

LISTE DES FINANCEMENTS ACCORDES PAR L'AFM-TELETHON EN 2021

POLITIQUE D'APPEL D'OFFRES

Projets soumis à l'appel d'offres

Commission : Myologie fondamentale

Aides aux jeunes chercheurs post-doctorants

BIANCONI Valeria, Roma, Italie

Deciphering the role of Prdm16-mediated H3K9 methylation in the control of Fibro-Adipogenic Progenitors identity and skeletal muscle repair

DE MARIO Agnese, Paris, France

Amorolfine, a positive modulator of the Mitochondrial Calcium Uniporter, as a tool to counteract atrophy and sarcopenia

HENRIQUE FERNANDES Carlos Alexandre, Paris, France

Structural characterization of a Kir potassium channel and its involvement in Andersen syndrome

Financements de projets Tremplins

SAURIN Andrew, Marseille, France

Transcriptional regulation of oxidative phosphorylation during Drosophila myogenesis

WALTZER Lucas, Clermont-Ferrand, France

Decrypting the function of the epigenetic enzyme TET in adult muscle progenitor maintenance and differentiation

Financements de projets

CARVAJAL Jaime, Seville, Espagne

Contribution of the Mrf4 gene towards muscle function during exercise-mediated hypertrophy, regeneration and ageing

DUPREZ Delphine, Paris, France

Molecular interactions between connective tissue fibroblasts and muscle cells in order to build a full musculoskeletal system

FAURE Sandrine, Montpellier, France

Neuronal regulation of intestinal smooth muscle cell differentiation and functionality

LEBRETON Gaëlle, Toulouse, France

Muscle-hematopoietic organ crosstalk: alary somatic muscle as a secretory organ regulating hematopoiesis in Drosophila

MAIRE Pascal, Paris, France

Fast myosin heavy chain locus regulation

MIQUEROL Lucile, Marseille, France

Exploring ventricular conduction system structure and function in the regenerating heart and DMD mouse models

MORO Cédric, Toulouse, France

Myolipotoxicity as a trigger of muscle weakness in metabolic diseases and aging?

MOZZETTA Chiara, Roma, Italie

Deciphering the role of Prdm16-mediated H3K9 methylation in the control of Fibro-Adipogenic Progenitors identity and skeletal muscle repair

MUNOZ-CANOVES Pura, Barcelona, Espagne

Role of circadian communication between skeletal muscle and brain to preserve muscle homeostasis and prevent age-associated muscle wasting

PENDE Mario, Paris, France

Role of mitochondrial YAP and TAZ in muscle growth, regeneration and mechanotransduction

POLGE Cécile, Saint-Genès-Champagnelle, France

Toward the inhibition of ubiquitination of contractile proteins for preserving muscle mass

ROMANELLO Vanina, Padova, Italie

Deciphering a novel link between the ubiquitin proteasome system and mitochondrial function to control muscle mass

SANDRI Marco, Padova, Italie

Dissecting the role of an uncharacterized FoxO-dependent gene that controls autophagy and ageing

SHI De-Li, Paris, France

Rbm24-controlled cytoplasmic polyadenylation in the post-transcriptional regulation of muscle cell differentiation

SOTIROPOULOS Athanassia, Paris, France

Functional cell heterogeneity underlying muscle plasticity

ZERVAS Christos, Athens, Grèce

Unravelling the mechanotransduction-dependent functions of Integrin-Linked Kinase (ILK) at myotendinous junctions

Commission : Bases Moléculaires et Physiopathologie des Dystrophies Musculaires

Aides aux jeunes chercheurs post-doctorants

REGGIO Alessio, Pozzuoli, Italie

Pharmacological reconstitution of the WNT5A/ β -catenin axis to counteract fibroadipogenic degeneration in Duchenne Muscular Dystrophy

RENZINI Alessandra, Roma, Italie

Proteomic study of HDAC4 cytoplasmic signaling, partners and secretome in DMD skeletal muscle

