

LISTE DES PROJETS ET AIDES AUX JEUNES CHERCHEURS FINANCES PAR L'AFM-TELETHON EN 2015

POLITIQUE D'APPEL D'OFFRES

Projets soumis à l'AO

Commission : Myologie fondamentale

Aides aux jeunes chercheurs

PESSINA Patrizia, BARCELONE, Espagne

Analysis of the cellular mechanisms underlying fibrosis development in dystrophic muscle

ROBERT-PAGANIN Julien, PARIS, France

Insights for novel treatment of HCM: structural studies of beta cardiac myosin to understand the impact of HCM mutations and the mechanism of modulators of force generation

Financements de projets Tremplins

COPPEE Frédérique, MONS, Belgique

Functional study of the DUX4c double homeodomain protein and its involvement in human muscle regeneration

LE GOFF Carine, PARIS, France

Role of ADAMTSL2 in skeletal muscle development

MEILHAC Ségolène, PARIS, France

Role of primary cilia in the growth of the cardiac muscle

VON HOFSTEN Jonas, UMEÅ, Suède

Induced regeneration of unique muscle identities

Financements de projets

ARMAND Anne-Sophie, PARIS, France

Regulation of dystrophin expression by the NFAT transcription factors during skeletal myogenesis

BLAAUW Bert, PADOUE, Italie

The role of S6K in the regulation of skeletal muscle mass and function

BORYCKI Anne-Gaëlle, SHEFFIELD, Royaume-Uni

Role of sonic hedgehog signalling in adult skeletal muscles

BOUTER Anthony, PESSAC, France

Annexins in membrane repair of human skeletal muscle

BUSCHBECK Marcus, BARCELONE, Espagne

How does the histone variant macroH2A regulate muscle metabolism in health and disease?

CABELLO-VERRUGIO Claudio Alejandro, SANTIAGO, Chili

Anti-atrophic role of Angiotensin 1-7 on skeletal muscle

CASAS François, MONTPELLIER, France

Influence of mitochondrial homeostasis in the control of the regeneration and skeletal muscle mass

CHAZAUD Bénédicte, VILLEURBANNE, France

Molecular coupling between myogenesis and angiogenesis

CHRETIEN Fabrice, PARIS, France

The role of CXCL12 and the paradigm of chemokine immobilization in muscle regeneration

DE SANTA BARBARA Pascal, MONTPELLIER, France

Function of double-strain RNA-Binding Protein and Collagen during digestive smooth muscle cell plasticity

DEFOSSEZ Pierre-Antoine, PARIS, France

Epigenetics of muscle cell differentiation: the role of DNA methylation and methyl-DNA-binding proteins

DUPREZ Delphine, PARIS, France

Link between and the CXCL12//CXCR7 signalling pathway in the connective-tissue-mediated control of muscle formation

GRAZIANI Andrea, NOVARA, Italie

Ghrelin peptides as novel anti-atrophic factors acting directly in the skeletal muscle: identification of their molecular mechanisms and of their role in cancer cachexia

HEIDMANN Thierry, VILLEJUIF, France

Role of the 'captured' fusogenic syncytin genes in muscle formation, regeneration and pathology

MAMMUCARI Cristina, PADOUE, Italie

Modulation of mitochondrial calcium signalling to combat skeletal muscle atrophy

METZGER Daniel, ILLKIRCH, France

Characterisation of signalling pathways controlled by androgens, glucocorticoids and miRNAs in skeletal muscles and identification of new targets for muscle diseases

MUNOZ-CANOVES Pura, BARCELONE, Espagne

Role and mechanisms of action a new regulator of skeletal muscle growth and wasting: sestrin

RONJAT Michel, GRENOBLE, France

Role of domain A of the skeletal muscle dihydropyridine receptor in excitation contraction coupling

ROSA Frederic, PARIS, France

Muscle maturation, myofibril assembly regulation and RNA binding proteins

SCHAEFFER Laurent, LYON, France

Histone variant epigenetic player H2A.Z and muscle plasticity

SOTIROPOULOS Athanassia, PARIS, France

Role of Serum Response Factor as a mediator of mechanotransduction in skeletal muscle

TAJBAKSHH Shahragim, PARIS, France

Developmental origins and genetic regulation of oesophagus skeletal muscles

VOLK Talila, REHOVOT, Israël

Mechanobiology of myonuclei in health and disease

ZERVAS Christos, ATHENE, Grèce

Function and Regulation of Parvin and Parvin/ILK interaction in Drosophila muscle attachment sites

Commission : Bases Moléculaires et Physiopathologie des Dystrophies Musculaires

Aides aux jeunes chercheurs

DEWULF Melissa, PARIS, France

Functional analysis of Cav3 mutations in muscular dystrophy diseases

KEMALADEWI Dwi, TORONTO, Canada

Elucidating the role of polyamine in laminin-deficient congenital muscular dystrophy

Financements de projets Tremplins

COLUSSI Claudia, ROME, Italie

Linking chromatin structure to dystrophic cardiomyopathy: role of Nucleoporin 153

JAGLA Krzysztof, CLERMONT-FERRAND, France

New Drosophila model of Myotonic Dystrophy type 1: Phenotypic and transcriptomic characterization of functional and structural cardiac defects

