FONDATION maladies rares

LES PROJETS SOUTENUS 2015

1ER APPEL À PROJETS « SÉQUENÇAGE À HAUT DÉBIT »

RESPONSABLE DU PROJET	VILLE	TITRE DU PROJET
Pauline ARNAUD	Paris	Identification of new genes involved in Marfan Syndrome and Familial Thoracic Aortic Aneurysm and Dissection (FTAAD).
Isabelle AUDO	Paris	Gene defect identification in X-linked retinitis pigmentosa cases excluded for currently known gene defects.
Nadia BAHI-BUISSON	Paris	Investigating novel modular basis for Periventricular Nodular Heterotopia.
Stéphanie BAULAC	Paris	Identification of new genes for autosomal dominant focal epilepsies.
Patrick CALLIER	Dijon	Identification of the gene for Pai syndrome through Whole Genome Sequencing.
Vincent CANTAGREL	Paris	Genetic basis of childhood-onset cerebellar atrophy associated with intellectual disability.
Nadia CERF-BENSUSSAN	Paris	Determination of mendelian causes of intestinal inflammation.
Nicolas CHASSAING	Toulouse	Analysis of regulatory elements sequences in microphthalmia/anophthalmia.
Jamel CHELLY	Strasbourg	Genetics of Focal Cortical Dysplasias.
Alexandre FABRE	Marseille	Identification of new genes associated with syndromic diarrhea tricho-hepato-enteric syndrom.
Sylvain LATOUR	Paris	Molecular identification of novels forms of inherited lymphoproliferation syndromes associated with a susceptibility to EBV infection.
Valérie MALAN	Paris	Unmasking of a recessive mutation: what role in the incomplete penetrance of CNVS.
Sandrine MARLIN	Paris	Identification of the molecular basis of cochlear nerve aplasia.
Sophie NAUDION	Bordeaux	Identification of the molecular bases of a new phenotype of multiple malformations in a multiplex family.
Véronique PAQUIS-FLUCKLINGER	Nice	Identification of new genes responsible for mitochondrial disorders by exome sequencing.
Nathalie ROUX-BUISSON	Grenoble	Identification of new genes in Exertional Heat Stroke syndrome (EHS) with positive in vitro contracture test and no mutation in RYR1 gene.
Stéphane VIVILLE	Strasbourg	Genetics of male infertility : genes implicated in non-obstructive azoospermia.
Christina ZEITZ	Paris	Identification of novel gene defects underlying retinitis pigmentosa in France by whole exome sequencing.

2ND APPEL À PROJETS « SÉQUENÇAGE À HAUT DÉBIT »

Marie Christine ALESSI	Marseille	Identification of new genes involved in platelet dysfunction.
Christine BELLANE CHANTELOT	Paris	Identification of new genes in congenital neutropenia.
Alain CALENDER	Lyon	Identification of genes involved in familial forms of sarcoidosis.
Pascale DE LONLAY	Paris	Identification of the gene(s) responsible for recessive rhabdomyolysis and Reye syndrome in 3 patients from 2 families presenting the same phenotype.
Muriel GIRARD	Paris	Identification of a common signaling pathway involved in biliary atresia.
Céline HUBER	Paris	Identification of the molecular basis of the Asphyxiating Thoracic Dysplasia.
Vincent LAUGEL	Strasbourg	Investigating novel molecular basis for Cockayne syndrome.
Caroline MICHOT	Paris	Dissection of molecular bases of microcephalic osteodysplastic primordial DWARFISMS : Identification of new genes in pre-screened patients.
Véronique PAQUIS-FLUCKLINGER	Nice	Progressive external ophtalmoplegia with multiple mitochondrial DNA deletions: Identification of new genes.
Laurent PASQUIER	Rennes	Identification of a gene involved in rhombencephalosynapsis.
Magana PRASAD	Strasbourg	Identification of novel genes underlying Bardet-Biedl and Bardet-Biedl-like syndromes.
Pierre RAY	Grenoble	Identification of genetic causes of human non-obstructive azoospermia (NOAZOO).
Caroline ROORYCK-THAMBO	Bordeaux	Identification of a gene involved in familial right ventricular hypoplasia.
Agnès ROTIG	Paris	Identification of a nuclear gene responsible of abnormal respiratory chain assembly.

