LISTE DES FINANCEMENTS ACCORDES PAR L'AFM-TELETHON EN 2024

Au titre de sa mission Guérir (hors financements accordés à Généthon, CECS-Istem, Institut de Myologie)

POLITIQUE D'APPEL D'OFFRES

Projets soumis à l'appel d'offres annuel

Commission thématique : Myologie fondamentale

Aides aux jeunes chercheurs post-doctorants

KUROSAWA	**Tamaki**	PARIS	FRANCE	Regionalization of fibroblasts of muscle attachments
PONTHEAUX	**Florian**	BANYULS-SUR-MER	FRANCE	Smooth muscles as mechano-chemical regulators of repair processes: nature of the actin-based topological defects during jellyfish regeneration

Financements de projets Tremplins

SEBTI	**Yasmine**	LILLE	FRANCE	Deciphering the identity and role of Type 2 Innate Lymphoid Cells in skeletal muscle homeostasis
SILVA-ROJAS	**Roberto**	MADRID	ESPAGNE	TiTransMyoMech: Titin Transcriptional impact on Myotube Mechanosignaling
THERET	**Marine**	VANCOUVER	CANADA	Role of the TAK1 signaling pathway in FAP in tissue remodeling

Financements de projets

CARNESECCHI	**Julie**	MONTPELLIER	FRANCE	Illuminating the isoform-regulatory function of the Hox transcription factor Ultrabithorax during muscle development
CIENIEWSKI-BERNARD	**Caroline**	LOOS	FRANCE	Trojan-CryAB: an innovative therapeutic approach for skeletal muscle diseases resulting from proteotoxicity and protein aggregation
COMAI	**Glenda**	PARIS	FRANCE	Homeostasis and repair of extraocular muscle myofibres
HUBE	**Florent**	PARIS	FRANCE	SNORNAs in normal and pathological human muscle
KELLY	**Robert G.**	MARSEILLE	FRANCE	Defining the unique developmental program of the trapezius muscle at the head trunk interface
MAMMUCARI	**Cristina**	PADOVA	ITALIE	The role of mitochondrial calcium signalling in aging skeletal muscle
MARCELLE	**Christophe**	VILLEURBANNE	FRANCE	Epaxial Muscle Patterning
NEVES	**Joana**	LISBOA	PORTUGAL	The contribution of immune aging to skeletal muscle regenerative failure
PIERCY	**Richard J.**	LONDON	UK	Defining dystrophin's transcriptional domains in healthy and damaged muscle
SANDRI	**Marco**	PADOVA	ITALIE	DISSECTING THE ROLE OF THE UBIQUITIN LIGASE MUSA1/Fbxo30 IN MUSCLE PHYSIOLOGY AND PHYSIOPATHOLOGY
SAURIN	**Andrew**	MARSEILLE	FRANCE	Hox control of Drosophila adult myogenesis
SINIGAGLIA	**Chiara**	BANYULS-SUR-MER	FRANCE	Jellyfish smooth muscles in development, tissue repair and regeneration

Commission thématique : Bases Moléculaires et Physiopathologie des Dystrophies Musculaires

Aides aux jeunes chercheurs post-doctorants

LESSARD	**Lola**	OTTAWA	CANADA	AMPK signaling in Type I Myotonic Dystrophy, a potential therapeutic target
MASSENET	**Jimmy**	LA JOLLA	USA	Identification of cell autonomous alterations of the 3D epigenome caused by Dystrophin deficiency

Financements de projets Tremplins

BOIVIN	**Manon**	ILLKIRCH-GRAFFENSTADEN	FRANCE	OculoPharyngoDistal Myopathies: decipher the pathogenic mechanisms of their causing GGC-expansion mutations to develop therapeutic approaches
GREEN	**Alexander**	OTTAWA	CANADA	A Protective Role for GCN5/PCAF in Neuromuscular Disease
KIRBY	**Tyler**	AMSTERDAM	PAYS-BAS	Nuclear trafficking of giant mRNA molecules in LMNA-related muscular dystrophies
LÓPEZ-MÁRQUEZ	**Arístides**	ESPLUGUES DE LLOBREGAT	ESPAGNE	CMG2 receptor: Deciphering its role in the pathophysiology of COL6-RDs and its potential as a therapeutic target
TIMPANI	**Cara**	MELBOURNE	AUSTRALIE	Uncovering the mechanisms of Faecal Microbiota Transplantation as a novel therapy in the treatment of neuromuscular disease
VIOLLET	**Benoit**	PARIS	FRANCE	Implication of NRLP3 inflammasome and AMPK signaling in inflammation-induced skeletal muscle atrophy