STRAPPAZZON Flavie, ROME, Italie

Recovery of dystrophic phenotype by modulation of Ambra1, a novel primer of mitophagy

VALLEJO Ainara, SAN SEBASTIAN, Espagne

Targeting calcium handling proteins in LGMD2A muscular dystrophy

Financements de projets

AUBOEUF Didier, LYON, France

Regulation and function of alternative splicing during muscle differentiation: an integrated view

BASSEREAU Patricia, PARIS, France

Molecular mechanisms of caveolin sorting in membranes. Role in muscular dystrophies

BERGHELLA Libera, ROME, Italie

Molecular mechanisms for neuromuscular junction (NMJ) disruption and reduced mitochondrial function in Duchenne Muscular Dystrophy

- BODEGA Béatrice**, MILAN, Italie
Epigenetic role for DNA repeats and ncRNAs in FSHD manifestation
- BROWN Susan**, CAMDEN TOWN, Royaume Uni
Investigations into why long term over-expression of LARGE is deleterious in dystrophic muscle
- CANCELA José-Manuel**, ORSAY, France
Characterization of a new model of mdx mice deficient in the enzyme CD38: towards the protective role on the Ca²⁺ homeostasis deregulation
- CHARLET-BERGUERAND Nicolas**, ILLKIRCH, France
Novel animal models of Myotonic Dystrophies
- COGNARD Christian**, POITIERS, France
Local stretch-activated events in mdx dilated cardiomyopathy
- DONATO Rosario**, PEROUSE, Italie
Molecular modulators of muscle remodeling in Duchenne Muscular Dystrophy: Role of RAGE
- DURBEEJ Madeleine**, LUND, Suède
Characterization of metabolic alterations in MDC1A and evaluation of potential therapies to improve muscle structure and function
- ESTEVEZ Raul**, BARCELONE, Espagne
Development and characterization of a zebrafish model of myotonia congenital
- FLANIGAN Kevin**, COLUMBUS – OHIO, Etats-Unis
Alternate translational initiation and amelioration of phenotype in the DMD gene
- FRIGUET Bertrand**, PARIS, France
Protein Damage and Repair in Muscular Dystrophies
- GAILLY Philippe**, BRUXELLES, Belgique
Role of TRPV2 and TRPV4 ion channels in normal and dystrophic muscle
- GOMES-PEREIRA Mario**, PARIS, France
Synaptic and cytoskeleton dysfunction in DM1 transgenic mice
- JASMIN Bernard**, OTTAWA, Canada
Translational Regulation of Utrophin A as a Novel Therapeutic Strategy for DMD
- LE RUMEUR Elisabeth**, RENNES, France
Structure and interactions of dystrophin and their modifications in Becker muscular dystrophy
- MARI Bernard**, VALBONNE, France
Regulation of human muscle progenitors fibrotic potential by micrnas
- MARTELLI Fabio**, SAN DONATO MILANESE, Italie
microRNA function and use as biomarkers in Myotonic Dystrophy type 1
- MENEVERI Raffaella**, MONZA, Italie
Post-genomic approaches to decipher the pathogenesis of Facioscapulohumeral dystrophy (FSHD)
- MERONI Germana**, TRIESTE, Italie
Structure and function of TRIM32, the ubiquitin ligase mutated in Limb Girdle Muscular Dystrophy 2H
- MILLER Jeffrey Boone**, BOSTON, Etats-Unis
Pathogenesis of congenital muscular dystrophy Type 1A (laminin-alpha-2-deficiency)
- MORALES Fernando**, SAN JOSÉ, Costa Rica
Myotonic dystrophy type 1: analyzing how somatic heterogeneity contributes to the age of onset and progression of the disease
- MOREL Véronique**, LYON, France
Drosophila nesprin-1 a model of Emery Dreifuss muscular dystrophy. Contribution to muscle function
- PLAISIER Emmanuelle**, PARIS, France
Pathogenic role of basement membrane defects and endoplasmic reticulum stress in the myopathy related to COL4A1 HANAC mutations
- RUBINSTEIN Eric**, VILLEJUIF, France
Control of muscle cells fusion by tetraspanins: mechanisms and potential implications for myotonic dystrophies
- RUEGG Markus**, BÂLE, Suisse
Repair of merosin-deficient congenital muscular dystrophy (MDC1A) with synthetic linker proteins
- SEUTIN Vincent**, LIÈGE, Belgique
Excessive daytime sleepiness in Myotonic Dystrophy type 1 patients: an unsolved clinical target requiring a translational research approach
- SORRENTINO Vincenzo**, SIENNE, Italie
Role of obscurin in skeletal muscle function and in muscular dystrophy
- TUFFERY-GIRAUD Sylvie**, MONTPELLIER, France
Defining the trans-acting factors that regulate normal DMD pre-messenger RNA splicing: a combination of RNA interference and targeted RNA-seq approaches

- TUPLER Rossella Ginevra**, MODENE, Italie
Whole exome sequencing to dissect genetic complexity in Fasciocalpulo humeral dystrophy
- ZAMMIT Peter**, LONDRES, Royaume Uni
Role of RET in muscle stem cell function and FSHD pathology
- ZINN-JUSTIN Sophie**, GIF-SUR-YVETTE, France
Structural analysis of protein complexes mutated in Emery-Dreifuss Muscular Dystrophy

