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	Strasbourg	• 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 in human pluripotent stem cells to identify early disease markers and potential therapeutic targets
Amélie PITON	Strasbourg	• 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»

RESPONSABLE DU PROJET	VILLE	TITRE DU PROJET
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	Strasbourg	• Combining high throughput sequencing approaches to define the genetic bases of myopathies
Roland LIBLAU	Toulouse	• Human immune signatures of narcolepsy with cataplexy
Hervé MOINE	Strasbourg	• 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 4GalT7 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»

RESPONSABLE DU PROJET	VILLE	TITRE DU PROJET
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'égalité des chances à l'école