Francois Lamy – VP, AFM Téléthon

We represent more than merely a patient organisation – we operate alongside pharma, and we possess our own voice that demands attention

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Francois LAMY, Vice President of AFM-Téléthon and father of a child with Duchenne muscular dystrophy, shares how the organisation grew from a family-led initiative into a major force in rare disease research and advocacy. He highlights AFM's role in launching Genethon and advancing gene therapy, while criticising the lack of ambition in France's current rare disease plan in terms of drug development and innovation. Emphasising patient-led governance, sustainable innovation, and equitable access, He stresses AFM's long-term vision and belief that today's rare disease breakthroughs will shape tomorrow's medicine.

Could you begin by providing our international audience with an introduction to AFM-Téléthon today, and perhaps touch upon the historical context that shaped the organisation?

Before delving into our history, I believe it is essential to establish the foundation of our governance structure. I serve as Vice President of AFM-Téléthon, having held this position since 2017. What distinguishes our organisation is that all board members are either patients themselves or family members of patients. In my case, my son is affected by Duchenne muscular dystrophy, which forms the basis of my commitment to this organization.

The disease affecting my son represents my curriculum vitae in this domain – it is my sole credential and yet my most profound competency. It embodies our driving force: the relentless

battle against disease and the daily progression of illness that I witness in my own child. This reality is shared by every board member at AFM. Consequently, all strategic decisions, all investment priorities, and all resource allocation choices are driven by families directly affected by progressive, devastating, and in most cases, fatal diseases. This patient-centric governance has remained our cornerstone since our inception in 1958.

The association was established as a modest organisation led by a handful of families in France during an era when rare diseases were neither recognised nor understood as a distinct category. The disease affecting my son was virtually unknown; only one physician in the entire country possessed any substantive knowledge about this condition, and all affected families found their way to him. This situation persisted for two decades before meaningful change began to emerge in the early 1980s, as AFM expanded and families coalesced around a shared purpose.

During this period, families united to collect funds and finance research aimed at understanding these diseases and developing technical solutions to support patients. A pivotal moment arrived in 1986 during a scientific conference organised by AFM in Tours. At this gathering, a young American researcher presented ground-breaking work: the discovery of the gene responsible for Duchenne muscular dystrophy. This revelation proved transformative, as it defined precisely what we were confronting. The families, advised by expert scientific counsel, recognised that genetics – though not yet established as a formal scientific discipline – represented the most promising avenue for research investment.

This understanding necessitated substantial capital deployment – not modest sums, but millions of euros – merely to comprehend the underlying mechanisms of these genetic diseases. Consequently, in 1986, the association conceived the idea of organising a major fundraising initiative, which materialised as the Téléthon in 1987. The inaugural event exceeded all expectations, generating remarkable public response and substantial funds. The President at that time made a commitment to donors: complete transparency regarding fund utilisation and a strategic focus on genetic research. This commitment led directly to the establishment of Genethon, our research laboratory, born from the success of that first Téléthon.

Nearly four decades later, whilst much has evolved, the Téléthon remains a vital annual event that unites the nation. We collect approximately 90 million euros annually through pure public generosity. These funds support extensive research portfolios and provide direct assistance to families through more than 250 social workers who deliver daily support and enable affected families to pursue fulfilling lives despite their challenges.

France has a well-structured rare disease system, with strong patient groups and a unique reference centre network. In your view, what are its key strengths, and where is there room to improve?

To be direct: most elements of the rare disease ecosystem in France were initiated by AFM. From the very first Téléthon, a fundamental principle has guided our approach: the recognition that fragmentation serves no one. We understood that if each rare disease created its own organisation and collected modest sums independently, we would accomplish nothing beyond communication expenses. Unity was essential.

Whilst AFM originated from families affected by Duchenne muscular dystrophy – which remains symbolically significant to our history – the organisation is fundamentally a neuromuscular disease association encompassing several hundred rare conditions. These diseases present diverse clinical manifestations: whilst many affect young children, others manifest in adulthood, presenting entirely different challenges when individuals already have established families and careers. Despite these variations, our core principle persists: we must work in unified direction.

