FONDATION maladies rares

LES PROJETS SOUTENUS 2016

1ER APPEL À PROJETS «GENOMICS : SÉQUENÇAGE À HAUT DÉBIT & MALADIES RARES»

RESPONSABLE DU PROJET	VILLE	TITRE DU PROJET
Genevieve BAUJAT	Paris	Molecular basis dissection of isolated Klippel Feil syndrome: identification of new genes
Stéphane BÉZIEAU	Nantes	Trio-based whole-genome sequencing of patients with syndromic and non-syndromic severe intellectual disability
Célia CRÉTOLLE	Paris	Genotype-endophenotype correlation study in patients with a MNX1 gene non mutated Currarino syndrome
Yanick CROW	Paris	Whole genome sequencing in Aicardi-Goutières syndrome and related type I interferonopathies
Albertina DE SARIO	Montpellier	DNA methylation and pulmonary disease in cystic fibrosis patients
Christel DEPIENNE	Illkirch	Identification of the unconventional genetic basis for familial cortical myoclonic tremor and epilepsy
Pascale GUICHENEY	Paris	Elucidation of the molecular variants responsible for sudden cardiac death in two large families
Alice HADCHOUEL	Paris	Identification of a new gene in a familial form of pulmonary alveolar proteinosis
Eric LE GUERN	Paris	Identification of new genes for familial forms of generalized epilepsies
Guy LENAERS	Angers	Genetic analysis of dominant optic atrophy
Caroline MICHOT	Paris	Dissection of molecular bases of Cornelia de Lange syndrome: identification of new genes in pre-screened patients
Jean MULLER	Strasbourg	Identification of novel genes underlying Bardet-Biedl Syndrome using Next Generation Sequencing
Christian PINSET	Evry	Studying myogenesis and the onset of Duchenne muscular dystrophy (DMD) in human pluripotent stem cells to identify early disease markers and potential therapeutic targets
Amélie PITON	Illkirch	Evaluation of RNA-sequencing strategies to better diagnose intellectual disability
Angela TINGAUD-SEQUEIRA	Bordeaux	Exome sequencing to find new candidate genes involved in Goldenhar Syndrome and Oculo-Auriculo-Vertebral Spectrum
Sandrine VUILLAUMIER-BARROT	Paris	Identification of a gene underlying same coagulation factors abnormalities and CDG II profile in two unrelated family with anticipated dominant transmission.

2ÈME APPEL À PROJETS «GENOMICS : SÉQUENÇAGE À HAUT DÉBIT & MALADIES RARES»

MARIE-CHRISTINE ALESSI	Marseille	Unraveling molecular mechanisms of ETV6-releted thrombocytopenia
MATHIEU BARBIER	Paris	In search of genetic modifiers to predict the Age at Onset in Frontotemporal-Lobar Dementia
LOUISE BENARROCH	Paris	Identification of new genes involved in Marfan syndrome and Familial Thoracic Aortic Aneurysm and Dissection
ANGE-LINE BRUEL	Dijon	Identification of new genes implicated in oral-facial-digital syndromes, in exome-negative patients
VALÉRIE CORMIER-DAIRE	Paris	Identification of the molecular basis of genochondromatosis
ALEXANDRE FABRE	Marseille	Genetic bases of Primary Intestinal lymphangiectasia
DELPHINE HERON	Paris	Identification of genes for isolated agenesis of the corpus callosum without intellectual deficiency
ANNE JOUTEL	Paris	Dissecting molecular pathways involved in COL4A1-related intracerebral hemorrhage
JOCELYN LAPORTE	Illkirch	Combining high throughput sequencing approaches to define the genetic bases of myopathies
ROLAND LIBLAU	Toulouse	Human immune signatures of narcolepsy with cataplexy
HERVÉ MOINE	Illkirch	Identification of the FMRP binding site on its neuronal mRNA targets by CLIP-seq in the Fmr1-KO mouse model of the fragile X syndrome
AGNES ROTIG	Paris	Identification of nuclear genes of mitochondrial diseases with neurological involvement
CHRISTEL THAUVIN	Dijon	Identification of new genes implicated in undiagnosed developmental anomalies following a genotype-first approach using genome sequencing, in trio-exome-negative patients

APPEL À PROJETS «CRIBLAGE À HAUT DÉBIT DE MOLÉCULES À POTENTIEL THÉRAPEUTIQUE»

RESPONSABLE DU PROJET	VILLE	TITRE DU PROJET
Valérie DESQUIRET-DUMAS	Angers	Screening of pharmacological molecules to restore oxidative metabolism in rare diseases associated to mitochondrial complex I mutations
Michel FONTES	Marseille	High throughput screening to identify molecules correcting connexon activity in X-linked Charcot-Marie-Tooth disorder
Sylvie FOURNEL-GIGLEUX	Nancy	Search for inhibitors of the galactosyltransferase β4GalT7 by High Throughput Screening : towards a specific substrate reduction therapy in mucopolysaccharidoses
Mathieu RODERO	Paris	Identification of molecules able to control interferon beta transcription in patients with gain-of-function mutations in TMEM173

APPEL À PROJETS «CRÉATION DE MODÈLES ANIMAUX»

Yann AUDIC	Rennes	Xenopus epidermis development in the search for modifier genes of epidermolysis bullosa
Charles-Henry COTTART	Paris	A new rat model for cystic fibrosis carrying the F508DEL mutation in the CFTR gene
Benjamin DEHAY	Bordeaux	Physiopathological characterization of a rat model of Kufor-Rakeb syndrome
Bruno DELLA GASPERA	Paris	Xenopus tropicalis model of spinal muscular atrophy with respiratory distress
Laurence LEGEAI-MALLET	Paris	Zebrafisch model of FGFR3-related skeletal disorders
Brigitte LELONGT	Paris	Impact of ANKS3 human mutation in rat model
Sylvie MAZOYER	Lyon	Study of the physiopathology of RNU4ATAC-associated diseases
Gilles MILLAT	Lyon	PRKAG2 mutations as a molecular explanation on patients with dilated cardiomyopathy and endocardial fibroelastosis?
Christophe SIRAC	Limoges	Establishment of a rat model fro AL amyloidosis

APPEL À PROJETS « SCIENCES HUMAINES & SOCIALES »

RESPONSABLE DU PROJET	VILLE	CHAMP DISCIPLINAIRE	TITRE DU PROJET
Hélène AMIEVA	Bordeaux	Psychologie	Prise en charge pluridisciplinaire et personnalisée des troubles du comportement dans la Dégénérescence Lobaire Fronto-Temporale
Delphine DELLACHERIE	Lille	Psychologie	TEMps et DANse comme outil de remédiation du fonctionnement dans les anomalies de développement du CErvelet
Caroline DESOMBRE	Lille	Psychologie	Élèves touchés par l'hémoPHILie et autres maladies hémorragiques familiales : cOmMENt rétablir l'Egalité des chances à l'école