RUPARELIA Avnika, Clayton, Australie

Identification of therapies for Collagen VI-related congenital muscular dystrophy

Financements de projets Tremplins

GARCIA Susana, Helsinki, Finlande

Mechanisms of RNA toxicity in Myotonic Dystrophy – the role of mitochondrial dysfunction

GRASSI Francesca, Roma, Italie

Role of glia in neurological comorbidities of Duchenne muscular dystrophy

SCHIAVONE Marco, Brescia, Italie

Identification of novel druggable targets for Duchenne muscular dystrophy

SPITALI Pietro, Leiden, Pays-Bas

Understanding histopathological alterations in neuromuscular disorders by spatial transcriptomics

Financements de projets

COWAN Kyle, Ottawa, Canada

Defining the Role and Therapeutic Potential of Pannexin 1 Channels in Duchenne Muscular Dystrophy using Mouse Models and Patients Cells

HUBE Florent, Paris, France

Newly identified non-coding RNAs from alternatively spliced introns in normal and pathological muscle differentiation

MAMMUCARI Cristina, Padova, Italie

Targeting the Mitochondrial Calcium Uniporter to counteract Duchenne Muscular Dystrophy

MERONI Germana, Trieste, Italie

Role of TRIM32, the Limb Girdle Muscular Dystrophy type R8 gene, in neuro-muscular differentiation and homeostasis

MORALES Fernando, San José, Costa Rica

Comparative expression profiling of multiple tissues in myotonic dystrophy

MORESI Viviana, Roma, Italie

Unveiling the cytoplasmic functions of HDAC4 in dystrophic skeletal muscle

MUNOZ-CANOVES Pura, Barcelona, Espagne

Novel strategies to ameliorate Duchenne Muscular Dystrophy

PERROTTA Cristiana, Milano, Italie

An integrated pharmacological/antioxidant approach for Duchenne Muscular Dystrophy: acid sphingomyelinase as new therapeutic target

RAVEL-CHAPUIS Aymeric, Ottawa, Canada

Role of the RNA-binding protein HuR in Myotonic Dystrophy type 1

SUELVES Monica, Badalona, Espagne

Exploring HDAC11 functions in Duchenne Muscular Dystrophy

Commission : Bases moléculaires et physiopathologie des autres maladies neuromusculaires

Aides aux jeunes chercheurs post-doctorants

CHAKOURI Nourdine, New York, Etats-Unis

Dissecting the role of stac3 in skeletal muscle excitation contraction coupling and congenital myopathies

LIONELLO Valentina Maria, London, Royaume-Uni

Complex modeling of myotubularin myopathy using human bioengineered skeletal muscles

PALMA Alessandro, Pozzuoli, Italie

Dissecting the role of ER-phagy in Pompe Disease

PIDOUX Ludivine, Valbonne, France

Investigation of the role of ASIC channels in spinal sensitization mechanisms associated to lipid-induced chronic muscle pain

Financements de projets Tremplins

SAARI Sina, Helsinki, Finlande

Role of mito-nuclear compatibility in the metabolic landscape of mitochondrial diseases

Financements de projets

BATONNET-PICHON Sabrina, Paris, France

Diving into the pathophysiological mechanisms of desminopathies: a comparative study of KI-mouse models and human isogenic cells

CHAHINE Mohamed, Québec, Canada

Omega currents cause cardiac arrhythmias and dilated cardiomyopathy

COLLE Marie-Anne, Nantes, France

Effects of enzyme replacement therapy on lysosome-autophagosome-mitochondria cross-talk and muscle repair in Pompe disease (LYSAUMI)

DEVAL Emmanuel, Valbonne, France

Acid-sensing ion channel 3 in the pathophysiology of chronic muscle pain

HNIA Karim, Toulouse, France

Molecular functions of MTM1-related phosphoinositides in XLCNM pathophysiology

MAZAT Jean-Pierre, Bordeaux, France

How to bypass mitochondrial ATP synthase deficiencies? The role of mitochondrial carriers MitoBAD (Mitochondrial Bypass ATPsynthase Deficiencies)