Numerous rare disease associations exist in France, but AFM established the Rare Disease Alliance in Paris – an umbrella organisation that coordinates all individual rare disease associations nationally. This structure, whilst independent from AFM, receives substantial funding from us. The Alliance enables smaller organisations to access resources whilst providing a collective voice for advocacy with public authorities. Two hundred associations possess no influence if they speak disparately; united, they become formidable.

Our independence principle is paramount. We maintain one board member within the Alliance, but we do not hold the presidency. They possess their own voice, though typically we work towards shared objectives.

This collaborative model extended beyond France. Two decades ago, we recognised the importance of European health policy. However, as a national organisation, we believed European advocacy was not our direct mandate. Consequently, we created EURORDIS, which now actively advocates for rare diseases at the European level.

Regarding the expert centres: in the 1990s, we recognised that our core neuromuscular diseases, whilst originating in muscle tissue, affect the entire body. The heart is a muscle requiring cardiologic expertise; the diaphragm affects respiration, necessitating respiratory specialists;

digestive function involves muscular activity. A single muscle disease demands coordination amongst six to ten specialists for proper management.

We advocated for multidisciplinary consultations – enabling patients to access multiple specialists in the same location, ideally on the same day. This advocacy contributed to France's first National Rare Disease Plan, launched in the early 2000s. A primary achievement of this initial plan was establishing the healthcare networks – 23 different disease groups for rare diseases in France, each supported by a network of expert centres.

We championed the creation of this established network, which now functions effectively. When an expert in one region has questions, the network enables collaboration. This French model subsequently influenced the creation of European Reference Networks (ERNs). France developed 23 national networks; Europe established 24 ERNs. The model originated nationally in France, expanded to Europe, and now drives efforts to replicate similar structures across all European nations. Italy, for example, has developed its own variant adapted to its less centralised structure.

You played a key role in launching the National Rare Disease Plan, now in its fourth phase. With 95% of rare diseases still lacking treatment, do you believe the current plan is ambitious enough to drive real progress?

We remain closely involved in preparing each plan, even if we are not the final decision-makers. Working with the Rare Disease Alliance, our view is that the current plan lacks ambition. It largely repeats the previous plan without introducing real innovation. The continued focus on diagnostics, genetic platforms and high-throughput sequencing simply highlights that we have not yet achieved the goals set in the earlier plan and must keep pursuing them.

Our main concern is the insufficient focus on therapies and innovative treatments. This is a major omission, particularly for ultra-rare diseases. We pushed hard for stronger commitments in this area, but the final plan did not go far enough.

Take Duchenne muscular dystrophy as an example. It is still a rare disease, and developing treatments requires massive investment. Even when companies succeed, the resulting medicines are often extremely expensive, which creates major access challenges. But at least a pathway exists: the science is active, several companies are engaged and therapies do reach patients, even if imperfectly.

The real problem lies with ultra-rare diseases, where only a handful of patients exist in a country like France. For those conditions, no viable economic model exists. We have even seen cases where a company secured regulatory approval in Europe, yet could not obtain reimbursement in enough countries to sustain commercial supply. As a result, despite the product's approval, European patients still cannot access it. From a patient's perspective, that situation is simply unacceptable.

What solutions do you envision? What innovation programmes might address this model failure?

For ultra-rare diseases, we advocate forcefully for public intervention – specifically, a national fund dedicated to financing compassionate use of treatments. With such limited patient populations, demonstrating efficacy and safety through conventional clinical trials becomes impossible before treatment administration. Insufficient patient numbers preclude proper clinical trials capable of demonstrating efficacy through regulatory pathways.