Financements de projets

ASIOLI	**Sofia**	BOLOGNA	ITALIE	Role of TNPO3 in the pathogenetic mechanism of LGMD D2: comparison between disease models
BOUTER	**Anthony**	PESSAC	FRANCE	Interplay of annexins, macrophages and fibroadipogenic progenitors in the development of muscular dystrophies
CHARLET-BERGUERAND	**Nicolas**	ILLKIRCH	FRANCE	Decipher the mechanisms underlying muscle weakness in Myotonic Dystrophy
COLLAS	**Philippe**	OSLO	NORVÈGE	Deregulation of the epigenome and transcription factor networks in LMNA-related muscular dystrophies
D'AMBROSI	**Nadia**	ROME	ITALIE	Targeting S100A4 to improve inflammation and fibrosis in Duchenne Muscle Dystrophy
DI PIETRO	**Lorena**	ROME	ITALIE	DIssecting the pathogenetic RolE played by the different Cell types within the skeletal muscle sTem cell nIche Of FSHD patieNts
DÍAZ-MANERA	**Jordi**	NEWCASTLE	UK	MATRIX: Understanding the role of extracellular matrix in the process of muscle degeneration in muscular dystrophies
ESCANDE-BEILLARD	**Nathalie**	ISTANBUL	TURQUIE	Novel Muscular Dystrophy caused by SNUPN Mutations: From Innovative modeling to therapeutic insights
GABELLINI	**Davide**	MILAN	ITALIE	Structure-Function characterization of a novel DUX4 inhibitor to develop a drug-like treatment for FSHD muscular dystrophy
PREVITALI	**Stefano Carlo**	MILAN	ITALIE	Modulation of modifier genes to improve muscle regeneration and metabolism in LAMA2-RD animal models
RAVEL-CHAPUIS	**Aymeric**	OTTAWA	CANADA	Characterization of the DM1 myosecretome and its multisystemic impact
TEDESCO	**Francesco Saverio**	LONDON	UK	Studying the impact of nuclear shape abnormalities in laminopathies using advanced in vitro models
WANSINK	**Derick G.**	NIJMEGEN	PAYS-BAS	DM1 repeat length-dependent disease heterogeneity in muscles

Commission thématique : Bases moléculaires et physiopathologie des autres maladies neuromusculaires

Aides aux jeunes chercheurs post-doctorants

DE CALBIAC	**Hortense**	PARIS	FRANCE	Identifying and understanding new genetic causes of acute rhabdomyolysis predisposition
GABILLARD	**Claudie**	ANGERS	FRANCE	Modulation of mitochondrial DYNAmics as a therapeutic approach to maintain mtDNA integrity and mitigate the accumulation of deficient myofibers in mitochondrial MYOpathies

Financements de projets Tremplins

BRISCHIGLIARO	**Michele**	MIAMI	USA	Mitochondrial RNA Folding: Mediation in Encephalomyopathies
LAITILA	**Jenni**	HELSINKI	FINLANDE	Determining the role of muscle myosin in the pathogenesis of neuromuscular disorders – a novel target for therapy?

Financements de projets

BARIS	**Olivier**	ANGERS	FRANCE	Modulation of mitochondrial DYNAmics as a therapeutic approach to maintain mtDNA integrity and mitigate the accumulation of deficient myofibers in mitochondrial MYOpathies
CHEVROLLIER	**Arnaud**	ANGERS	FRANCE	Induced neurons resulting from direct conversion of fibroblasts from patients with mitochondrial diseases: a new model to explore ATP synthase reverse function and its role in mitochondrial fission
COLLE	**Marie-Anne**	NANTES	FRANCE	Effects of enzyme replacement therapy on lysosome-autophagosome-mitochondria cross-talk and muscle repair in Pompe disease (LYSAUMI)
COPIC	**Alenka**	MONTPELLIER	FRANCE	Establishment of molecular mechanisms leading to protein aggregation and amyloid formation in distal myopathy patients using multidisciplinary approaches including genetic screening, bioinformatics and biochemical and structural analysis
DEVAL	**Emmanuel**	VALBONNE	FRANCE	ACID-SENSING ION CHANNEL 3 IN THE PATHOPHYSIOLOGY OF CHRONIC MUSCLE PAIN
HEARD	**Edith**	HEIDELBERG	ALLEMAGNE	Allele-specific epigenetic regulation of Bag3: physiopathological implications and new strategies for dilated cardiomyopathy
KÜHL	**Inge**	GIF-SUR-YVETTE	FRANCE	Investigating the role of an uncharacterized mitochondrial RNA Polymerase-associated Factor: towards a finer understanding of mammalian OXPHOS biogenesis
METODIEV	**Metodi**	PARIS	FRANCE	Molecular basis of impaired OXPHOS biogenesis caused by deficient protein maturation in mitochondria
ROSSI	**Daniela**	SIENA	ITALIE	CCDC78: from basic research to mechanisms of disease

Commission thématique : Système nerveux - Motoneurone et jonction neuromusculaire

Aides aux jeunes chercheurs post-doctorants

KLEIJWEGT	**Constance**	MONTPELLIER	FRANCE	Chromatin proteomics of extended GAA/TTC trinucleotides repeats : Implication for Friedreich Ataxia
MAINO	**Eleonora**	BASEL	SUISSE	Characterization of pathophysiological mechanisms involved in the selective vulnerability of muscles in Myasthenia Gravis
MILIOTO	**Carmelo**	PADOVA	ITALIE	Investigating spinal motor neurons vulnerability in C9orf72 DPR knock-in mouse models
NABAVIZADEH	**Nasrinsadat**	ISTANBUL	TURQUIE	Deciphering the function of a novel gene in an ALS-like syndrome