Commission : Bases moléculaires et physiopathologie des autres maladies neuromusculaires

Aides aux jeunes chercheurs

- METODIEV Metodi**, PARIS, France
Identification of novel genes of mitochondrial translation deficiencies in human

Financements de projets Tremplins

- GOMES José-Eduardo**, BORDEAUX, France
Investigating the neuro-muscular effects of Adenylosuccinate Lyase (ADSL) deficiency using *C. elegans* as model organism
- KASTANIOTIS Alexander**, OULLU, Finlande
A cryptic cache of mitochondrial disease genes

Financements de projets

- AGBULUT Onnik**, PARIS, France
Exploring cardiac pathogenesis associated to desmin mutations: a novel approach based on AAV-mediated expression
- ALLARD Bruno**, VILLEURBANNE, France
Physiopathology of voltage-activated Ca²⁺ influx in normal and diseased skeletal muscle
- AUTHIER François Jérôme**, CRETEIL, France
Myofiber expression of HLA-DR in dysimmune myopathies
- BATONNET-PICHON Sabrina**, PARIS, France
Desminopathies: mutations in desmin, what impact on cell-cell or cell-matrix adhesion ?
- BENDAHHOU Said**, NICE, France
Pathomechanisms in Andersen's syndrome: insights into excitable and non excitable tissues
- BOYER Olivier**, ROUEN, France
Necrotizing myopathies: pathogenic role of autoantibodies and development of new immuno-assays
- CARRA Serena**, MODENE, Italie
Identification of HSPB3 mutations in myopathic patients: understanding the mechanisms of disease
- DEVAUX Jérôme**, MARSEILLE, France
Identification of Novel Biomarkers and Treatments for Inflammatory Demyelinating Neuropathies
- DUFOUR Eric**, TAMPERE, Finlande
Altering complex I response to OXPHOS dysfunctions; a new tool to combat mitochondrial diseases
- FERNANDEZ-VIZARRA Erika**, CAMBRIDGE, Royaume-Uni
Patho-physiology of mitochondrial respiratory chain complex III assembly
- GOILLOT Evelyne**, LYON, France
Regulation of protein aggregation through NBR1 and p62 phosphorylation: implication for protein aggregate myopathies
- ILLA Isabel**, BARCELONE, Espagne
Danger signals promoting innate immunity in dermatomyositis
- JACQUEMOND Vincent**, VILLEURBANNE, France
Phosphoinositides and Ca²⁺ signaling in normal and diseased skeletal muscle
- LOMBES Anne**, PARIS, France
Pathophysiology of human mitochondrial disorders: searching for common treatable mechanisms
- MARTY Isabelle**, LA TRONCHE, France
The calcium release complex: targeting and maintenance in normal and pathological situation
- MITRANI-ROSENBAUM Stella**, JERUSALEM, Israël
Characterization of the microRNA profile of GNE Myopathy

- OTTENHEIJM Coen**, AMSTERDAM, Pays-Bas
Why Are Muscles Weak In Nemaline Myopathy And How Can We Treat It?
- PENDE Mario**, PARIS, France
Autophagic vacuolar myopathies and the functional role of class III PI3K
- RAVENSCROFT Gianina**, NEDLANDS, Australie
Neuromuscular disease presenting in utero - gene discovery and pathobiology
- ROSSIGNOL Rodrigue**, BORDEAUX, France
Adaptative pathways of energy transduction in Mitochondrial diseases: implication for therapy
- VERMOT Julien**, ILLKIRCH, France
Generation of desminopathy models through loss and gain of function approaches in zebrafish
- WALLGREN-PETTERSSON Carina**, HELSINKI, Finlande
Nemaline myopathy and related disorders: diagnostic methods, disease-gene identification, pathogenesis and genotype-phenotype correlations
- WALLGREN-PETTERSSON Carina**, HELSINKI, Finlande
Nemaline myopathy and related disorders: molecular genetics, pathogenesis and development of RNA-based therapy

Commission : Système nerveux : Motoneurone et jonction neuro-musculaire

Aides aux jeunes chercheurs

- CIURA Sorana**, PARIS, France
Physiological analysis of C9orf72 depletion, a major genetic cause in ALS using zebrafish models
- COQUE Emmanuelle**, MONTPELLIER, France
The contribution of effector immunity in the pathophysiology of ALS
- JOASSARD Olivier**, OTTAWA, Canada
HuR and AU-rich elements regulate the induction of AChR mRNAs after skeletal muscle denervation
- STREPPA Laura**, LYON, France
Biomechanical study of neuromuscular junction and its impact on myopathies
- TORRES-BENITO Laura**, COLOGNE, Allemagne
Deregulated Calcium Signalling and Homeostasis in Spinal Muscular Atrophy