METODIEV Metodi, Paris, France

Deficient protein processing in mitochondrial diseases

MITRANI-ROSENBAUM Stella, Jerusalem, Israël

Cellular Models to explore GNE functions in muscle

SCHMIDT Jens, Göttingen, Allemagne

Epigenetic modulation of pro-inflammatory cell stress in inclusion body myositis

SCORRANO Luca, Padova, Italie

Enhancing Opa1-dependent cristae structure to combat mitochondrial diseases

STENZEL Werner, Berlin, Allemagne

Studies of protein quality control and proteolysis in the genesis of acute and chronic myositis – the example of Immune-Mediated Necrotizing Myopathy (IMNM) and Inclusion Body Myositis (sIBM)

SWAN Laura, Liverpool, Royaume-Uni

INPP5K-mediated congenital muscular dystrophy models of motor neuron branching and function

ZORZANO Antonio, Barcelona, Espagne

Mitochondrial dynamics as a key process to prevent muscle inflammation and search of novel therapies in inflammatory myopathies

Commission : Système nerveux : Motoneurone et jonction neuromusculaire

Aides aux jeunes chercheurs post-doctorants

JHA Narendra, New York, Etats-Unis

Identification of modifier gene(s) of spinal muscular atrophy in model mice

SARDINA Francesca, Roma, Italie

Phenotypic evaluation of microtubules network as prognostic and predictive marker in response to spastin elevating approaches in Hereditary Spastic Paraplegia type 4

Financements de projets Tremplins

BOEYNAEMS Steven, Stanford, Etats-Unis

sPAM: Developing peptide-based drugs to target ataxin-2 in neuromuscular disease

Financements de projets

BOMONT Pascale, Montpellier, France

Analysis of the first phenotypical mouse model for Giant Axonal Neuropathy

BONDURAND Nadège, Paris, France

Role of ADAR1 and RNA editing in Schwann cells development and myelin maintenance

BRANCHEREAU Pascal, Bordeaux, France

Chloride-related inhibition in spinal motoneurons during the ALS disease progression in mouse model

CARRA Serena, Modena, Italie

Unraveling HSPB3 physiological functions to understand its implication in neuromuscular diseases

CASSEREAU Julien, Angers, France

Metabolomic exploration of dysregulated lipid metabolism in MFN2-related CMT2A

DUPUIS Luc, Strasbourg, France

Muscle contribution to FUS-associated ALS: genetic and mechanistic insights

FASSIER Coralie, Paris, France

Preventing motor neuron degeneration associated with spastin haploinsufficiency through TTLL-mediated tubulin polyglutamylation

FERRARO Elisabetta, Pisa, Italie

Metabolic reprogramming and activation of microglia in amyotrophic lateral sclerosis (ALS): a pivotal role for serotonin?

JORDANOVA Albena, Antwerpen, Belgique

Profiling the spatial RNA and protein signatures of peripheral neurons in YARS associated Charcot-Marie-Tooth neuropathies

LEVY-LAHAD Ephrat, Jerusalem, Israël

The motor neuron disease gene VPK1: a conditional VPK1 knockout mouse as a novel model for neuromuscular disease

LIEVENS Jean-Charles, Montpellier, France

Deciphering the beneficial effects of Sigma-1 receptor in amyotrophic lateral sclerosis

MONANI Umrao, New York, Etats-Unis

Investigating the cellular and molecular basis of muscle defects in spinal muscular atrophy

PENNUTO Maria, Padova, Italie

Targeting AR co-regulators to attenuate spinal and bulbar muscular atrophy

RIVAL Thomas, Marseille, France

Characterizing Charcot-Marie-Tooth disease-associated alleles of mitofusin with enhanced mitochondrial fusion activity and interfering with their neurotoxicity in vivo