Financements de projets Tremplins

AMOR BARRIS	**Silvia**	ANTWERPEN	BELGIQUE	Unveiling the role of HINT1 in health and disease with a special focus on Ca2+ dynamics
FERREIRA DA SILVA	**Tiago**	PORTO	PORTUGAL	Deciphering the mechanisms of impaired neuron-muscle synapses caused by loss of plasmalogens
MARZULLO	**Marta**	ROME	ITALIE	Dissecting the epigenetic control of TDP-43 expression during aging
PIOL	**Diana**	PADOVA	ITALIE	Elucidating the role of axonal androgen receptor signaling in spinal and bulbar muscular atrophy

Financements de projets

ARRIBAT	**Yoan**	MONTPELLIER	FRANCE	Organelle coordination in hereditary spastic paraplegia: deciphering protein network and restoring lipid balance
CESCON	**Matilde**	PADOVA	ITALIE	Targeting Beclin 1-regulated pathways to counteract peripheral demyelination
D'ANTONIO	**Maurizio**	MILAN	ITALIE	Axonal degeneration in CMT2J/I neuropathies: molecular mechanisms and therapeutic strategies
D'ANTONIO	**Maurizio**	MILAN	ITALIE	Molecular mechanisms and therapeutic approaches for CMT1E neuropathies
DUPUIS	**Luc**	STRASBOURG	FRANCE	Muscle contribution to FUS-associated ALS: genetic and mechanistic insights
MADARO	**Luca**	ROME	ITALIE	RETINOIC ACID SIGNALLING IN THE REGULATION OF MUSCLE INNERVATION
MAIRE	**Pascal**	PARIS	FRANCE	Delineating Mechanisms of Neuromuscular Crosstalk in Myofiber and Motoneuron Diversity
MANGIN	**Jean-Marie**	PARIS	FRANCE	Understanding how the electrical activity of midline radial glia participates to neuromuscular development and function
MONANI	**Umrao R.**	NEW YORK	USA	How does a novel spinal muscular atrophy modifier suppress motor neuron disease?
OURY	**Franck**	PARIS	FRANCE	Decipher the role of the Ocn-induced autophagy in motoneurons and its anti-aging therapeutic potential
ROJO	**Manuel**	BORDEAUX	FRANCE	Cellular assays to establish the consequences and infer the pathogenicity of MFN2 variants
SELLIER	**Chantal**	STRASBOURG	FRANCE	Identify the role of NUP50 in motor neuron physiology and ALS pathology

Commission thématique : Cellules souches

Aides aux jeunes chercheurs post-doctorants

CHRYSOSTOMOU	**Eleni**	PARIS	FRANCE	Fibrillin-1 as a modulator of quiescence in skeletal muscle stem cells
LY	**Ha My**	OTTAWA	CANADA	The role of PINK1/PARKIN mediated the mitophagy in the regulation of muscle stem cell fate choices and muscle regeneration
POITOU MOUCAUD	**Blandine**	MONTRÉAL	CANADA	Optogenetics and muscle atrophy: delivery of optogenetic contractile properties to skeletal muscle by MuSCs

Financements de projets Tremplins

COSSEC	**Jack-Christophe**	PARIS	FRANCE	Uncovering Rare Disease Signatures in Cell Fate Transitions through a Genetic Perturbation Screen in Embryo-Like Structures
MORIN	**Xavier**	PARIS	FRANCE	Unequal mitotic mitochondrial segregation as an asymmetric fate determinant in neural stem cells
VILLALOBOS	**Elisa**	NEWCASTLE UPON TYNE	UK	Stiffness-DMD: Deciphering the role of stiffness in the commitment of fibro-adipogenic progenitors in Duchenne muscular dystrophy

Financements de projets

BURELLE	**Yan**	OTTAWA	CANADA	Targeting mitochondrial quality control in stem cells to promote muscle repair
GUARDIOLA	**Ombretta**	NAPLES	ITALIE	Exploring the complexity of chemokine/chemokine receptor dynamics in muscle stem cell population: insights into CXCR2 signalling and heterogeneity
HELMBACHER	**Françoise**	MARSEILLE	FRANCE	In vivo modelling and modulating fibroadipogenic differentiation
LI	**Zhenlin**	PARIS	FRANCE	Involvement of synemin in cardiomyopathies using human induced pluripotent stem cells (hiPSC) and cell type-specific knock-out murine models
MODJTAHEDI	**Nazanine**	LYON	FRANCE	Regulation of the AIF/CHCHD4-dependent mitochondrial import pathway in human skeletal muscle stem cells
OLGUIN	**Hugo**	SANTIAGO	CHILI	Nedd4-1 promotes regeneration by regulating autophagy in differentiating muscle stem cells
SENGENES	**Coralie**	TOULOUSE	FRANCE	Harnessing Adipose stromal cell (ASCs) Migration: a therapeutic LEver for muscular healTh in the elderly?