Financements de projets

- BESSEREAU Jean-Louis**, VILLEURBANNE, France
Control of acetylcholine receptor expression by novel mechanisms identified at the C. elegans neuromuscular junction
- BOMONT Pascale**, MONTPELLIER, France
Development of in vivo model for Giant Axonal Neuropathy
- BRITES Pedro**, PORTO, Portugal
Establishing the role and function of plasmalogens in neurons and neuron-muscle synapses
- BURGO Andrea**, EVRY, France
Understanding the molecular bases of the axonopathy in hereditary spastic paraplegias
- CHARLET-BERGUERAND Nicolas**, ILLKIRCH, France
Role of C9ORF72 in amyotrophic lateral sclerosis & frontotemporal dementia
- CHRISTADOSS Premkumar**, GALVESTON, Etats-Unis
MuSK specific IgG4 B cells as biomarker for MuSK myasthenia gravis
- DUPUIS Luc**, STRASBOURG, France
Motor neuron loss triggered by defective nuclear import of RNA-binding proteins: mechanistic studies using FUS as a prototypical example
- GALINDO Máximo Ibo**, VALENCE, Espagne
Metabolic and functional characterization of clinically relevant Charcot-Marie-Tooth genotypes in a Drosophila model
- GALLI Thierry**, PARIS, France
Neuronal secretory mutants - characterization of neuromuscular junctions
- JORDANOVA Albena**, ANVERS, Belgique
Identification of disease mechanisms and therapeutic targets of CMT neuropathies in drosophila
- KABASHI Edor**, PARIS, France
Development of zebrafish models for C9orf72, the major genetic cause in Amyotrophic Lateral Sclerosis and related neurological diseases

- KREJCI Eric**, PARIS, France
A novel natural mutation in mouse ColQ: muscle and non-muscle alteration
- LAMOTTE D'INCAMPS Boris**, PARIS, France
Corelease of acetylcholine and glutamate by motoneurons
- LEGAY Claire**, PARIS, France
The Wnt binding domain in MuSK: role in neuromuscular junction formation and maintenance
- LEGAY Claire**, PARIS, France
Post-transcriptional control of Acetylcholine receptor mRNA; implications for neuromuscular diseases
- LEGENDRE Pascal**, PARIS, France
Microglia instruct fate of embryonic neurons at the onset of developmental cell death in the embryonic spinal cord in vivo
- MANUEL Marin**, PARIS, France
Is hyperexcitability protective or harmful in ALS?
- MILLER Ariel**, HAIFA, Israël
DNA methylation and gene expression profiling of monozygotic twins concordant and discordant for Myasthenia Gravis
- NICOLE Sophie**, PARIS, France
The sodium channel Nav 1,4 at the neuromuscular junction: role in congenital myasthenic syndromes
- NOBREGA Clevio**, COIMBRA, Portugal
Ataxin-2 as a new molecular target in Machado-Joseph disease: from translation regulation to disease alleviation
- POLETTI Angelo**, MILAN, Italie
Selective autophagic response to proteotoxicity in motorneurons and muscle of motor neuron diseases
- RODGERS David**, LEXINGTON, Etats Unis
Cavities in Choline Acetyltransferase and Neuromuscular Disorders
- SAOUDI Abdelhadi**, TOULOUSE, France
Investigation of Vav1 signalosome and analysis of its implication in myasthenia gravis
- SCAMPS Frédérique**, MONTPELLIER, France
Calcium activated chloride channels and motoneuron pathophysiology
- SCHAEFFER Laurent**, LYON, France
Physiopathology of a new congenital myasthenic syndrome caused by a deficit of agrin secretion by motoneurons
- SORARU Gianni**, PADOUE, Italie
Skeletal muscle: target tissue to cure Spinal and Bulbar Muscular Atrophy (SBMA)
- TALBOT Kevin**, OXFORD, Royaume-Uni
Novel Disease Mechanisms in Hereditary Neuropathy

Commission : Cellules souches

Aides aux jeunes chercheurs

- FRANCOIS Stéphanie**, MONZA, Italie
Unravelling differences between Satellite Cells and Mesoangioblasts: modulation of the Rho/ROCK pathway in the activation of Satellite Cells migration
- MAYEUF-LOUCHART Alicia**, LILLE, France
Role of Rev-erb in myogenic versus adipogenic cell fate decisions and homeostasis
- MITUTSOVA Violeta**, MONTPELLIER, France
Skeletal muscle stem cell cardiogenic and neurogenic differentiation: an in vitro and in vivo analysis
- STUELSATZ Pascal**, SEATTLE, Etats Unis
EOM satellite cells: high performance myo-engines for muscular dystrophy therapy
- WAHANE Shalaka**, MONTPELLIER, France
Phenotypic and stem cell properties of VEGFR3+ and Msx1+ cells in the normal and pathological spinal cord

Financements de projets Tremplins

- HSIAO Edward**, SAN FRANCISCO, Etats Unis
Human iPS cell derived muscle stem cells in musculoskeletal disease

QUATTROCELLI Mattia, LOUVAIN, Belgique

Assessment of biodistribution and regenerative potential of human bipotent progenitor cells in a murine model of muscular dystrophy

SACCONI Valentina, ROME, Italie

Soluble mediators of the functional interactions between fibro-adipogenic progenitors and satellite cells in the pathogenesis and treatment of Duchenne Muscular Dystrophy

Financements de projets

AIT-SI-ALI Slimane, PARIS, France

Epigenetic regulation of muscle stem cells: cooperation between Polycomb group/H3K27 methylation and H3K9 methylation pathways

