries out projects in collaboration with health actors while involving the people concerned through their patient organizations.

And all the consequences of these disabilities are a determining factor in social exclusion, especially if they are combined with behavioral problems. The constant presence of a family member is necessary for the patient and, as a result, personal and social life is reduced to its simplest expression.

The patient and his family are never off work, neither during the day nor at night, because the hours of care are added to the daily work. The patients require direct help from a charity that can quickly decide to support them financially, either on a one-time or regular basis.

This is why these patients need financial support.

If our association can provide relief, even temporarily, to children who are stricken by the injustice of an incurable disease, then it will have achieved its objectives.

First Step

The «First Step» therapy initiated by Mr. Shai Silberbush from Tel Aviv allows children with motor or sensory disabilities to significantly improve their well-being by offering them the possibility to acquire greater autonomy in their daily lives.

After having met a mother living in Zurich whose son had been following this therapy for several months with great success, our Association brought Shai Silberbush to Lausanne for a few days in February 2011 in order for him to meet children suffering from one or another rare disease and who could benefit from the First Step method.

Since then, our Association has helped by financing this new therapy which is practiced in Switzerland and in Tel Aviv. Several children of our Association practice this therapy which allows them to significantly improve their motor and sensory behavior and offers them the possibility of acquiring greater autonomy in their daily lives.

For each child, an evaluation is done by the therapist and, during two hours, he observes and tests the child in different situations. Afterwards, the therapist talks to the parents and explains to them **what could be improved and how to improve it**. This assessment session is done for each child in order to judge the opportunity to start this therapy. Several points are evaluated, such as the family's ability to devote several hours a week to practicing the exercises taught, since the parents are involved as partner-therapists. Observing the parents' activities with the child reveals much about the child's dynamic development.

After exploring the child's potential, First Step sets up a **customized work process**. Even in cases of brain damage or different syndromes, the ability to learn exists. Little by little, the child will learn all the steps he has skipped and by receiving the right stimulus, the brain responds and reorganizes the network of its connections.

The First Step program includes integration with the child's entire environment, especially the parents. The therapist not only approaches the child, but also works to modify the parents' behavior to guide the child toward change.

The focus is on the child's potential, not just the disease. First Step does not say "the child does not know" but "the child has not yet learned". It is often necessary to take a step backwards in order to move forward. A child must first learn to crawl, crawl, sit before learning to walk. This method allows the growth age to catch up with the chronological age.

Once the process is in place, the family will need to apply everything they have learned in the intensive treatment phase to everyday life. Follow-up sessions will be implemented by the therapists to continue the treatment according to their lifestyle.

Balance work for Ismael.

Coordination work for Diego.

Regularly, videos will be sent to the therapist who will make the necessary adjustments. To date, we are financing this therapy for 21 families: 8 families in the canton of Vaud, 1 family in the canton of Valais, 1 family in Geneva, 10 families in the German part of Switzerland, 1 family in the canton of Fribourg.

All of the children have made tremendous progress: Many of them have taken their first steps or learned to eat by themselves. Each family that has benefited from this therapy has seen their lives change radically and some teachers in the institutes where these children are placed have been so impressed with the rapid results obtained that they are now applying the exercises of this method.

Many families have shown their gratitude through small messages. Here are a few excerpts:

Alicia, who has Sturge Weber syndrome, started walking without assistance five months after the beginning of the therapy. Today she is an independent little girl who speaks, writes and goes to a regular school twice a week.

“Until we learned about First Step, all the therapists told us everything Alicia would be unable to do. With First Step, we heard everything she could do. This therapy has been a sea change that has led us to these wonderful results”.
Parents of Alicia born in May 2007 in Lausanne.

Mila was born with “Flat Brain” syndrome, a very rare syndrome. After two weeks of an intensive process, Mila started crawling on her belly and today she walks with support and communicates better with her environment.

“It has been and still is a fundamental experience for the development of Mila. This journey is not yet finished but it has allowed us to find ourselves where we are today”.
Parents of Mila born in August 2007 in Lugano.

Loane was born with V-deficiency, a rare genetic disease. At the age of five and a half, she could not walk or talk and could only pronounce the word “no”. Today Loane walks almost freely. Even when she falls, she knows how to get up. Her progress surprises even the doctors and teachers at the institute.

“Before the therapy, Loane was like a shell. In Tel Aviv, the shell broke and Loane became open to the world around her. She is cheerful and happy in life”.
Parents of Loane born in August 2005 in Lausanne.

In view of the progress made, we have decided to continue financing this therapy in order to bring as much improvement as possible to the children. It is true that this represents an important cost (about CHF 9,000.- for a child) since we finance not only the therapy, but also the travel for the family, the rent of an apartment, a translator when the parents do not speak English.

Motor skills work for Estéban.

Excerpts from testimonials

We have selected a few extracts of testimonies sent by the families we support. We can't publish them in their entirety, due to lack of space, but all the words received, the cards and the drawings of the children have touched us enormously by their spontaneity and their kindness.

A young man with cardiomyopathy

“ Thank you for your help, every month I can continue my school despite my health problems, thank you all ”

Mother of a child with a rare disease with autistic disorders

“ When we met for the 1re time the AEMO association, 5 years ago, our son was 3 years old. He was not sitting up, crawling or moving around. He only ate blended food and categorically refused pieces. A lot of outside stimulation seemed to assault him and he stayed locked in his little bubble.

