LES PROJETS DE RECHERCHE LAUREATS 2017

APPEL À PROJETS «GENOMICS : SÉQUENÇAGE À HAUT DÉBIT & MALADIES RARES»

RESPONSABLE DU PROJET	VILLE	TITRE DU PROJET
Francis COUTURAUD	Brest	Extending identification of new inherited Thrombophilia in selected families
Stéphane SAVARY	Dijon	Novel CRISPR-mediated mutant microglial cell models to better understand the physiopathogenesis of peroxisomal leukodystrophies and identify novel therapeutic targets by NGS RNAseq
Virginie CARMIGNAC	Dijon	Unraveling the genetic basis of mutation-negative mosaic overgrowth syndromes through deep whole exome sequencing
Brahim BELBELLAA	Illkirch	Multisystem approach analysis of Friedreich Ataxia cardiomyopathy in frataxin deficient mice models and hIPS-derived cardiomyocytes for the elucidation of pathophysiological mechanisms and biomarkers identification
Veronique PAQUIS-FLUCKLINGER	Nice	Identification of new genes and possible de novo mutations in early-onset mitochondrial disorders
Claire FRANCASTEL	Paris	GenOmics of the ICF syndrome: when studying a rare disease also sheds new light on the “old” field of DNA methylation
Caroline NAVA	Paris	Identification of novel genes responsible for Dravet syndrome
Cécile JEANPIERRE	Paris	Identification of novel genes and mutational mechanisms for renal hypodysplasia
Geneviève DE SAINT BASILE	Paris	Genetic basis of various phenotypes segregating in a large inbred family
Véronique PINGAULT	Paris	Molecular bases of Waardenburg syndrome type 2
Capucine TROLLET	Paris	Long and small non coding RNA in Oculopharyngeal Muscular Dystrophy
Sandrine MARLIN	Paris	Identification of the second gene responsible for isolated hearing impairment with enlarged vestibular aqueducts
Martine COHEN-SALMON	Paris	Deciphering the gliovascular functions of MLC1 underlying megalencephalic leukoencephalopathy
François VIALARD	St Quentin en Yvelines	Identification and characterization of gene alterations in patients with a spermatogenesis maturation arrest
Stéphane VIVILLE	Strasbourg	Exome sequencing in a consanguineous family with spontaneous ovarian hyperstimulation cases with unknown triggers

APPEL À PROJETS «RECHERCHE PRÉCLINIQUE : MODÈLES EXPÉRIMENTAUX»

RESPONSABLE DU PROJET	VILLE	TITRE DU PROJET
Pierre CATTAN	Paris	Full thickness circumferential esophageal replacement by a tissue engineered construct
Aziz EL-AMRAOUI	Paris	Modeling the Usher syndrome type I (USH1) retinopathy: physiopathology and gene therapy
Olivier GOUREAU	Paris	Preclinical validation of a cell-derived Retinal Pigmented Epithelium (RPE)
Gérard LAMBEAU	Nice	Membranous nephropathy, a rare autoimmune kidney disease: establishing the first model

APPEL À PROJETS «MODÈLES MURINS DE MALADIES RARES»

RESPONSABLE DU PROJET	VILLE	TITRE DU PROJET
Jean-Vianney BARNIER	Paris	Mutation in the PAK3 gene associated to Intellectual Deficiencies
Jean-Jacques BOFFA	Paris	Pathophysiological role of Isthmin-1 in idiopathic nephrotic syndrome
Anne DEBANT	Montpellier	Contribution of *de novo* mutations in the trio gene in intellectual disability: development of a trio knock-in mouse model mimicking the human disease
Juliette GODIN	Illkirch	Understanding the roles of tRNAs modifications in cerebral corticogenesis through the investigation of an ADAT3 knock-in model
Denis HERVE	Paris	A mouse model for studying pathophysiological mechanisms of ADCY5-related dyskinesia
Pascal HOUILLIER	Paris	A Claudin 16-knock-in mouse as a model of Familial hypomagnesemia with hypercalciuria and nephrocalcinosis
Metodi METODIEV	Paris	A mouse model to understand the pathophysiolgy and tissue-specificity of mitochondrial disease caused by mutations in the RNA stability factor LRPPRC
Benoît MIOTTO	Paris	Study of a mouse model of Meier-Gorlin Syndrome based on a mutation in the conserved BAH domain of ORC1
Stéphane NEDELEC	Paris	Development and characterization of preclinical human and mouse models of Spinal Muscular Atrophy to determine the mechanisms of selective motor neuron impairments
Miria RICCHETTI	Paris	A mouse model for Cockayne syndrome
Laurent SCHAEFFER	Lyon	Light up the neuromuscular junction to monitor muscle innervation
Jacques YOUNG	Paris	Knock-in mouse model as a proof of concept for human hyperandrogenism, anovulation associated with activating LHCGR mutation

APPEL À PROJETS «RECHERCHE TRANSLATIONNELLE & CLINIQUE DANS LES MALADIES RARES» CONJOINT GCS-HUGO

RESPONSABLE DU PROJET	VILLE	TITRE DU PROJET
Dominique BONNEAU	Angers	Multi-Omics and IPSCs to Improve Diagnosis of Rare Intellectual Disabilities
Jean-Baptiste GOURRAUD	Nantes	CIQTP prolongation : role and mechanism in sudden cardiac death
Laurent PLANTIER	Tours	Acoustic Waves and Helium/oxygen for Aerosol Treatment of Idiopathic Pulmonary Fibrosis (IPF)

APPEL À PROJETS « SCIENCES HUMAINES & SOCIALES »

RESPONSABLE DU PROJET	VILLE	TITRE DU PROJET
Pierre ANCET	Dijon	Etude Naevus géant congénital : Psychologie, Info-Com, Santé, Transdisciplinarité, Ethique
Lionel DANY	Marseille	Conséquences psycho-sociales du lupus érythémateux systémique : une étude auprès des malades et de leurs conjoints
Agnès DUMAS	Villejuif	Plateformes de transition : Comprendre les attentes des parents de jeunes porteurs de maladies rares
Stéphanie MAZZA	Lyon	Etude du parcours scolaire et professionnel des patients narcoleptiques
Maria POPA-ROCH	Strasbourg	Maladie Rare Invisible et Scolarité des Enfants
Virginie POSTAL	Bordeaux	La communication dans le syndrome Prader-Willi : Etude du contrôle émotionnel lié aux troubles du comportement, de leurs répercussions au quotidien et examen de thérapies innovantes
Sébastien RUFFIE	Pointe à Pitre	Drépanocytose, troubles neurocognitifs et participation sociale