We advocate for dedicated funding enabling academic laboratories or small biotechnology companies to bring treatments to patients, even when commercial viability is mathematically impossible. This must constitute a collective intervention – companies cannot provide treatments indefinitely without compensation, and academic laboratories lack funds to deliver treatments to patients independently.

This situation applies specifically to ultra-rare diseases. However, we have also created another structure: a start-up incubator called Ampleïa, designed to assist rare disease biotechnology companies in organising clinical phases. We recognised that academic laboratories or small biotechs – particularly those founded by researchers who have discovered promising interventions – face substantial difficulties raising capital in the rare disease field. Moreover, researchers typically lack expertise in fundraising and business plan development.

Ampleïaattracts investors by presenting a portfolio of biotechnology companies working in rare disease, assists biotechs in developing business plans and presenting projects to investors, and enables investors to diversify risk by investing across multiple biotechs. This creates an essential interface between researchers possessing promising ideas worthy of clinical development and investors possessing capital but limited knowledge of the narrow, specialised rare disease field.

Whilst not entirely recent, establishing credibility and attracting both investors and identifying promising biotechs requires substantial time. Ampleïamust build confidence whilst also identifying and supporting biotechs by helping them recruit CEOs, CFOs, and other capabilities researchers typically lack. Currently, we work with French, European, and worldwide biotechs.

Europe is often characterised as a ‘valley of death’ for biotechnology, particularly regarding local innovation. What role do you envision for patient-driven organisations in helping local innovation thrive?

The challenge is substantial. I can speak with particular insight about Genethon, which pursues numerous projects and represents our oldest laboratory, established in 1990. Initially, Genethon produced the first human genome maps. This example illustrates an important dynamic: once initial genome maps were decoded – sufficient for basic understanding but far from complete – Genethon scientists advocated continuing this work. Complete genome maps would not be produced for another five to ten years.

However, the families leading our association made a decisive intervention: “We possess sufficient knowledge to work on therapeutics. Whilst we understand the scientific interest in completing this work, our priority is drugs. This genomic work can continue in other public laboratories.” Indeed, Genoscope continues purely genetic research adjacent to Genethon. However, Genethon converted into a drug development laboratory.

From the beginning, AFM-Téléthonfocused on Duchenne muscular dystrophy and other neuromuscular diseases. A principle we deeply value became critical: patients can only succeed together in finding therapies. In the mid-1990s, gene therapy existed as a theoretical concept amongst researchers, but treating the extensive muscle tissue throughout the body seemed beyond reach. Intermediate steps proved necessary.

Consequently, the first gene therapy developments financed by AFM addressed not muscles but autoimmune diseases, leading to Professor Alain Fischer’s breakthrough treatment for bubble babies around 2000 – the world’s first successful gene therapy for autoimmune disease. Muscles remained challenging, so we financed projects on ocular gene therapy. The eye, being a localised organ and immunologically isolated from the body, presented a more manageable target. Moreover, unlike ex vivo gene therapy for autoimmune conditions – where processes occur in laboratories with extensive quality control – ocular therapy required direct in vivo injection, representing a significant advancement.

We subsequently financed hepatic research. The liver, whilst larger and internal, naturally attracts many drugs, making it easier to target than muscle tissue. By approximately 2010, Genethon began serious research on muscle diseases, including spinal muscular atrophy, which led to highly successful gene therapy reaching market in 2019, now owned by Novartis. The foundational pre-clinical discoveries for that treatment were made at Genethon, using patents we deposited.

Given the wide scope of rare diseases and limited resources, how do you prioritise and select areas for funding and investment?

Our selection process operates through two essential stages. First, we conduct scientific assessment by the foremost experts in respective domains – truly top-tier specialists – who provide rigorous project evaluations. Subsequently, the AFM board makes all final decisions regarding project approval. This ensures every project receives highly qualified scientific assessment followed by what I term the ‘patient filter’ – not truly an assessment, but rather ensuring we consistently advance towards patient benefit.

