AFM-TELETHON PIPELINE
of clinical trials

Clinical trials (ongoing or in preparation)

	DISEASES	TYPE OF THERAPY	PRODUCT	PHASE
NEUROMUSCULAR DISEASES	Spinal muscular atrophy with progressive myoclonic epilepsy (SMA-PME) and Farber disease	GT	AAV-ASAH1	Preclinical
	Duchenne muscular dystrophy	GT	AAV-microdystrophin	Phase I/II/III
	LGMD R1 (calpain)	GT	AAV-CAPN3	Preclinical
	LGMD R2 (dysferlin)	P	Bazedoxifen	Preclinical
	LGMD R3 (α-sarcoglycan)	P	Givinostat	Preclinical
	LGMD R5 (γ-sarcoglycan)	GT	AAV-SGCG	Phase I/II
		P	Givinostat	Preclinical
	LGMD R9 (FKRP)	GT	AAV-FKRP	Phase I/II
	Myotonic dystrophy type 1 (Steinert)	GT	AAV-MBNLΔ	Preclinical
	Type 3 glycogenosis (Cori-Forbes disease)	GT	AAV-GDE	Preclinical
	Charcot-Marie-Tooth disease	P	PXT3003	Phase III*
		P	IFB-088	Phase I completed
	Pompe disease	GT	AAV-GAA	Phase I/II
	Myotubular myopathy	GT	AAV-MTM	Phase I/II*
	Inclusion body myositis	P	Rapamycin (Sirolimus)	Phase III
		P	Ruxolitinib	Phase II* in preparation
	Sarcopenia	P	recGDF5	Preclinical
	Amyotrophic lateral sclerosis	GT	AAV-SOD1	Preclinical
		P	IFB-088	Phase II
		P	Interleukin 2	Phase IIb
OTHER DISEASES	Fanconi anemia	GT	Hematopoietic stem cells + LV-FANCA	Phase II*
	Spinocerebellar ataxia 3 (SCA3)	GT	AOM-skipex10	Preclinical
	Dilated cardiomyopathy	CT	extracellular vesicle-enriched secretome of cardiovascular progenitor cells	Phase I*
	Immune deficiencies – Artemis deficiency	GT	Hematopoietic stem cells + LV-Artemis	Phase I/II
	Immune deficiencies – X-linked severe combined immunodeficiency (X-SCID)	GT	Hematopoietic stem cells + LV-XSCID	Phase I/II*
	Immune deficiencies – Chronic granulomatosis	GT	Hematopoietic stem cells + LV-CGD	Follow-up study (15 years) Phase I/II USA
	Immune deficiencies – Wiskott-Aldrich syndrome	GT	Hematopoietic stem cells + LV-WAS	Follow-up study (15 years)
	Dystrophic epidermolysis bullosa	GT	Genetically modified autologous skin cells	Phase I/II*
	Junctional epidermolysis bullosa	GT	Genetically modified autologous skin cells	Étude pilote*
	Glycogen storage disease type 1a (Von Gierke disease)	GT	AAV-G6PC	Preclinical
	Metachromatic leukodystrophy	GT	ARSA	Preclinical
	Disseminated lupus erythematosus	CT	Mesenchymal stem cells	Phase I/II
	Crigler-Najjar disease	GT	AAV-UGT1A1	Phase III confirmatory
		GT	IDES + AAVUGT1A1	Preclinical
	Retinitis pigmentosa	CT	Embryonic stem cells	Phase I/II
		GT	AAV-RdCVF	Phase I/II
	Multiple sclerosis	CT	Cytotoxic T cells	Phase I
	Phelan-McDermid syndrome (genetic form of autism)	P	Lithium	Phase III*
	Syndrome de Wolfram	P	Valproic acid	Phase II
	Sickle cell skin ulcers	CT	Embryonic stem cells	Preclinical

GT Gene therapy CT Cell therapy P Pharmacology

* Preclinical or previous clinical phases funded by AFM-Telethon

Approved drugs

Cuprior® P
Wilson disease

Firdapse® P
Lambert-Eaton syndrome

Namuscla® P
Myotonic syndromes

Skysona® TG
Adrenoleukodystrophy

Strimvelis® TG
X-SCID

Zynteglo® TG
β-thalassemia

Zolgensma® TG
Spinal muscular atrophy

Off-label drugs

Lumevoq® TG
Leber optic neuropathy

Metformin P
Steinert myotonic dystrophy

Patient databases

The AFM-Telethon supports **research databases** that collect medical data of neuromuscular diseases patients.
In 2023, it created **a health data warehouse**.