Commission thématique : Thérapie Génique et/ou Cellulaire des Maladies Rares

Aides aux jeunes chercheurs post-doctorants

AKINYELE	**Oluwaseun**	PITTSBURG	USA	Development of a Gene Therapy Approach in an Inborn Error of Polyamine Metabolism
ALMEIDA	**Camila**	COLUMBUS	USA	Evaluation of AAV9.U7snRNAs in Myotonic Dystrophy type 1 preclinical mouse models
ANGELINI	**Giuseppe**	MONTPELLIER	FRANCE	Silencing of GIRK4 by AAV as a treatment of sinus node dysfunction in mouse models and patient-specific pacemaker-like hiPS-derived cardiomyocytes
TIBERIO	**Federica**	ROME	ITALIE	Development of functionalized FGF2-decoRated PLGA-PEG-bis-sulfone nanoparticles for targeted delivery of therapeutic siRNAs in Crouzon syndrome

Financements de projets Tremplins

BRUSSON	**Mégane**	PARIS	FRANCE	Development of genome editing strategies for alpha-thalassemia
KEOGH	**Michael**	NEWCASTLE UPON TYNE	UK	Developing a gene therapy for desminopathy
MALERBA	**Alberto**	EGHAM	UK	Enhancing muscle strength in muscular dystrophies through RPL3L inhibition
MIRANDA	**Catarina**	COIMBRA	PORTUGAL	Direct Glia Reprogramming and Mutant Ataxin-3 Silencing as a New Combined Therapeutic Strategy to Treat Spinocerebellar Ataxia Type-3 - a Proof-of-Principle Study
PULMAN	**Juliette**	PARIS	FRANCE	Gene repression using CRISPRi for the treatment of autosomal dominant retinitis pigmentosa

Financements de projets

ARRIBAT	**Yoan**	MONTPELLIER	FRANCE	Development of the first system of organelle optimisation and transplantation to treat mitochondrial diseases
BELENGUER	**Pascale**	TOULOUSE	FRANCE	Proof of principle of an original gene therapy for a rare hereditary mitochondrial-related optic atrophy
EL AMRAOUI	**Aziz**	PARIS	FRANCE	Humanized mice to expedite effective translation of reliable inner ear gene therapies
HOVNANIAN	**Alain**	PARIS	FRANCE	Base editing-mediated correction of recurrent mutations in COL7A1 to treat RDEB
KALATZIS	**Vasiliki**	MONTPELLIER	FRANCE	CRISPR/Cas genome-editing to treat autosomal dominant retinitis pigmentosa caused by the G56R mutation in NR2E3: pre-clinical proof-of-concept in human retinal organoids
LOLLO	**Giovanna**	VILLEURBANNE	FRANCE	Lipid nanoparticles delivery of antisense oligonucleotides to correct the pathological cause of muscular dystrophies
MINCZUK	**Michal**	CAMBRIDGE	UK	Mitochondrial Genome Base Editing for Advancing Therapies of Neuromuscular Diseases
MONANI	**Umrao R.**	NEW YORK	USA	Modulating SLC2A1 activity with a novel lncRNA to treat infantile-onset Glucose Transporter-1 deficiency syndrome
PARENTI	**Giancarlo**	NAPLES	ITALIE	Targeting secondary dysregulation of cellular pathways and functions to improve gene therapy for Pompe Disease
PIGUET	**Françoise**	PARIS	FRANCE	Combined gene therapy approach for Pelizaeus Merzbacher disease
ROSSI	**Andrea**	DÜSSELDORF	ALLEMAGNE	Small molecules that enhance Prime Editing in therapeutic applications
RUZZENENTE	**Benedetta**	PARIS	FRANCE	Gene therapy cure for mitochondrial disease caused by mutations in the mitochondrial RNA stability factor LRPPRC
WEIN	**Nicolas**	COLUMBUS	USA	Preclinical evaluation of AAV.U7snRNAs in Myotonic Dystrophy type 1 mouse models

Commission thématique : Thérapie Pharmacologique des maladies neuromusculaires et Recherche Translationnelle

Aides aux jeunes chercheurs post-doctorants

BOULINGUIEZ	**Alexis**	EGHAM	UK	Pharmacological approaches to improve folding of microdystrophin in Duchenne muscular dystrophy
FABBRIZIO	**Paola**	MILAN	ITALIE	Modulating the P2X7 axis in Spinal and Bulbar Muscular Atrophy muscle cell populations as an innovative approach to promote myogenesis

Financements de projets Tremplins

GOOSSENS	**Remko**	LEIDEN	PAYS-BAS	Functional in vitro modelling of Duchenne AON therapies using 3D muscle bundles
OTTRIA	**Roberta**	MILAN	ITALIE	The endocannabinoid system as new target in Duchenne's muscular dystrophy: getting inside the mechanistic and the anti-inflammatory aspects
SANDONÀ	**Martina**	ROME	ITALIE	Liquid biopsy for non-invasive monitoring of DMD progression: identification of a diagnostic and prognostic signature in Extracellular vesicle released from muscle into the blood

