

AFM Téléthon

France



www.afm-telethon.fr



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1. About

The French Muscular Dystrophy Association (AFM-Telethon) is an association of activists, patients and parents of patients, affected by rare, progressive and significantly disabling genetic diseases, known as genetic neuromuscular diseases. It was created in 1958 out of a conviction and a desire to cure diseases that were long considered to be incurable.

AFM-Telethon is also the Telethon, a fundraising event which brought rare diseases out of scientific and medical obscurity and triggered a three-fold revolution: genetic, social, and medical.

“CURE Neuromuscular diseases, take CARE of patients and their family, Communicate broadly”

2. Mission

Committed to scientific research as well as to supporting patients and their families, AFM-Telethon acts according three main objectives: Cure, Care, and Communicate.

- Curing neuromuscular diseases is the Association’s primary mission with a strategy of innovation and public interest to benefit all rare diseases as well as the most common diseases.
- Reducing disabilities caused by disease means helping patients and their families to live their lives while awaiting treatments.
- The Communicate aims to improve knowledge of rare diseases, long ignored and even misunderstood, to share and explain progress in research as well as therapeutic progress.

3. Focus

Telethon: On the first weekend of December each year, there is a unique momentum that draws thousands of volunteers and partners who organize events for the Telethon all over France, including overseas territories. Almost 20,000 events are organized each year throughout France and abroad, and close to 90 partners take part in the Telethon.

Généthon, a unique scientific and human adventure, turns 30 this year. Funded by the Téléthon, this laboratory has put France at the forefront of genome exploration and of the gene therapy revolution. In 1993, P Melki performs to identify the SMN gene, and in 2012 D M Barkats demonstrates efficacy of SMA Gene therapy on mouse.

4. Coming Up

France has a lot of difficulties to implement newborn screening for SMA. Two pilots are currently on-going, but only 20 of babies are screened. The discussion with the national authorities are very challenging.

Social care in France is very supportive regarding professional personal assistant for disabled people, for example 24h/day can be reimbursed by national insurance. Unfortunately, there's a lack of trained workers, so it's more and more challenging for patients to live independently.