AUDA-BOUCHER Gwenola, NANTES, France

Identification and characterization of the myogenic enhancing factors from foetal preadypocyte secretome

COMI Giacomo Pietro, MILAN, Italie

Optimized transplantation of hiPSC derived LEX+CXCR4+VLA4+ neural stem cells as a therapy for SMARD1

DEMENEIX Barbara, PARIS, France

Timing of thyroid hormone action during adult neurogenesis

HUGNOT Jean-Philippe, MONTPELLIER, France

Molecular and cellular characterization of the spinal cord stem cell niche activation by physical exercise and damage

IMBRIANO Carol, MODENE, Italie

NF-YA as a molecular switch with therapeutic potential in muscle regeneration

JARRIAULT Sophie, ILLKIRCH, France

Understanding direct reprogramming as a gateway to safely manipulate cell identity and efficiently re-direct differentiation for cellular therapy purposes

KELLY Robert, MARSEILLE, France

Investigation of the emergence of craniofacial muscle progenitor cells in pharyngeal mesoderm

KOISTINAHO Jari, KUOPIO, Finlande

Human Model of Neuromuscular Junctions in the Research of Amyotrophic Lateral Sclerosis

MAIRE Pascal, PARIS, France

Myogenic fate of satellite cells

MOREAU-GAUDRY François, BORDEAUX, France

Safety management of induced pluripotent stem cells (iPSCs) in regenerative medicine

MOUNIER Rémi, VILLEURBANNE, France

AMPK α 1 as a regulator of adult muscle stem cell fate choice

RICCHETTI Miria, PARIS, France

Repair of DNA breaks in mouse skeletal muscle stem cells: implications in muscle regeneration in the adult

SERRANO Antonio, BARCELONE, Espagne

Satellite cell regulation by IL-6 in muscle regeneration

TABTI Nacira, PARIS, France

Insights into the physiology of human iPSC-derived skeletal myocytes and model development for the study of hereditary Na⁺ channelopathies

Commission : Thérapie Génique et/ou Cellulaire des Maladies Rares

Aides aux jeunes chercheurs

CHAPPERT Pascal, PARIS, France

Cross-tolerance approaches for AAV-mediated muscle gene transfer

KHABOU Hanen, PARIS, France

Retinitis Pigmentosa: gene therapies to prevent and restore vision loss

SANCHEZ-DUFFHUES Gonzalo, LEYDE, Pays-Bas

Putting the brakes on fop: development of novel strategies to block heterotopic ossification

TORRES TORRONTERAS Javier, BARCELONE, Espagne

Long-term biosafety and efficiency pre-clinical studies of an adeno-associated liver targeted vector as a gene therapy strategy for mitochondrial neurogastrointestinal encephalomyopathy

VACCA Ophelie, ORSAY, France

AAV-Mediated Dystrophin-Dp71 Gene Therapy In The Central Nervous System

Financements de projets Tremplins

ATHANASOPOULOS Takis, WOLVERHAMPTON, Royaume Uni

Development of dual/triple RNA and protein transpliced AAV vectors to restore quasi/full length co-dystrophin variants to muscle

SALVETTI Anna, LYON, France

Deciphering AAV vector genome uncoating from in vitro physical properties measured at the single particle level

Financements de projets

BARDONI Barbara, VALBONNE, France

Destabilizing FMR1 mRNA as a therapeutic strategy to treat FXTAS

BELAYEW Alexandra, MONS, Belgique

Evaluation of DUX4 silencing tools in mice

DEGLON Nicole, LAUSANNE, Suisse

In vitro and in vivo gene editing using viral delivered CRISPR system for Huntington's disease

GONCALVES Manuel A., LEYDE, Pays-Bas

High-capacity adenovectors for homology-directed correction of dystrophin-defective myogenic cells

LOPEZ Bernard, VILLEJUIF, France

Stimulating homologous recombination for gene correction of monogenic diseases

MECHALI Marcel, MONTPELLIER, France

Replication Origins containing Episomes for Gene Therapy

NOWAK Kristen, PERTH, Australie

ACTA1 congenital myopathies: evaluating viral and gene therapy

RECCHIA Alessandra, MODENE, Italie

Development of a new tool for gene therapy approach for autosomal dominant retinitis pigmentosa

TAYLOR Naomi, MONTPELLIER, France

Intrathymic hematopoietic stem cell transplantation for the correction of severe combined immunodeficiency

VANDENDRIESCHE Thierry, BRUXELLES, Belgique

Gene therapy for hereditary muscle diseases using novel AAV immune stealth nanotechnology

Commission : Thérapie Pharmacologique des maladies neuromusculaires et Recherche Translationnelle

Aides aux jeunes chercheurs

DURAND Sebastien, LYON, France

Characterization of INT6/EIF3E functions during Nonsense Mediated Decay (NMD) and development of new NMD inhibitors

JIA Jieshuang, LILLE, France

Characterization of new NMD inhibitors and/or activators of readthrough to correct premature termination codons in genetic diseases

MADARO Luca, ROME, Italie

Correction of the epigenetic landscape in dystrophic macrophages and satellite cells by HDAC blockade

PROKHOROVA Irina, ILLKIRCH, France

Structural basis for stop-codon read-through therapies on the eukaryotic ribosome