Financements de projets

BRICHARD	**Sonia**	BRUXELLES	BELGIQUE	Adiponectin and its mimics for the treatment of muscular dystrophies
CIRAK	**Sebahattin**	COLOGNE	ALLEMAGNE	Biomarker discovery and validation for LGMD2I/FKRP-related muscular dystrophy.
DE STEFANI	**Diego**	PADOVA	ITALIE	Fight TMEM65 diseases: from mitochondrial calcium to neuromuscular degeneration
DI SCHIAVI	**Elia**	NAPLES	ITALIE	A combinatorial pharmacotherapeutic approach to counteract Spinal Muscular Atrophy
DORCHIES	**Olivier**	GENEVA	SUISSE	Duchenne muscular dystrophy: Phenotyping and validation of better murine models for improving preclinical research and clinical translation – a step further
DUPUIS	**Luc**	STRASBOURG	FRANCE	Preclinical evaluation of dual orexin receptor antagonist in amyotrophic lateral sclerosis
HORNSTEIN	**Eran**	REHOVOT	ISRAËL	microRNA biomarkers for ALS progression - revised
HORNSTEIN	**Eran**	REHOVOT	ISRAËL	microRNA as biomarkers for phenoconversion in pre familial ALS
LATELLA	**Lucia**	ROME	ITALIE	SerpinE1 inhibitor as therapeutic tool in the treatment of DMD
LEVY-LAHAD	**Ephrat**	JERUSALEM	ISRAËL	VRK1-related motor neuron disease: biological models for drug repurposing
MALERBA	**Alberto**	EGHAM	UK	Improving protein folding to enhance microdystrophin expression in Duchenne muscular dystrophy
MALTECCA	**Francesca**	MILAN	ITALIE	Efficacy of IFB-088 on the neuromuscular phenotype of the Spastic-Ataxia type 5 mouse model
PUCEAT	**Michel**	MARSEILLE	FRANCE	Cardiomyopathy Arrhythmia and Conduction defects Therapy
RYBALKA	**Emma**	MELBOURNE	AUSTRALIE	Toward the Clinic: Evaluating the long-term efficacy of re-purposed dimethyl fumarate for the treatment of Duchenne Muscular Dystrophy
SAENZ PENA	**Amets**	DONOSTIA	ESPAGNE	Wnt signaling pathway activation in muscular dystrophy models: a search of therapeutic use in LGMDR1 muscular dystrophy
SANDONÀ	**Dorianna**	PADOVA	ITALIE	Target validation approach to unveil the mechanism of action of C17 in sarcoglycanopathies
VENEREAU	**Emilie**	MILAN	ITALIE	Preclinical Evaluation of a Designer HMGB1 as a Drug Candidate for Duchenne Muscular Dystrophy

Commission thématique : Recherche Médicale

Aides aux jeunes chercheurs post-doctorants

ANDRADE	**Ricardo**	NANTES	FRANCE	Innovative Multiparametric UtraSound biomarkers for muscular dystrophies: towards a comprehensive monitoring of musCLE degeneration and responses to therapy

Financements de projets

CORTESE	**Andrea**	LONDON	UK	Long-read Sequencing In Undiagnosed Neuromuscular Diseases
GAGNON	**Cynthia**	JONQUIERE	CANADA	Understanding the impact of central nervous system impairments on the performance of instrumental activities of daily living in people living with myotonic dystrophy type 1
MARTINEZ ALEJOS	**Roberto**	ROUEN	FRANCE	Conditions for Optimizing Mechanical Insufflation-Exsufflation Settings to Improve Clearance of Pulmonary Secretions
SORARU	**Gianni**	PADOVA	ITALIE	Autonomic nervous system involvement in Spinal and Bulbar Muscular Atrophy (SBMA) - AutoSBMA
TASCA	**Giorgio**	NEWCASTLE UPON TYNE	UK	Dista-Myo part 2: Natural history of distal and myofibrillar myopathies assessed by clinical and technological outcome measures - a two-year extension.
VUILLEROT	**Carole**	BRON	FRANCE	Development and validation study of a Motor Function Measure digitalized playful completion modules
WAHBI	**Karim**	PARIS	FRANCE	Venous Thromboembolism in Myotonic Dystrophy Type 1: Unravelment of Molecular Mechanisms and Biomarker Development

