LES LAUREATS DU 3ème APPEL À PROJETS 'MOUSE MODELS AND RARE DISEASES'

JUILLET 2017

RESPONSABLE DU PROJET	LABORATOIRE	TITRE DU PROJET
Jean-Vianney BARNIER	UMR 9197 CNRS - Univ. Paris Sud, Institut des Neurosciences Paris-Saclay (Neuro-Psi), Paris	Mutation in the PAK3 gene associated to Intellectual Deficiencies
Jean-Jacques BOFFA	INSERM UMRS1155, Service de Néphrologie et dialyses, Hôpital Tenon, Paris	Pathophysiological role of Isthmin-1 in idiopathic nephrotic syndrome
Anne DEBANT	Centre de Recherche en Biologie Cellulaire (CRBM) - CNRS UMR 5237, 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	IGBMC, Médecine translationnelle, CNRS Université de Strasbourg UMR 7104 - INSERM U964, Illkirch	Understanding the roles of tRNAs modifications in cerebral corticogenesis through the investigation of an ADAT3 knock-in model
Denis HERVE	INSERM UMRS839, Institut du Fer à Moulin, Paris	A mouse model for studying pathophysiological mechanisms of ADCY5-related dyskinesia
Pascal HOUILLIER	INSERM UMRS1138, Centre de Recherche des Cordeliers, Paris	A Claudin 16-knock-in mouse as a model of Familial hypomagnesemia with hypercalciuria and nephrocalcinosis
Metodi METODIEV	INSERM U1163, Institut Imagine, Hôpital Necker-Enfants Malades, Paris	A mouse model to understand the pathophysiology and tissue-specificity of mitochondrial disease caused by mutations in the RNA stability factor LRPPRC
Benoît MIOTTO	INSERM UMRS1138, Centre de Recherche des Cordeliers, Paris	Study of a mouse model of Meier-Gorlin Syndrome based on a mutation in the conserved BAH domain of ORC1
Stéphane NEDELEC	INSERM UMRS839, Institut du Fer à Moulin, 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	Institut Pasteur, Cellules souches et développement, Paris	A mouse model for Cockayne syndrome
Laurent SCHAEFFER	Institut Neuromyogène, Interactions neurone-muscle, Lyon	Light up the neuromuscular junction to monitor muscle innervation
Jacques YOUNG	INSERM U1185, Faculté de médecine Paris Sud, Le Kremlin-Bicêtre	Knock-in mouse model as a proof of concept for human hyperandrogenism, anovulation associated with activating LHCGR mutation