In March 2017 we started, with the support of AEMO, First Step therapy. Since 2017 we went as a family 2 times to Tel Aviv and the Israeli therapists came 5 times to our house. We have regular contact via Skype.

Our son has made great progress since then. He has learned to crawl, then to crawl on his hands and knees, and now he is practicing climbing stairs and walking. He is communicating with the outside world better and better and seems more comfortable. Even though food is still a complicated area, he is able to eat on his own and is getting the hang of chewing and chunking. What is even more wonderful is that he is playing and interacting with his little sister and that is a wonderful gift. What the doctors refused to give us, AEMO offered us: hope. Hope to see him progress and above all, proof that every child, despite his difficulties, has abilities and that by stimulating him in the right way he can break out of his shell and move forward in this world.

All this is certainly a team effort. The families, the therapists, AEMO but also you, each person who, in one way or another supports us and the other families, by walking, by offering your time, or donations. Without you all this would not be possible. We would like to thank you from the bottom of our hearts! Thank you for believing in him! ”

Parents of a young person with Retinoblastoma

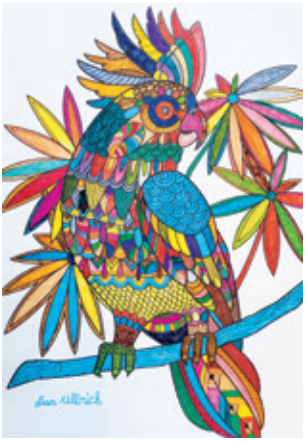
“ Dear Members of the Association, this short note is to convey to you all our gratitude and thanks for your precious financial help.

Raising a child with a disease like cancer is a daily struggle. We go through difficult times and yet, we must move forward. There are brothers and sisters and we have to go on, even if we sometimes feel alone and often misunderstood.

Thanks to your monthly support, we have been able to continue the therapy with the horse and our son follows with more and more assiduity the courses in a heated pool. These efforts bring him joy and help him to develop in life. ”

Drawings

Some drawings of sick children that AEMO supports. Thank you, dear kids!



OUR ASSISTANCE/SERVICES

Helping families

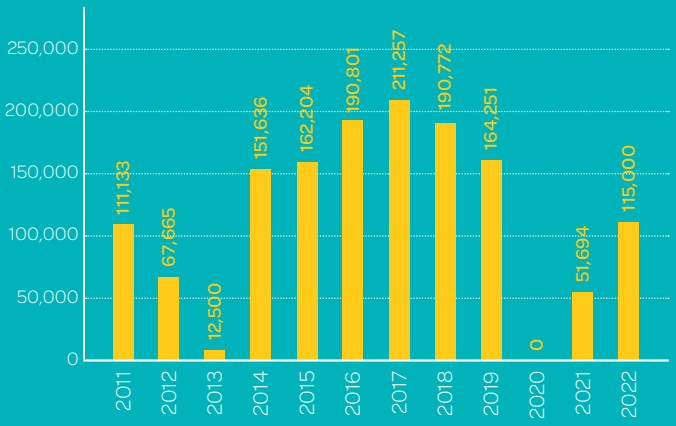
Each year, AEMO organizes a fundraising campaign to provide direct assistance to families living in Switzerland.

> ***Currently, AEMO supports 130 families, 120 of which are in French-speaking Switzerland; it assumes responsibility for costs and care not covered by insurance.***

Funding therapies

Therapies like First Step help families improve the daily life of their sick children. With the help of these therapies, these patients progress in their gestures, their language, their behaviors, their autonomy... the family regains a little color and hope.

First Step grants (in CHF)



The small amounts allocated to First Step in 2020 and 2021 are directly related to the health crisis (inability to assemble, cancelled therapies, etc.).

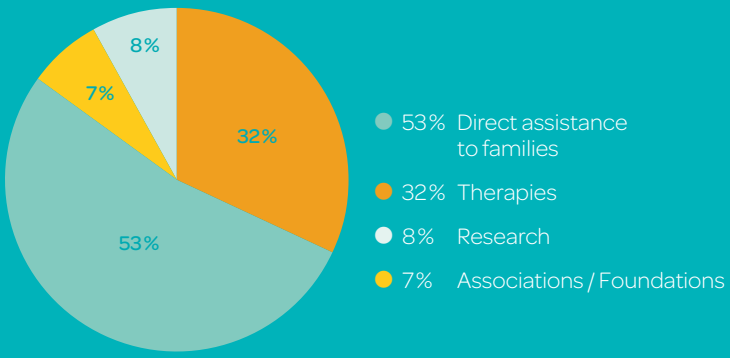
Supporting hospitals, researchers, associations or dedicated foundations

AEMO financially supports specific parts of research programs that concern rare or orphan diseases as well as the activities of institutions such as Orphanet.

Inform and educate

To provide the general public with information to make them aware of the difficulties encountered by families and the possibilities of help available to them. Concretely, we contact each year by phone nearly 30,000 people and we distribute a detailed brochure with 15,000 copies.

Distribution of aid for 2022



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