Appel d'offres Doctorants

AGRESTI	**Laura**	LYON	FRANCE	Compartmentalization of the C. elegans sarcolemma by the dystrophin-associated protein complex
ALCOLEI	**Allan**	LYON	FRANCE	Characterization of a novel role for Troponin I in muscle aging
BATAILLARD	**Méghane**	CASTELNAU-LE-LEZ	FRANCE	Systemic gene therapy using NCS1 for treating Wolfram syndrome
BOURGETON	**Tiffany**	NANTES	FRANCE	Effect of Enzyme replacement therapy (ERT) on skeletal muscle pathophysiology in Pompe disease : specific focus on satellite cells and autophagy
DAY	**Lucie**	PARIS	FRANCE	Role of ADAR1 and RNA editing in peripheral myelin maintenance
GISBERT	**Vincent**	MONTPELLIER	FRANCE	Organelle alterations converge on neurodegeneration and lack of regeneration in hereditary spastic paraplegia
GUTKOWSKI	**Lisa**	TOULOUSE	FRANCE	Study of the pathophysiology of muscular defects associated to Noonan syndrome
HAUTBOIS	**Marie**	CLAMART	FRANCE	Base editing-mediated correction of recurrent mutations in COL7A1 to treat RDEB
MACAUX	**Gaspard**	PARIS	FRANCE	The crosstalk between motoneurons and myofibers
MAIELLANO	**Greta**	LYON	FRANCE	Role of the TMED proteins in acetylcholine receptor biosynthesis and implication in neuromuscular diseases
MATOUK	**Meriem**	MONTIGNY LE BRETONNEUX	FRANCE	Characterize satellite cell associated dystrophin in muscle stem cells
MOURIER	**Hugo**	MONTPELLIER	FRANCE	The NRF2 pathway: a possible therapeutic approach in amyotrophic lateral sclerosis
MOURJI	**Kenza**	PARIS	FRANCE	Single-chain antibodies against pathogenic CTG/CUG repeats: diagnosis and therapeutic applications for myotonic dystrophy
PHONGSAVANH	**Xaysongkhame Micky**	LUDRES	FRANCE	Evaluation of HDAC inhibitors to increase dystrophin rescue in DMD following exon skipping therapy
ROLLAND	**Fanny**	MONTPELLIER	FRANCE	Characterization of the first organelle factory for therapeutic transplantation
ROMAN	**Olga**	STRASBOURG	FRANCE	Identification of the role of NUP50 in motor neuron physiology and ALS pathology
ROUÉ	**Clémence**	MONTPELLIER	FRANCE	Autoimmune Nodopathy : DEtermine the RoleS Of ADAM10/17 in Neuropathy
ROUSSET	**Célia**	MARSEILLE	FRANCE	Defining the unique developmental program of the trapezius muscle
RUBENS	**Paula**	PARIS	FRANCE	DNA Methylation Landscape of Normal, Mitochondrial Mutated, and Three-Parent Preimplantation Embryos
SANTINI	**Annalisa**	PARIS	FRANCE	Development of in vivo gene therapy approaches for β-hemoglobinopathies
SARDE	**Liza**	PARIS	FRANCE	Muscle stem cell division and migration
SOYLU	**Talha**	MONTIGNY-LE-BRETONNEUX	FRANCE	Understanding how subcellular dystrophin protects muscle fibers from degeneration
THIBAULT	**Chloé**	BORDEAUX	FRANCE	Development of novel Cas9-nickases for gene therapy of rare genetic diseases: application to the congenital erythropoietic porphyria
VAHDAT	**Juliette**	MARSEILLE	FRANCE	Exploring ventricular conduction system structure and function in DMD mouse models
VAUCOURT	**Mathilde**	TOULOUSE	FRANCE	Selective autophagy in X-linked centronuclear myopathy: molecular mechanisms and pathophysiological relevance
YTIER	**Charline**	MARSEILLE	FRANCE	Modeling, modulating, and monitoring intramuscular fibro-fatty infiltrates

Projets soumis à l'appel d'offres thématique Recherche Dystrophie Myotonique

FURLING	**Denis**	PARIS	FRANCE	Tricyclo-DNA antisense oligonucleotide treatment for Myotonic Dystrophy

Partenariats institutionnels

DUPONT	**Jean-Baptiste**	PARIS	FRANCE	Next generation modelling of human myogenesis and skeletal muscle diseases with pluripotent stem cells
KIM	**Minchul**	PARIS	FRANCE	The biology of syncytial cells: Dissecting the mechanisms and functions of nuclear differentiation inside skeletal muscle syncytium
LAUGEL	**Vincent**	STRASBOURG	FRANCE	Cohorte de suivi longitudinal SMA-DNN: Dépistage néonatal de l'Amyotrophie Spinale - Projet pilote SMA-DNN France
ZLATANOVA	**Ivana**	PARIS	FRANCE	Decoding cell-type specific chromatin landscape to elucidate heart regeneration

Partenariats associatifs

Cure CMD	LAKEWOOD	USA	Targeting Primary Cilium-Related Molecular Pathways to Correct Tendon Cell Defects in Collagen VI Congenital Muscular Dystrophies - CENNI Vittoria
IFCAH (International Fund raising for research on Congenital Adrenal Hyperplasia)	PARIS	FRANCE	Advancing Adrenal Stem Cell Transplantation for Precision Therapies in Congenital Adrenal Hyperplasia - OIKONOMAKOS Ioannis
RETINA France	COLOMIERS	FRANCE	Optogenetic engineering of stem cell-derived photoreceptors to improve visual restoration - ORIEUX Gaël
SMA EUROPE	CHIPPING CAMDEN	UK	12th international Call for SMA Research Projects - Financial contribution granted to: - Leveraging SMN role in translation to develop the next-gen of biomarkers for SMA - VIERO Gabriella - Skeletal Muscle Stem Cells as untapped therapeutic targets for SMA long-term treatment - DIDIER Nathalie - Deciphering the Molecular Landscape of Neuromuscular Development in Spinal Muscular Atrophy - GAZZOLA Morgan - Investigating Calcium-induced mitochondrial dysfunction in zebrafish and iPSC models of SMA - CIURA Sorana - Are microvascular defects relevant in Spinal Muscular Atrophy ?: Characterization of the mouse model - PARSON Simon
VAINCRE LA MUCOVISCIDOSE (VLM)	PARIS	FRANCE	Appel à projets scientifiques 2024 : - Édition génique au locus CFTR par Homology-independent targeted insertion (HITI) : Évaluation de la stratégie moléculaire dans des cellules épithéliales et des organoïdes des voies respiratoires - BERGOUGNOUX Anne - Ciblage des ARNt pour une suppression traductionnelle spécifique de mutations non-sens dans le gène CFTR - NAMY Olivier