TARESTE David, Paris, France

Core Molecular Mechanisms and Lipid Determinants of Mitofusin-mediated Mitochondrial Fusion

TAWK Marcel, Le Kremlin Bicêtre, France

Dissect the role of ADCY6 in arthrogryposis and peripheral myelination

VETTORI Andrea, Verona, Italie

Involvement of BMP signaling pathway in Hereditary Spastic Paraplegia: identification of new therapies based on in-vivo drug screening

VIERO Gabriella, Povo, Italie

Ribosome-based functions of the SMN protein: from fundamental biology to second-generation therapies for SMA

Commission : Cellules souches

Aides aux jeunes chercheurs post-doctorants

ALESSANDRINI Francesco, Chicago, Etats-Unis

Develop an iPSC-based platform for interrogating sporadic ALS disease proteome compartmentalization

DI GIROLAMO Daniela, Paris, France

Metabolic regulation of muscle stem cell fates in physiological and pathological conditions

GALLAY Laure, Genève, Suisse

Pivotal role of muscle stem cells in idiopathic inflammatory myopathies pathogenesis

SANDONÀ Martina, Roma, Italie

Study of FAPs-derived Extracellular vesicles ability to restore dystrophic muscle integrity upon systemic administration: a new piece of the puzzle

Financements de projets Tremplins

EVANO Brendan, Paris, France

Niche determinants of muscle stem cell in vivo dynamics

ORTEGA CANO Juan Alberto, Chicago, Etats-Unis

Defining the spinal cord matrisome to design more effective ALS models and treatments

PARFITT David, Leiden, Pays-Bas

Investigating cell-specific pathology in Huntington's disease and spinocerebellar ataxia iPSC-derived brain assembloids before and after treatment with targeted antisense oligonucleotides

Financements de projets

AIT-SI-ALI Slimane, Paris, France

Induced pluripotent stem cells-based strategies and disease modeling to unravel signaling-induced epigenetic networks in healthy and in Duchenne muscle dystrophic muscles: the TGFβ/Wnt regulated histone lysine methylome

AMTHOR Helge, Montigny-le-Bretonneux, France

The role of dystrophin in establishing the satellite cell niche

BIRCHMEIER Carmen, Berlin, Allemagne

Muscle stem cell maintenance and self-renewal: Prerequisites for a healthy muscle

BIRESSI Stefano, Povo, Italie

Novel satellite cell heterogeneity in healthy and pathological regeneration

ELVASSORE Nicola, Padova, Italie

Intravital 3D bioprinting approach for muscle stem cell delivery

FUKADA So-Ichiro, Osaka, Japon

Identification of factors inducing MuSC expansion from overloaded muscle

MAYEUF-LOUCHART Alicia, Lille, France

The circadian clock of muscle stem cells

MOREY Céline, Paris, France

Investigating the function of the FTX long non-coding RNA in the definition of spinal motoneuron identities

SEBILLE Stéphane, Poitiers, France

Functional characterization of muscle cells derived from healthy and DMD human induced Pluripotent Stem Cells

THORSTEINSDOTTIR Solveig, Lisbon, Portugal

Understanding the responses of fetal muscle stem cells and myoblasts to their niche in a mouse model for LAMA2-CMD

Commission : Thérapie Génique et/ou Cellulaire des Maladies Rares

Aides aux jeunes chercheurs post-doctorants

BOFFA Yolanda, Napoli, Italie

New gene therapy strategies for the treatment of gyrate atrophy of the choroid and retina (GACR), a progressive retinopathy due to the deficiency of the enzyme ornithine amino-transferase

RICHARD Elodie, Montpellier, France

Systemic gene therapy approach to treat Wolfram syndrome

TASFAOUT Hichem, Seattle, Etats-Unis

Expression of large dystrophins using Intein-mediated protein trans-splicing

Financements de projets Tremplins

INDRIERI Alessia, Pozzuoli, Italie

Therapeutic efficacy of miR-181a/b down regulation in inherited optic neuropathies

