

AFM-Téléthon THERAPEUTICS PIPELINE

NEUROMUSCULAR DISEASES

DISEASES	TYPE OF THERAPY	PRODUCT	PHASE
Amyotrophic lateral sclerosis	GT	AAV-SOD1	Preclinical
	P	IFB-088	Phase II completed
Charcot-Marie-Tooth disease	P	IFB-088	Phase I completed
Duchenne muscular dystrophy	GT	AAV-microdystrophin (GNT0004)	Phase III
	GT	Tricyclo-DNA (SQY51)	Phase I/II*
Inclusion body myositis	P	Rapamycin	Phase III*
	P	Ruxolitinib	Phase II
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 (ATA-200)	Phase I/II
	P	Givinostat	Preclinical
LGMD R9 (FKRP)	GT	AAV-FKRP (ATA-100)	Phase I/II
Mitochondrial myopathy m.3243A>G	CT	Autologous mesoangioblasts	Phase II
Myotonic dystrophy type 1 (Steinert)	GT	AAV-MBNLΔ	Preclinical
	GT	Tricyclo -DM1	Preclinical
	P	Metformine	Phase III*
Myotubular myopathy	GT	AAV-MTM	Phase I/II* completed
Pompe disease	GT	AB-1009	Phase I/II*
Spinal muscular atrophy with progressive myoclonic epilepsy (SMA-PME) and Farber disease	GT	AAV-ASAH1	Preclinical
Type 3 glycogenosis (Cori-Forbes disease)	GT	AAV-GDE	Preclinical

Approved drugs

Cuprior® **P**
Wilson disease

Firdapse® **P**
Lambert-Eaton syndrome

Itivisma® **GT** (USA)
Spinal muscular atrophy
(2 years of age and older)

Namuscla® **P**
Myotonic syndromes

Sklysona® **GT** (USA)
Adrenoleukodystrophy

Strimvelis® **GT** (Europe)
ADA-SCID

Waskyra™ **GT**
Wiskott-Aldrich syndrome

Zynteglo® **GT** (USA)
β-thalassemia

Zolgensma® **GT**
Spinal muscular atrophy
(less than 21 kg)

OTHER DISEASES

Crigler-Najjar disease	GT	AAV-UGT1A1	Phase III (confirmatory study)
	GT	IDES + AAVUGT1A1	Phase II
Dilated cardiomyopathy	CT	extracellular vesicle-enriched secretome of cardiovascular progenitor cells	Phase I*
Dravet Syndrome	GT	CAV2	Preclinical
Dystrophic epidermolysis bullosa	GT	Genetically modified autologous skin cells	Phase I/II*
Fanconi anemia	GT	Hematopoietic stem cells + LV-FANCA	Phase II
Glycogen storage disease type 1a (Von Gierke disease)	GT	AAV-G6PC	Preclinical
Immune deficiencies	GT	Hematopoietic stem cells + LV-Artemis	Phase I/II
	GT	Hematopoietic stem cells + LV-XSCID	Phase I/II*
	GT	Hematopoietic stem cells + LV-CGD	Follow-up study (15 years) phase I/II USA
	GT	Hematopoietic stem cells + LV-WAS	Follow-up study (15 years)
Metachromatic leukodystrophy	GT	ARSA	Préclinical
Multiple sclerosis	CT	Cytotoxic T cells	Phase I
Retinitis pigmentosa	CT	Embryonic stem cells	Phase I/II
	GT	AAV-RdCVF	Phase I/II
Sickle cell disease	GT	LV-βAS3	Phase I/II
Sickle cell skin ulcers	CT	Embryonic stem cells	Preclinical
Spinocerebellar ataxia 3 (SCA3)	GT	AON-skipex10	Preclinical
Wolfram Syndrome	P	Valproic acid	Phase II
X-linked hypophosphatemia (XLH)	GT	AAV-cFGF23	Preclinical

Off-label drugs

Lumevoq® **GT**
Leber optic neuropathy
(Expanded access)

Patient databases

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

GT Gene therapy

CT Cell therapy

P Pharmacology

* Preclinical or previous clinical phases funded by AFM-Telethon.