However, we maintain several investment tiers. We fund early-stage and fundamental research through worldwide calls for proposals – we finance high-quality projects in any country with strong scientific ratings. We also fund what we term ‘strategic projects’ – more elaborate initiatives that may include fundamental research but possess clear clinical pathways. We provide extensive funding to several laboratories: Genethon is one; I-Stem focuses on stem cells; and a third, critically important facility is the Institute of Myology at Pitié-Salpêtrière hospital, where research occurs adjacent to clinicians and diagnostic laboratories. They conduct biological research alongside consultations with clinicians, and also develop assessment tools – NMR imaging, strength measurement instruments – for evaluating clinical trials and disease progression.

In addition since 2010, we increasingly invest in biotechnology companies, as we approach closer to actual drug development. We invest in or create biotech companies. Yposkesi provides an excellent example – initially created in 2012 as a Genethon spin-off, it became a fully private biotechnology company in 2016.

Today, Yposkesi produces gene therapy platforms for various companies, correct?

Indeed. Genethon remains an excellent client – which is precisely why we created the facility – but Yposkesi serves other clients as an independent company.

Regarding funding philosophy: what we do not fund is equally important as what we do fund, particularly concerning partnerships. Especially now, with biotechnology ventures approaching market stages – whilst recognising market entry remains challenging for rare diseases – we avoid being solitary or declaring our work complete when projects show promise. We seek fair compensation, not excessive returns, because treatments must reach patients. We firmly believe excessively high pricing prevents patient access, so we advocate for elevated but balanced pricing – reflecting bioproductionand R&D costs, but remainingfair.

We fund collaboratively with partners, such as France's public investment bank (Bpifrance). We created a rare disease investment fund in partnership with them.. Whenever possible, we pursue such partnerships. Genethon also licenses projects occasionally. We consistently advocate for fair end pricing, although this becomes challenging when we no longer control project development. Nevertheless, we monitor this carefully.

What role does big pharma play in rare diseases today? How do you engage with them, and is current collaboration sufficient – or do you expect more?

Current engagement is insufficient – far from sufficient, particularly regarding partnerships with us. When we possess high-quality projects, attracting large pharmaceutical companies proves difficult. And yet, these projects are at the forefront of science and adaptable to more common diseases—e.g., CAR-T Cell—and it is also unfortunate for them not to see the innovative therapies and rare diseases sectors as opportunities.

We have achieved some biotechnology partnerships successfully, but large pharma remains elusive.

Why do you believe this difficulty exists with large pharma? In other markets, such companies typically engage readily with patient-oriented associations.

We represent more than merely a patient organisation – we are not simply patients appreciating pharmaceutical work. We operate alongside pharma, and we possess our own voice that demands attention, because we have accomplished substantial work ourselves. Typically, patient organisations attend meetings but lack true influence, lacking financial strength, expertise, or other leverage. Perhaps this explains it – I cannot provide a definitive answer, but the situation is truly regrettable.

When we created Yposkesi, we recognised it as a sovereignty tool for France – for health independence. When seeking a partner – because neither an association nor the public investment bank should operate such an entity indefinitely – we could not attract a French or European partner. Today, a Korean entity owns the company. We retain a small board presence, but this represents a genuine shame.

Looking ahead to the next three years, what excites you most? What are your primary objectives?

Within the next three years, we have one highly symbolic project. The young man featured in our upcoming Téléthon campaign imagery is affected by Duchenne muscular dystrophy. Genethon currently leads a clinical trial for Duchenne – a micro-dystrophin gene therapy that commenced in 2021. d Phase I and II has completed with exceptionally encouraging results. The pivotal phase of this seamless Phase I/II/III trial design commences in September. We invest tremendous hope in this success. Thus far, Phase II results have proven genuinely encouraging.

Beyond this, Genethon develops numerous other gene therapy products for limb-girdle myopathies and hepatic conditions such as Crigler-Najjar syndrome. These products also demonstrate encouraging preliminary clinical results. Our challenge – Genethon ‘s challenge – is that whilst we have many stimulating, exciting projects, we lack sufficient funds to bring them all to market, particularly given difficulties finding partners, especially large pharmaceutical companies.