MALERBA Alberto, Egham, Royaume-Uni

Establishing a mouse model of disease to test combined antisense oligonucleotides and AAV systemic gene therapy as new treatment for Oculopharyngeal muscular dystrophy

Financements de projets

BETUING Sandrine, Paris, France

Efficient Gene and Sterol regulations by gene transfer for striatal protection in Huntington's Disease

BOYER Olivier, Rouen, France

CAAR-T cells: toward an antigen-specific immunotherapy for Immune Mediated Necrotizing Myopathies

DALKARA Deniz, Paris, France

Non viral gene editing for autosomal dominant retinitis pigmentosa

ERCEG Slaven, Valencia, Espagne

The cell therapeutic strategy for hereditary retinal dystrophies in small and large animals: MERTK associated Retinitis pigmentosa

HOVNANIAN Alain, Paris, France

Base editing-mediated correction of recurrent mutations in COL7A1 to treat RDEB

KEMALADEWI Dwi, Pittsburgh, Etats-Unis

Therapeutic genetics and disease modeling in LAMA2-CMD

LATTANZI Wanda, Roma, Italie

Personalized non-invasive nanotherapy of Crouzon syndrome through FGFR2 gene knock-down by recombinant human ferritin-based targeted siRNA delivery

ZIMMERMANN Valérie, Montpellier, France

Combinatorial treatment with gene and cell therapy for the treatment of SCID

Commission : Thérapie Pharmacologique des maladies neuromusculaires et Recherche Translationnelle

Aides aux jeunes chercheurs post-doctorants

DORT Junio, Montréal, Canada

Using specialized pro-resolving mediators to treat Duchenne muscular dystrophy

Financements de projets Tremplins

BEN-ELIEZER Noam, Tel Aviv, Israël

A quantitative MRI platform for simultaneous and automatic quantification of fat infiltration and T2 relaxation times in neuromuscular disorders

DUARTE Sonia, Coimbra, Portugal

Repurposing FDA-approved Drugs as microRNA-specific modifiers towards a new and promising therapeutic approach for Machado-Joseph disease/spinocerebellar ataxia type 3 (MJD/SCA3)

SACCHETTO Roberta, Legnaro, Italie

Use of small molecules to correct defective SERCA Proteins causing BRowdy disease

SNOWDEN Stuart, London, Royaume-Uni

Identifying metabolic signatures of pathology in muscle and blood to identify biomarkers of oculopharyngeal muscular dystrophy

Financements de projets

BRYSON-RICHARDSON Robert, Melbourne, Australie

Pre-clinical drug screen for LAMA2 congenital muscular dystrophy

BURATTI Emanuele, Trieste, Italie

Novel therapeutic targets derived from modulation of RNA metabolism in late onset Pompe disease

CIRAK Sebahattin, Köln, Allemagne

Biomarker discovery and validation for LGMD2I/FKRP-related muscular dystrophy

DE LUCA Annamaria, Bari, Italie

Selected growth hormone secretagogues (GHS) with wide mechanism of actions as disease modifiers in Duchenne Muscular Dystrophy: a multidisciplinary proof-of-concept study in mdx mouse model

DEVAUX Jérôme, Montpellier, France

Pathogenic mechanisms of anti-neurofascin 155 IgG4: Role of bivalency and novel therapeutic approaches

DORCHIES Olivier, Genève, Suisse

Duchenne muscular dystrophy: Phenotyping and validation of better murine models for improving preclinical research and clinical translation

HORNSTEIN Eran, Rehovot, Israël

Rigorous, integrated miRNA-DNA plasma biomarkers for amyotrophic lateral sclerosis

LAZARIDIS Konstantinos, Athens, Grèce

Preclinical study of antigen-specific tolerance induction for the treatment of myasthenia gravis

MESSINA Graziella, Milano, Italie

Drug repurposing of ERK inhibitors to target the transcription factor Nfix in dystrophic muscles: development of a new proof-of-concept study to hinder Muscular Dystrophies