Projets Ignition

BARTHELEMY	**Inès**	MAISONS-ALFORT	FRANCE	Adoptive Cell Therapy In Fibrosis : application to Duchenne Muscular Dystrophy
BENCHOUA	**Alexandra**	CORBEIL-ESSONNES	FRANCE	Paediatric application of dopamine synthesizing lentiviral vectors in genetic dopamine deficiency disorders: Lesch-Nyhan Disease as proof of concept
DELPRAT	**Benjamin**	MONTPELLIER	FRANCE	Original systemic gene therapy approach to treat Wolfram syndrome
NICOLE	**Sophie**	MONTPELLIER	FRANCE	Nav1.4 activators for a correction of SCN4A-related muscle weaknesses
SUMARA	**Izabela**	ILLKIRCH	FRANCE	Therapeutic dissolution of aberrant nucleoporin condensation in Fragile X syndrome using gene therapy approaches
VISCOMI	**Carlo**	PADOVA	ITALIE	Harnessing mitophagy to treat mitochondrial myopathies

Projets soumis ou suivis hors calendrier Appel d'Offres

SACCONI	**Sabrina**	NICE	FRANCE	A 12 months prospective natural history study to gain insight FSHD2 pathophysiology and disease progression

ACTIONS STRATEGIQUES

Projets stratégiques

AGBULUT	**Onnik**	PARIS	FRANCE	DESmin-related CARdiomyopathy ThErapeuticS Development - DESCARTES
BOMONT	**Pascale**	LYON	FRANCE	Therapy for Giant Axonal Neuropathy
CORRAL-DEBRINSKI	**Marisol**	PARIS	FRANCE	Neuroglobin gene overexpression: a promising tool for treating a large spectrum of neurological disorders thanks to its ability to safely and sustainably preserve mitochondrial integrity
DECRESSAC	**Michael**	LA TRONCHE	FRANCE	Implementation of a gene therapy strategy for MTFMT-associated Leigh syndrome
DUMONCEAUX	**Julie**	LONDON	UK	DUX4 in FSHD: pathophysiology and therapeutic approaches
FRANCO	**Brunella**	POZZUOLI	ITALIE	miR-181a/b downregulation: a pan therapeutic approach to treat rare diseases associated with mitochondrial dysfunction
GALY	**Anne**	CORBEIL-ESSONNES	FRANCE	Sickle cell disease gene therapy program
LAPORTE	**Jocelyn**	ILLKIRCH	FRANCE	Pathophysiology and therapeutic proof-of-concepts for congenital myopathies
OLIVIER-FAIVRE	**Laurence**	DIJON	FRANCE	PERIGENOMED-CLINICS 1: a pre-pilot to assess the feasibility and acceptability of newborn screening using panel-based genome sequencing in France
POURQUIE	**Olivier**	BOSTON	USA	Toward cell therapy for Duchenne Muscular Dystrophy: characterization of regenerative potential of hIPS derived Pax7+ cells
SEREDA	**Michael**	GÖTTINGEN	ALLEMAGNE	A multi-omic approach to the identification of novel biomarkers in early Charcot-Marie-Tooth 1A disease (CMT1A)
VAILLEND	**Cyrille**	ORSAY	FRANCE	DECODING CENTRAL DEFECTS IN DYSTROPHINOPATHIES : FROM DIAGNOSTIC TO REMEDIATION
VILQUIN	**Jean-Thomas**	PARIS	FRANCE	Comparative study of iPSC-derived myogenic precursors proposed for cell therapy in a model of Duchenne muscular Dystrophy
ZAKHIA	**Raymond**	EVRY	FRANCE	Collaborative program for a systematic, mecanistic and pharmacologic approach of rare neuromuscular diseases

Pôles stratégiques

MAGDINIER	**Frédérique**	MARSEILLE	FRANCE	Modelisation and Therapeutic Approaches for Rare Diseases : Pathologies liées au gène STIM1 ; Dysferlinopathies ; CMT ; FSHD ; Cardiomyopathies génétiques (LMNA ; Rasopathies…) ; Encephalopathie épileptique KCQN2 ; Syndrome de Rett ; Pathologies du vieillissement (Progeria, progeria like, MADaM syndrome, Hallermann-Streiff syndrome…) ; David syndrome
RELAIX	**Frédéric**	CRÉTEIL	FRANCE	An integrated translational program for neuromuscular disorders : Dystrophies musculaires : DMD / FSHD ; SMA ; Myopathies congénitales ; Myopathies inflammatoires
SCHAEFFER	**Laurent**	LYON	FRANCE	MyoNeurALP2, The Research Network dedicated to Neuromuscular Disorders in Rhone Alpes Auvergne : Dystrophies musculaires : DMD / DM1 ; Myopathies inflammatoires ; Myopathie centronucléaire ; Myopathies congénitales / Myopathie congénitale à « central cores » ; Hyperthermie maligne ; Ataxies, dont Ataxie de Friedreich ; Maladies du motoneurone (SLA / SMA / CMT) ; Myasthénie ; Neuropathies sensorielles