YUSEIN-MYASHKOVA Shazie, ANVERS, Belgique

A chemical genetic screen for candidate drugs rescuing CMT-associated phenotypes in Drosophila

Financements de projets Tremplins

GONDIN Julien, MARSEILLE, France

Combined MRI and 31P-MRS investigations of tyrosine supplementation in two murine models of nemaline myopathy with actin mutation

HATHOUT Yetrib, WASHINGTON, Etats Unis

Novel Tools to Monitor Disease Progression and Response to Therapy in Duchenne Muscular Dystrophy

HOLMBERG Johan, LUND, Suède

Circulating microRNAs: non-invasive biomarkers for congenital muscular dystrophy type 1A

TISO Natascia, PADOUE, Italie

POLYGON: POLG-related diseases: mutation analysis and drug screening in a zebrafish-based system

Financements de projets

BAAS Frank, AMSTERDAM, Pays-Bas

Inhibition of synthesis of terminal Complement Components as therapeutic strategy for peripheral neuropathies

BLOT Stéphane, MAISONS-ALFORT, France

Longitudinal cardiac, respiratory and locomotor follow-up of GRMD with production of dedicated biomarkers

DESAPHY Jean-François, BARI, Italie

Riluzole, lubeluzole and benzothiazolamine derivatives as new potent antimyotonic drugs

DORCHIES Olivier, GENEVE, Suisse

Enhancing estrogenic signalling to fight devastating muscular dystrophies: Mechanisms of action and repurposing estrogenic drugs approved for Human use

GIRARD Christian, PARIS, France

New nonsense-mediated mRNA decay (NMD) inhibitor molecules

LOCHMULLER Hanns, NEWCASTLE-UPON-TYNE, Royaume Uni

Coordination of the global patient registries for neuromuscular disorders

LOSSOS Alexander, JERUSALEM, Israël

Testing small molecules, glycogen autophagy and microtubule transport as treatments of glycogenosis

MARCHAND-PAUVERT Véronique, PARIS, France

Electrophysiological biomarkers of spinal neural activity in amyotrophic lateral sclerosis

PHYLACTOU Leonidas, NICOSIE, Chypre

Developing miRNA serum-based biomarkers for Myotonic Dystrophy type 1

PREVITALI Stefano Carlo, MILAN, Italie

Modulation of Jab1/p27 levels to rescue peripheral neuropathy and muscular dystrophy in Congenital Muscular Dystrophy type 1A

PURI Pier Lorenzo, ROME, Italie

HDAC/miR-regulated SWI/SNF sub-unit exchange & stage-specific response to HDAC inhibitors in dystrophic muscles

ROSSI Daniela Maria, PAVIE, Italie

Assessing the in vivo efficacy of peptide therapeutics towards the progression of spinal muscular atrophy

SANDONÀ Dorianna, PADOUE, Italie

Small molecule-based therapy for sarcoglycanopathies. Assessment of efficacy and tolerability in novel animal models

SPITALI Pietro, LEYDE, Pays-Bas

Identification of blood derived transcriptomic biomarkers for Duchenne muscular dystrophy

Commission : Médicale

Aides aux jeunes chercheurs

VEYTIZOU Julien, GRENOBLE, France

Spinal Muscular Atrophy (SMA) patients' evaluation using the Motor Function Measure (MFM-32) combined with low-cost innovative technology for improve the measurement quality

Financements de projets

- ALLENBACH Yves**, PARIS, France
Diagnostic Accuracy of Whole body Magnetic Resonance Imagery in Inflammatory Myopathies 2
- ANGEARD Nathalie**, PARIS, France
Social cognition and executive functions in childhood DM1
- COSTEDOAT-CHALUMEAU Nathalie**, PARIS, France
Muscular diseases and pregnancy
- GAGNON Cynthia**, JONQUIERE-QUEBEC, Canada
Selection and validation of clinical outcomes and related outcome measures in myotonic dystrophy type 1
- NORDEZ Antoine**, NANTES, France
Innovative tools to assess muscle function of healthy and pathological subjects
- PEPIN Jean-Louis**, LA TRONCHE, France
Sleep breathing disorders, a main Trigger for cardiac ARrhythmias in type I myotonic dystrophy?