PHYLACTOU Leonidas, Nicosia, Chypre

Development of serum miRNAs as biomarkers for the prognosis of Myotonic Dystrophy type I

VAN DEN BOSCH Ludo, Leuven, Belgique

Identification of therapies targeting lipid metabolism & myelination for Charcot-Marie-Tooth disease type 1A using patient derived Schwann cells

Commission : Médicale

Financements de projets

DIAZ-MANEIRA Jordi, New Castle, Royaume-Uni

Implementation of an artificial intelligence module on the web-based digital platform MyoShare for guiding the diagnosis of muscle diseases

LEOTARD Antoine, Garches, France

Early identification of respiratory exacerbations using NIV device monitoring in slowly progressive neuromuscular disorders

LETELLIER Guy, Nantes, France

Efficacy and safety evaluation of the ExoMS exoskeleton: an innovative impressed 3D upper limb assistive device for children with neuromuscular diseases. A Single-Case Experimental Design trial using Goal Attainment Scaling

SACCONI Sabrina, Nice, France

New clinical outcome measures to evaluate non-ambulant FSHD patients

TASCA Giorgio, Roma, Italie

Natural history of distal and myofibrillar myopathies assessed by clinical and technological outcome measures: a new toolbox for clinical trials

VUILLEROT Carole, Bron, France

Development and validation study of a Motor Function Measure digitalized playful completion modules

Appel d'offres Doctorants

- ALMYRE Claire**, Bordeaux, France
Investigation in human cells and mice of FDA-approved chemicals showing beneficial effects in yeast models of distinct metabolic disorders with a nuclear or mitochondrial genetic origin
- AMMAR Nourhene**, Toulouse, France
Live-imaging of adult muscle stem cell activation in Drosophila
- BEAUJARD Bettina**, Paris, France
L'annonce diagnostique d'une maladie neuromusculaire à l'âge adulte. Retentissements psychologiques des processus communicationnels entre médecin et patient
- BOUCHARD Laetitia**, Marseille, France
Evaluation of FGF10 as a target for promoting heart regeneration in Lmna-related dilated cardiomyopathy
- CHALUMEAU Anne**, Paris, France
Development of a universal prime editing approach to β -hemoglobinopathies
- CHEVREAU Robert**, Montpellier, France
Role of Hippo/YAP pathway in maintenance and differentiation of adult spinal cord stem cells: a potential cellular source for spinal cord degenerative pathologies
- CLAEYSSEN Charlotte**, Loos, France
O-GlcNAcylation and its interplay with phosphorylation: which impact on the cytoarchitecture and the function of skeletal muscle?
- D'AGATA Léna**, Paris, France
Annexins as genetic modifiers of human muscular dystrophies
- DA CUNHA Dylan**, Montpellier, France
Unravelling molecular mechanisms of Exon Junction Complex (EJC) in splicing regulation of the DMD gene
- DEBAR Louis**, Aubière, France
Disease-causing mutations in human mitochondrial DNA replication factors: A single-molecule study
- DELAFENETRE Arnaud**, Poitiers, France
Functional characterization of muscle cells derived from healthy and DMD human induced Pluripotent Stem Cells: focus on calcium release channels
- HOUQUES Chloé**, Montpellier, France
Combinatorial treatment with gene and cell therapy for the treatment of SCID
- KOBON Cassandra**, Paris, France
Control of myoblast cell-cell fusion - Crucial role of actin-based structures
- KOTAICH Farah**, Montpellier, France
Neurofilaments in health and Charcot-Marie-Tooth diseases
- LAPENDRY Audrey**, Lyon, France
Amino acid metabolism and alternative splicing
- MATHIEU Maxime**, Toulouse, France
Characterization and functional role of adipose-derived fibro-adipogenic progenitors (AD-FAPs) in skeletal muscle regeneration
- MATOUK Meriem**, Montigny-Le-Bretonneux, France
Elucidate the role of dystrophin in the muscle stem cell niche
- MOUNIER Coline**, Paris, France
Cell-Type-Specific Gene and Sterol Profiling following CYP46A1 gene therapy in Huntington's disease
- NAIT-SAIDI Rima**, Montpellier, France
Role of oxidative stress and endoplasmic reticulum stress in OPMD: molecular mechanisms and pharmacological treatments
- VAHDAT Juliette**, Marseille, France
Exploring ventricular conduction system structure and function in DMD mouse models
- VARUK Olena**, Marseille, France
Analysis of an allelic series of mitofusin MARF mimicking mutations associated to Charcot-Marie-Tooth type 2A disease in the Drosophila motor neurons
- YILDIRIM Zuleyha**, Illkirch, France
Role of PARP3 in the differentiation of muscle stem cells in mice
- ZERAD Lisa**, Paris, France
Role of ADAR1 and RNA editing in Schwann cells development and myelin maintenance