Structures stratégiques

FONDATION MALADIES RARES	PARIS	FRANCE	Appel à projets auprès des Filières de Santé Maladies Rares - Projets financés: - Orai1 Ca2+ channel: a new therapeutic target in PAH - ANTIGNY Fabrice - Promoting regenerative inflammation to improve muscle homeostasis in Duchenne Muscular Dystrophy - CHAZAUD Bénédicte - Consolidation of an innovative strategy for the treatment of calcitriol-mediated hypercalcemia - LAVERNY Gilles - Inducing albumin-mediated recycling of endogenous proteins: a novel strategy to treat quantitative deficiencies of plasma proteins - LENTING Peter - HDAC6 inhibitors to treat Duchenne Muscular Dystrophy - SCHAEFFER Laurent
FONDATION MALADIES RARES	PARIS	FRANCE	Subvention 2024
GENOPOLE	EVRY-COURCOURONNES	FRANCE	Participation financière de l'AFM au budget 2024 du GIP GENOPOLE

Outils stratégiques (bases de données)

BASSEZ	**Guillaume**	PARIS	FRANCE	International Myotonic Dystrophy Registry - iDM-Scope
DESGUERRE	**Isabelle**	PARIS	FRANCE	French Dystrophinopathies Registry (DYS Registry)
LAFORET	**Pascal**	GARCHES	FRANCE	Registre Français des Sarcoglycanopathies (Sarcoglycanopathy French Registry)
MALFATTI	**Edoardo**	CRETEIL	FRANCE	French national registry for Calpainopathies
SACCONI	**Sabrina**	NICE	FRANCE	French national registry for FacioScapuloHumeral muscular Dystrophy (FSHD)
WAHBI	**Karim**	PARIS	FRANCE	Cardiomyopathy database

Plateformes stratégiques

ATTARIAN	**Shahram**	MARSEILLE	FRANCE	Plateforme d'essais thérapeutiques Pédiatriques et Adultes Neuromusculaires - MARSEILLE
LAFORET	**Pascal**	GARCHES	FRANCE	Plateforme d'essais thérapeutiques Pédiatriques et Adultes Neuromusculaires - GARCHES
SOLE	**Guilhem**	BORDEAUX	FRANCE	Plateforme d'essais thérapeutiques Pédiatriques et Adultes Neuromusculaires - BORDEAUX

AUTRES ACTIONS

Manifestations scientifiques (congrès, colloques)

AGNETTI	**Giulio**	BOLOGNA	ITALIE	Gordon Research Conference on Intermediate Filaments 2024
BARIS	**Olivier**	ANGERS	FRANCE	12ème Colloque Meetochondrie
COLLOQUE JEUNES CHERCHEURS		PARIS	FRANCE	Colloque Jeunes Chercheurs 2024 - Myology 2024
DE SANTA BARBARA	**Pascal**	MONTPELLIER	FRANCE	International Forum on Visceral Myopathy 2024
FAURE	**Marie-Catherine**	SAINT ANDRÉ DE SANGONIS	FRANCE	4ème Congrès International Choroïdérémie
KINOSHITA	**June**	RANDOLPH	USA	31st annual FSHD International Research Congress
LEK	**Angela**	CHICAGO	USA	MDA Muscle Regeneration Summit 2024
MAIRE	**Pascal**	PARIS	FRANCE	Cochin symposium on "Muscle, Exercise and Health"
MILLAY	**Douglas P.**	CINCINNATI	USA	Skeletal Muscle Stem Cells in Development, Regeneration, and Adaptations
MOUNIER	**Rémi**	LYON	FRANCE	EMBO Workshop "Skeletal Muscle Development, Metabolism, and Repair during Homeostasis and Disease"
SCHMIDT	**Jens**	GÖTTINGEN	ALLEMAGNE	Support for 5th Global Conference on Myositis
SORCI	**Guglielmo**	PERUGIA	ITALIE	21st IIM International Meeting
STRAUB	**Volker**	NEWCASTLE UPON TYNE	UK	LGMDR9 Community Conference, Standards of Care Workshop and Physiotherapy Training of Trainers
TAILLANDIER	**Daniel**	ST GENÈS CHAMPANELLE	FRANCE	10th Proteasome & Autophagy Congress
TOME	**Stephanie**	PARIS	FRANCE	Long-read sequencing of expanded tandem repeats

Plateformes

DESVIGNES	**Cyril**	MEZILLES	FRANCE	Financement CEDS