Appel d'Offres Doctorants

- BABSKI Hélène**, MONTPELLIER, France
Getting wired up for locomotion: the premotor interneuron/motoneuron connection in Drosophila
- BERGUA Cécile**, ROUEN, France
Understanding the mechanisms of necrotizing autoimmune myopathies: pathogenic role of auto-antibodies in a mouse model and identification of new auto-antigens in patients
- COQUE Emmanuelle**, MONTPELLIER, France
The contribution of effector immunity in the pathophysiology of ALS
- DAHER Marie Thérèse**, PARIS, France
Bcl11b/CTIP2, a newly identified transcriptional repressor: its role in cardiac hypertrophy and commitment of cardiac stem cells
- DEWULF Melissa**, PARIS, France
Functional analysis of Cav3 mutations in muscular dystrophy diseases
- DIOUF Sarah**, TOULOUSE, France
Decryption of the roles of CBP methylation in human primary myoblast differentiation: cross-talk between nuclear and mitochondrial genomes
- DOS SANTOS Matthieu**, PARIS, France
Genetic control of adult muscle fiber type
- EL FISSI Najla**, MARSEILLE, France
Using drosophila as a model system to investigate how altered mitochondrial fusion triggers mitochondrial damages and neuromuscular disorders
- GAZALAH Hussein**, MONTPELLIER, France
Enigmatic perineuronal cells in the mouse and human spinal cord: properties, isolation and function
- JIMENEZ Gina**, LYON, France
Mouse in vivo gene therapy for Primary Ciliary Dyskinesia
- KHABOU Hanen**, PARIS, France
Retinitis Pigmentosa: gene therapies to prevent and restore vision loss
- KUTCHUKIAN Candice**, VILLEURBANNE, France
Phosphoinositides and Ca²⁺ signaling in normal and diseased skeletal muscle
- LAINÉ Viviane**, VILLEURBANNE, France
Characterization of the potassium channel SLO-2 in the regulation of acetylcholine receptors at the neuromuscular junction
- LAVERGNE Guillaume**, CLERMONT-FERRAND, France
Studying homing behaviour of Drosophila Adult Muscle Precursor (AMP) cells using genome wide cell specific approaches
- MIAS-LUCQUIN Dominique**, RENNES, France
Dynamics and mechanics of the myopathy-related protein dystrophin in macromolecular complexes with filamentous partners
- MORATAL Claudine**, NICE, France
Regulation of intramuscular adipogenic lineage in healthy and dystrophic human muscles
- NEY Michel**, ILLKIRCH, France
Implication of BIN1 in myotonic dystrophy type 1

- PAPAEFTHYMIUO Aikaterini**, PARIS, France
The role of the transcription factor Srf in muscle stem cells
- PLANTIÉ Emilie**, CLERMONT-FERRAND, France
New Drosophila model of Myotonic Dystrophy type 1: Phenotypic and transcriptomic characterization of functional and structural cardiac defects in adult DM1 flies
- RAESS Matthieu**, STRASBOURG, France
Deciphering the functional and molecular differences between MTM1 and MTMR2 to understand two neuromuscular diseases
- RENAUD Edith**, TOULOUSE, France
Translational regulation of gene expression during heart ischemia: applications to cardiac gene therapy
- ROUANET Sophie**, NICE, France
Genetic correction of Xeroderma Pigmentosum skin cell
- SAMSON Camille**, PARIS, France
Structural analysis of the emerin-lamin complex mutated in Emery-Dreifuss muscular dystrophy
- SANGARI Sina**, PARIS, France
Electrophysiological biomarkers of spinal neural activity in amyotrophic lateral sclerosis
- SCIONTI Isabella**, LYON, France
Epigenetics in muscle lipid metabolism
- SEBASTIEN Muriel**, LA TRONCHE, France
Mechanisms of Triad targeting of Calcium Release Complex proteins
- STREPPA Laura**, LYON, France
Biomechanical study of neuromuscular junction and its impact on myopathies
- SUTCU Haser**, PARIS, France
Implication of DNA Damage and Repair in Viability and Differentiation of Muscle Stem Cells
- TERRONE Sophie**, LYON, France
Interplay between epigenetic marks and alternative splicing during myogenesis
- TEYSSOU Elisa**, PARIS, France
In vitro and in vivo functional analysis of 2 genes identified in familial amyotrophic lateral sclerosis
- WATTIN Marion**, LYON, France
Comparative study of proteostasis during muscle degeneration in models for muscular dystrophies

Partenariats institutionnels

- NEDELEC Stéphane**, PARIS, France
Soutien du projet de recherche du candidat ATIP-Avenir 2015 : Stem cell approaches of human motor neuron diversity in development and diseases
- POREAU Brice**, GRENOBLE, France
Soutien du poste d'accueil ATIP-Avenir 2014 (année 2) : Huntingtine : rôle physiopathologique dans le muscle

Partenariats associatifs

- ARSEP/PLASSART-SCHIESS Emmanuelle**, IVRY-SUR-SEINE, France
Special Call for Research Proposals Fondation ARSEP - AFMTELETHON 2014: "Immunointervention in demyelinating diseases of the Central Nervous System"
- IFCAH/FINIDORI Joelle**, PARIS, France
Pathophysiology and therapeutic challenges of Congenital Adrenal Hyperplasia
- IRME/TADIE Marc**, PARIS, France
Traumatismes de l'encéphale et de la moelle épinière : Mécanismes et approches thérapeutiques
- POHLSCHMIDT Marita**, LONDON, Royaume Uni
Collagene VI Alliance
- RETINA France/MOSER Eric**, COLOMIERS, France
Appel d'offres Retina France 2015
- VLM/DE CARLI Paola**, PARIS, France
Appel à projets Recherche

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

BECANE Henri-Marc, PARIS, France

Study of the effect of preventive treatment with Nebivolol on the development and progression of cardiac dysfunction in children with Duchenne

DJOUADI Fatima, PARIS, France

Testing new compounds for pharmacological therapy of mitochondrial energy metabolism deficiencies

MELKI Judith, LE KREMLIN-BICETRE, France

Genetic and molecular bases of anomalies of development or function of motor neurons