APPEL À PROJETS « CRÉATION DE MODÈLES ANIMAUX »

RESPONSABLE DU PROJET	VILLE	TITRE DU PROJET
Johann BOHM	Illkirch	First mammalian model for tubular aggregate myopathy and Stormorken syndrome.
Pascale BOMONT	Montpellier	Development of an In vivo model for Giant Axonal Neuropathy.
Jamel CHELLY	Illkirch	Understanding NEDD4L-related MCD (Malformations of Cortical Development) through investigations of a Knock-In mouse model.
Hélène DOLLFUS	Strasbourg	Mouse modeling of a missense mutation in the essential gene PIK3R4 (VPS15) responsible fora ciliopathy-like disease.
Julie DUMONCEAUX	Paris	FacioScapuloHumeral Dystrophy (FSHD): targeting two alternative Fat1 exons with one mouse.
Christian HAMEL	Montpellier	Impg1 gene Knock-Out, a mouse model for human vitelliform macular dystrophy and retinitis pigmentosa.
Alain HOVNANIAN	Paris	Generation of a humanized mouse model for Recessive Dystrophic Epidermolysis Bullosa harbouring a recurrent COL7A1 mutation.
Sébastien LACROIX DES MAZES	Paris	Generation of a novel mouse model of hemophilia A constituted of mice transgenic for a human T cell receptor specific for therapeutic factor VIII.
Carine LE GOFF	Paris	SMAD4 and Myhre syndrome.
Delphine MEYNARD	Toulouse	Is Matriptase-2 involved in iron homeostasis regulation and anemia exclusively through the hepatocytes ?
Véronique PAQUIS-FLUCKLINGER	Nice	CHCHD10S59L mouse model: how mitochondrial dysfunction promotes motor neuron disease ?
Frédérique RENE	Strasbourg	Generation of an ALS-FTD mouse model based on a conditional CHMP2B intron 5 mutant knock-in.
Frédéric RIEUX-LAUCAT	Paris	Role of LRBA in the control of the immune response: implication in primary immunodeficiencies.

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

Pascale BOMONT	Montpellier	Reversing motor deficits in Glant Axonal Neuropathy.
Vincent GACHE	Lyon	Rescue myonuclear domains establishment in centronuclear myopathies with chemical compounds.
Yvon TROTTIER	Strasbourg	High Throughput screening for the identification of amyloid aggregation modulators in Huntington's disease.

APPEL À PROJETS « SCIENCES HUMAINES & SOCIALES »

RESPONSABLE DU PROJET	VILLE	CHAMP DISCIPLINAIRE	TITRE DU PROJET
Sophie ARBORIO	Villers les Nancy	Anthropologie	Syndrome de West : construction des savoirs et singularité des expériences des familles.
Michel CASTRA	Lille	Sociologie	Situations de handicap et discrimination en période pré-greffe. Etude comparativeentre deux maladies pulmonaires rares : la mucoviscidose et la fibrose pulmonaire idiopathique.
Dominique FARGE	Paris	Sociologie	Mise en place d'une e-plateforme collaborative pour identifier et caractériser les handicaps des patients atteints de maladies auto-immunes rares relevant de la thérapie cellulaire ou debiothérapies.
Anne MARCELLINI	Montpellier	Sociologie	Devenir adulte avec une anomalie du développement : obstacles et facilitateurs.
Grégoire MERCIER	Montpellier	Economie de la santé	Reste à charge des patients atteints de lymphœdème primaire : mesure des inégalités financières d'accès aux soins.
Remy POTIER	Paris	Psychologie	Déterminants psychosociaux de l'impact du handicap de surdicécité sur l'autonomie au sein du parcours de vie chez les personnes atteintes des syndromes de Usher, Wolfram et Stickler.
Sophie QUINTON	Lille	Droit	Sclérodermie et difficultés professionnelles : identifier pour mieux aider.