Appel d'offres thématique ARN médicament et cible

NAMY Olivier, Orsay, France

Translectin: A new termination codon readthrough inducer to expand therapeutics choices for nonsense genetic diseases

D'AMATI Giulia, Roma, Italie

Stabilization of tRNAs as a therapeutic strategy for diseases due to mutations in mt-tRNAs

Appel d'offres thématique Recherche Dystrophie Myotonique

FURLING Denis, Paris, France

Tricyclo-DNA antisense oligonucleotide treatment for Myotonic Dystrophy

Partenariats institutionnels

KIM Minchul, Montpellier, 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

Partenariats associatifs

Soutien à l'association Cure CMD

ZITO Ester, Torrance, Etats-Unis

Ablation of the maladaptive ER stress response improves altered calcium handling and restores diaphragm function in SELENON knock-out mice

Soutien au Fond de Dotation IFCAH

BABOT Ruiz, Dresden, Allemagne

Generation of human steroidproducing organoids: a new approach towards a treatment for CAH

Soutien à l'association Retina France

ROGER Jérôme, Saclay, France

Expression de CRX par thérapie génique visant des modèles de dystrophies rétiniennes indépendantes de mutations dans CRX

Soutien à l'association Vaincre la Mucoviscidose (VLM)

CADARS Magali, Montpellier, France

ONB-CFTR : Stratégie OligoNucléotide Bloqueur : un outil spécifique pour stabiliser ou corriger les transcrits CFTR

LEJEUNE Fabrice, Lille, France

Optimisation de la restauration de la fonction CFTR in vivo à partir d'un gène porteur d'une mutation non-sens par inhibition du NMD et activation de la translecture

Soutien à l'IRME, Paris, France

HUGNOT Jean-Philippe, Montpellier, France

Aging and Controlling the Fate of Human Spinal Cord stem cells

Projets soumis en dehors du calendrier AO ou sortis de l'AO pour des raisons de PI

SOLE Guilhem, Bordeaux, France

Evaluation of the effectiveness of COVID-19 vaccination by seeking neutralizing antibodies in patients with neuromuscular disease with severe amyotrophy

Projets Ignition

BÜNING Hildegard, Hannover, Allemagne

Rational-designed enveloped AAV: a novel all-in-one gene transfer system

DECRESSAC Michael, La Tronche, France

Towards gene replacement tHERapy for severe mitochondrial disease

MUSARO Antonio, Roma, Italie

Understanding the contribution of molecular mediators of inflammation to DMD progression and implications for therapy: studying the impact of IL-6 transsignaling on dystrophic muscle stability

NICOLE Sophie, Montpellier, France

Nav1.4 activators for a correction of SCN4A-related muscle weaknesses

PALACIOS Daniela, Roma, Italie

Functionalized nanoparticles for targeted genome editing in Duchenne Muscular Dystrophy