Consequently, we walk rather than run – or perhaps we run, but insufficiently quickly. For patients, nothing is ever fast enough. However, as patients, resignation is not an option. We will not abandon any project unless it demonstrably fails to work.

Could you address improvements in gene therapy technology?

What exists now might be characterised – perhaps somewhat crudely – as first-generation gene therapy. However, Genethon works extensively on improving these therapies: enhanced production yields, superior efficacy enabling lower patient doses, reduced immune response, ultimately better efficacy and lower cost. Genethon invests substantially in biotechnology development, which – I recognise researchers may object – I find profoundly relevant even if not seemingly exciting science. The drug functions; it “simply” requires optimisation. Filtration improvements, process simplification – whilst perhaps not intellectually thrilling, these advances will prove decisive for

patients.

These techniques being invented now will make substantial differences. When Yposkesi was created in 2012, it was the world's largest gene therapy production facility, simply because such facilities barely existed. Today, larger facilities exist, but this remains an emerging industry where everything requires creation – all quality processes, all production processes. Since creation from nothing was necessary, and current methods function, vast room for improvement exists. We are convinced this will prove transformative for patients.

Perhaps patients with extremely rare diseases will ultimately benefit, as gene therapy production may become less expensive, rendering it more economically viable for companies.

Do you have any final message for our international industry audience?

Our values – the fight against disease, the emergency driving all families, the commitment to general interest – truly distinguish our strategy. We do not fund research exclusively for our children's diseases or our own conditions. We maintain a genuine long-term vision, and this distinguishes us from other organisations.

Biotechnology companies operate on two-month horizons because they lack treasury – somewhat provocative, but essentially accurate. Pharmaceutical companies work on three-month cycles, dictated by quarterly financial reporting. AFM has maintained identical strategy and vision for four decades: finding cures for patients. When we commenced, virtually nothing existed. We did not know which gene was responsible. Observe where we stand today.

This represents our strength: this stable, sustained vision. We work on rare diseases, but we understand that what we create today constitutes tomorrow's medicine. We are certain that innovations developed for rare diseases will eventually benefit more common conditions. Ten years ago, people dismissed this assertion with laughter. Today, cancers are cured using gene therapy products derived from – inherited from – Alain Fischer's work. I am confident that what we accomplish today for rare diseases will serve more prevalent conditions in the future.

Our governance matters profoundly. Our board comprises exclusively patients and parents of patients, but we are not alone. We benefit from the highest-rated experts – the finest in their fields – who volunteer, assessing several hundred projects annually.

We engage deeply in care, not merely research, because families in their daily existence require immediate support. They cannot await future therapies. Through care, we improve quality of life and extend life itself. Quality of life includes public awareness. In France, if you ask someone about gene therapy, they likely connect it to the Téléthon, because it provides a communication window for gene therapy, innovative therapies, rare diseases, and disability.

We pioneered rare disease advocacy: The National Rare Disease Plan, the unique support model provided by our social workers who stand beside families, our advocacy that modified healthcare autonomy legislation alongside other organisations. We insist on cost transparency and pricing fairness.

A critical issue is new-born screening for treatment access. Treatment must reach patients at the optimal moment. For spinal muscular atrophy, the drug has been market-approved since 2019, but France lacked new-born screening. Consequently, patients developed symptoms, received treatment, but too late. Symptoms were halted, but for some, treatment arrived far too late, and they died despite receiving therapy.

From September first, new-born screening for spinal muscular atrophy became available in France – five years too late. We advocated forcefully for this change in perspective and for anticipating such situations. We promote the value of new-born screening in preventive care. Even when no therapy exists, knowing you are affected by a disease slightly before first symptoms appear enables families to adapt and construct their lives with the disease. We advocate extensively on this matter.

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