LES LAURÉATS DE L'APPEL À PROJETS 'GENOMICS'

JUILLET 2017

RESPONSABLE DU PROJET	LABORATOIRE	TITRE DU PROJET
Sandrine MARLIN	Hôpital Necker-Enfants Malades, AP-HP, Paris	Identification of the second gene responsible for isolated hearing impairment with enlarged vestibular aqueducts
Brahim BELBELLAA	IGBMC, Dpt. Médecine translationnelle, CNRS Université de Strasbourg UMR 7104 - INSERM U964, Illkirch	Multisystem approach analysis of Friedreich Ataxia cardiomyopathy in frataxin deficient mice models and hIPS-derived cardiomyocytes for the elucidation of patho-physiological mechanisms and biomarkers identification
Martine COHEN-SALMON	Centre Interdisciplinaire de Recherche en Biologie, CNRS UMR 7241 INSERM U1050, Collège de France, Paris	Deciphering the gliovascular functions of MLC1 underlying megalencephalic leukoencephalopathy
Claire FRANCASTEL	CNRS UMR7216, Epigénétique et destin cellulaire, Université Paris Diderot, Paris	GenOmics of the ICF syndrome: when studying a rare disease also sheds new light on the "old" field of DNA methylation
François VIALARD	Gamète, Implantation, Gestation (GIG) EA7404, Université Versailles Saint Quentin	Identification and characterization of gene alterations in patients with a spermatogenesis maturation arrest
Caroline NAVA	Département de Génétique, Hôpital de la Pitié-Salpêtrière, Paris	Identification of novel genes responsible for Dravet syndrome
Virginie CARMIGNAC	INSERM UMR1231, Génétique des Anomalies du Développement (GAD, Université de Bourgogne, Dijon	Unraveling the genetic basis of mutation-negative mosaic overgrowth syndromes through deep whole exome sequencing
Stéphane VIVILLE	Laboratoire de diagnostic génétique, Hôpitaux Universitaires de Strasbourg	Exome sequencing in a consanguineous family with spontaneous ovarian hyperstimulation cases with unknown triggers
Cécile JEANPIERRE	INSERM U1163, Institut Imagine, Hôpital Necker-Enfants Malades, Paris	Identification of novel genes and mutational mechanisms for renal hypodysplasia
Francis COUTURAUD	Département de médecine interne et pneumologie EA3878, CHU Brest, Université de Bretagne Occidentale	Extending identification of new inherited Thrombophilia in selected families

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Veronique PAQUIS-FLUCKLINGER	IRCAN, UMR 7284 INSERM U1081, Université de Nice Sophia Antipolis, Nice	Identification of new genes and possible de novo mutations in early-onset mitochondrial disorders
Geneviève DE SAINT BASILE	INSERM U1163, Institut Imagine, Hôpital Necker-Enfants Malades, Paris	Genetic basis of various phenotypes segregating in a large inbred family
Véronique PINGAULT	INSERM U1163, Institut Imagine, Hôpital Necker-Enfants Malades, Paris	Molecular bases of Waardenburg syndrome type 2
Stéphane SAVARY	BioPeroxIL EA7270 , Université de Bourgogne, Dijon	Novel CRISPR-mediated mutant microglial cell models to better understand the physiopathogenesis of peroxisomal leukodystrophies and identify novel therapeutic targets by NGS RNAseq
Capucine TROLLET	Centre de Recherche en Myologie, UMRS974, Institut de Myologie, Paris	Long and small non coding RNA in Oculopharyngeal Muscular Dystrophy