VISCOMI Carlo, Padova, Italie

Harnessing mitophagy to treat mitochondrial myopathies

ACTIONS STRATEGIQUES

Projets stratégiques

AGBULUT Onnik, Paris, France

Innovative bio-inspired cell and tissue models of genetic dilated cardiomyopathy for disease modelling and drug discovery

DUMONCEAUX Julie, London, Royaume-Uni

DUX4 in FSHD: pathophysiology and therapeutic approaches

GUEDAT Philippe, Nantes, France

A double-blind, randomized, placebo-controlled, parallel-group phase 2 study to evaluate the safety, efficacy, and pharmacokinetics of IFB-088 in patients with a Charcot-Marie-Tooth disease caused by either a duplication of PMP22 gene (CMT1A), a point mutation in the PMP22 gene (CMT1E) or a mutation in MPZ gene (CMT1B)

LAPORTE Jocelyn, Illkirch, France

Pathophysiology and therapeutic proof-of-concepts for congenital myopathies

MUNTONI Francesco, London, Royaume-Uni

Advances in oligonucleotide-mediated exon skipping for DMD and related disorders - WP3 - Natural history extension

MYOPHARM Program, Evry, France

Collaborative program for a systematic, mechanistic and pharmacologic approach of rare neuromuscular diseases

POURQUIE Olivier, Boston, Etats-Unis

Toward cell therapy for Duchenne Muscular Dystrophy: characterization of regenerative potential of hIPS-derived Pax7+ cells

PROCACCIO Vincent, Angers, France

Identifying candidate drugs in mitochondrial cardiomyopathies: From Mouse to Human

Pôles stratégiques

LEVY Nicolas, Marseille, France

Translational Research in Marseille: towards Therapeutic Development for Rare Diseases
Modelisation and Therapeutic Approaches for Rare Diseases

RELAIX Frédéric, Créteil, France

An integrated translational program for neuromuscular disorders

SCHAEFFER Laurent, Lyon, France

MyoNeurALP2, The Research Network dedicated to Neuromuscular Disorders in Rhone Alpes Auvergne

Structures stratégiques

- FONDATION MALADIES RARES**, Paris, France
Appel à projet auprès des filières de santé maladies rares
- FONDATION MALADIES RARES**, Paris, France
Subvention 2021
- GENOPOLE**, Evry, France
Participation financière de l'AFM au budget 2021 du GIP GENOPOLE

Outils stratégiques

- BASSEZ Guillaume**, Créteil, France
iDM-scope: the international French-Quebec myotonic dystrophy registry
- DESGUERRE Isabelle**, Paris, France
A national clinical (and genetic) database for dystrophinopathies (DYS Registry)
- LAFORET Pascal**, Garches, France
Sarcoglycanopathy French Registry
- MALFATTI Edoardo**, Créteil, France
French national registry for calpainopathies
- SACCONI Sabrina**, Nice, France
French National Registry for FacioScapuloHumeral muscular Dystrophy (FSHD)
- SACCONI Sabrina**, Nice, France
Resolve FSHD: clinical trial readiness to solve barriers to drug development in FSHD
- WAHBI Karim**, Paris, France
Cardiomyopathy database

AUTRES ACTIONS

Manifestations scientifiques (congrès, colloques)

- BERRIH-AKNIN Sonia**, Paris, France
ISNI Congress 2021
- BRYANT Kathryn**, Columbus, Etats-Unis
International Limb Girdle Muscular Dystrophy Conference
- GOMEZ Ana Maria**, Châtenay-Malabry, France
Calcium Signaling and Excitation-Contraction Coupling
- JAMAR Gaëlle**, Paris, France
ESGCT and national societies of Europe Virtual congress
- KINOSHITA June**, Lexington, Etats-Unis
28th Annual FSHD Society International Research Congress
- TEDESCO Francesco Saverio**, London, Royaume-Uni
Society for Muscle Biology (SMB) 2021 Frontiers in Myogenesis Conference "Skeletal muscle: development, regeneration and disease"

Plateforme non stratégique

- CARRE Monique**, Mézilles, France
Financement CEDS