UDD Bjarne, HELSINKI, Finlande

LGMD2D – natural history in R77C mutated patients

ZEITZ Christina, PARIS, France

Développement d'une thérapie génique pour restaurer la fonction de LRIT3 dans la cécité nocturne congénitale stationnaire

ACTIONS STRATEGIQUES

Projets stratégiques

GELPI Odile, PARIS, France

AAV-MPSIIIB Program: Gene therapy for neurodegeneration in Sanfilippo type B syndrome

HOVNANIAN Alain, PARIS, France

Ex vivo and in vivo gene therapy approaches for recessive and dominant dystrophic epidermolysis bullosa

LAPORTE Jocelyn, ILLKIRCH, France

AFM-IGBMC Partnership

LAPORTE Jocelyn, ILLKIRCH, France

Genetic bases, pathomechanisms and preclinical developments in congenital myopathies

MUNTONI Francesco, LONDRES, Royaume Uni

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

POURQUIE Olivier, ILLKIRCH, France

Anagenesis Biotechnologies (financement du projet de recherche) et Differentiating ES Cells or Induced Pluripotent Cells into Skeletal Muscle as Therapy for Muscular Dystrophies (iPS-2)

PUYMIRAT Jack, QUEBEC, Canada

Evaluation of peptide antisense oligonucleotides as gene therapy for myotonic dystrophy

ROTIG Agnès, PARIS, France

An integrated approach for MITOchondrial disorder THERApeutics from yeasts and worms to humans

SIMONELIG Martine, MONTPELLIER, France

Towards a clinical trial for OPMD

VASSETZKY Yegor, VILLEJUIF, France

Facio-scapulo-humeral dystrophy: from molecular mechanisms to correction of the genetic defect in immortalized myoblasts from FSHD patients

Plateformes stratégiques

BLOT Stéphane, MAISONS-ALFORT, France

Plateforme de recherche et d'expérimentation animale de l'ENVA

PERREAU-SAUSSINE Marianne, PARIS, France

I-MOTION : Création d'un centre de recherche clinique neuromusculaire pédiatrique Parisien

Pôles stratégiques

LEVY Nicolas, MARSEILLE, France

Translational Research in Marseille: towards Therapeutic Development for Rare Diseases

RELAIX Frédéric, CRETEIL, France

TRANSLAMUSCLE: An integrated translational program from basic research to biotherapies in stem cells and molecular medicine of the neuromuscular system

Structures stratégiques

FONDATION MALADIES RARES/LEVY Nicolas, PARIS, France

Subvention 2015

LEVY Nicolas, MARSEILLE, France

GIPTIS - Genetics Institute for Patients, Therapies, Innovation & Science

MOULLIER Philippe, NANTES, France

Atlantic Bio GMP

Outils stratégiques

BASSEZ Guillaume, CRETEIL, France

Base de données cliniques sur les dystrophies myotoniques (RIDM)

SACCONI Sabrina, NICE, France

French National FSHD patient registry for clinical trial planning and translational research

AUTRES ACTIONS

Manifestations scientifiques

BROWN Susan, CAMDEN TOWN, Royaume Uni

20th International Congress of the World Muscle Society

COLLOQUE JEUNES CHERCHEURS, EVRY, France

Colloque Jeunes Chercheurs 2015 12 juin 2015 - JDF Parc Floral

FSH SOCIETY/PEREZ Daniel Paul, BEDFORD – MA, Etats Unis

FSH Society Facioscapulohumeral Muscular Dystrophy [FSHD] 2015 International Research Consortium & Research Planning Meetings

GOURDON Geneviève, PARIS, France

10^{ème} congrès du consortium international sur les dystrophes myotoniques: IDMC10

JAMAR Gaëlle, PARIS, France

Annual conference of the European Society for Gene and Cell Therapy in collaboration with the Finnish society of Gene Therapy

JAMAR Gaëlle, PARIS, France

Ingestem Congress on Pluripotent Stem Cells, Reprogramming and Tissue Engineering

JAMAR Gaëlle, PARIS, France

Journées Thématiques de la SFTCG. Journée Ethique et Réglementaire et Journée Gene Regulation

KARDON Gabrielle, SALT LAKE CITY – UTAH, Etats Unis

Gordon Research Conference on Myogenesis: Molecular and Cellular Networks (June 21-26, 2015)

Gordon Research Seminar on Myogenesis (June 20, 2015)

MEZZINA Mauro, PARAY-VIEILLE POSTE, France

EMBO workshop: Modern DNA concepts/tools for safe gene transfer and modification

MOUNIER Rémi, VILLEURBANNE, France

13^{èmes} Journées de la Société Française de Myologie/Colloque Myogénèse

MUSARO Antonio, ROME, Italie

IIM-Myology Meeting 2015

PARKS Robin, OTTAWA, Canada

The 3rd Ottawa International Conference on Neuromuscular Biology, Disease & Therapy - September 24-26, 2015

PRIP-BUUS Carina, PARIS, France

8th Meeting of the "MeetOchondrie" network

TREVES Susan, BÂLE, Suisse

Gordon Research Conference on Muscle: Excitation/Contraction Coupling - Advancing Research and Leadership in EC Coupling

Plateforme CEDS

CARRE Monique, MEZILLES, France

Financement 2015 CEDS