Comprendre les maladies rares

• 1er appel à projets « Séquençage à haut débit »

Responsable du projet	Ville	Titre du projet
Nadia Bahi-Buisson	Paris	Identification of the genetic bases of Aicardi syndrome
Patrick Callier	Dijon	Genetic basis of frontonasal dysplasia
Valérie Cormier-Daire	Paris	Identification of the molecular basis of the Ellis-van Creveld (EVC) syndrome
Valérie Delague	Marseille	Identification of novel genes in Charcot-Marie-Tooth disease in lebanese consanguineous families
Marion Gérard	Caen	Whole-exome sequencing to identify genetic alterations associated with sirenomelia (ADEP project)
Pascale Guicheney	Paris	Identification of a new gene causing idiopathic ventricular fibrillation with short-coupled variant of torsade de pointes
Jean-Louis Mandel	Illkirch	Exome sequencing of patients with intellectual disability and no mutation identified in known genes
Sophie Nicole	Paris	Search for a new gene responsible for periodic paralysis by whole exome analysis of one family and sporadic cases
Véronique Paquis-Flucklinger	Nice	Early-onset neuromuscular presentations of mitochondrial disorders: Identification of new genes by exome sequencing
Karine Poirier	Paris	Identification of new genes involved in microlissencephaly
Patrick Revy	Paris	Identification of new genes involved in severe bone marrow failure associated with telomere and/or DNA repair defects
Damien Sanlaville	Lyon	Study of 9 complex chromosomal rearrangements by massively parallel sequencing: An unifying mechanism?
Florent Soubrier	Paris	Exome sequencing in two large pedigrees with multiple cases of IgA nephropathy
Giovanni Stevanin	Paris	Whole genome sequencing in 5 families with hereditary spastic paraplegia
Sandrine Vuillaumier-Barrot	Paris	Identification of a gene underlying dominant epilepsy in a SLC2A1/GLUT1 negative family (trio analysis)

• 2nd appel à projets « Séquençage à haut débit »

Responsable du projet	Ville	Titre du projet
Nadia Bahi-Buisson	Paris	Delineation of the molecular basis of cortical malformation in 22q11.2 deletion syndrome
Eric Bieth	Toulouse	Search for new genetic determinants of male infertility due to congenital bilateral absence of the vas deferens
Francis Couturaud	Brest	Identification of new inherited thrombophilia in selected families
Christel Depienne	Paris	Identification of novel genes responsible for autism-epilepsy phenotypes
Jamal Ghoumid	Lille	Molecular characterization of blepharocheilodontic (BCD) syndrome through exome sequencing in 5 families
Gaëlle Hardy	Grenoble	New genes involved in bradykinin-mediated angioedema with reduced C1-inhibitor function and no mutation in SERPING1 or F12 genes
Carine Le Goff	Paris	Identification of a new gene involved in Floating Harbor syndrome
Aude Magérus-Chatinet	Paris	Search for modifier genes in Autoimmune lymphoproliferative syndrome (ALPS) - Fas
Gaël Manes	Montpellier	Identification of novel genes in autosomal dominant retinitis pigmentosa in 21 fully screened families for known genes
Rolando Meloni	Paris	Research of a major gene for familial form of bipolar disorder in an extended pedigree with an ascertained founder effect
Sébastien Moutton	Bordeaux	Identification of molecular bases of omphalocele-exstrophy of bladder-imperforate anus-spinal defects (OEIS complex) in a multiplex family
Irène Netchine	Paris	Identifying new genes responsible for autosomal inheritance of Russell-Silver syndrome
Michel Polak	Paris	Identification of new genes involved in brain-lung-thyroid syndrome
Reiner Veitia	Paris	Genetics and genomics of primary ovarian insufficiency: an entry point to understand ovarian function

• Appel à projets « Création de modèles expérimentaux »

Responsable du projet	Ville	Titre du projet
Vincent Beringue	Jouy-en-Josas	Zebrafish model of prion disease
Nicolas Charlet-Berguerand	Illkirch	A rat model of amyotrophic lateral sclerosis & frontotemporal dementia
Kathrin Gieseler	Villeurbanne	Development of C. elegans models for human muscular dystrophies
Jamilé Hazan	Paris	Dangerous liaisons: The link between spastin, atlastin and BMP signaling in the pathogenesis of hereditary spastic paraplegia
Yann Hérault	Illkirch	A rat model for the 16p11.2 microdeletion syndrome to better understand and treat the cognitive and metabolic disorders induced in human
Jocelyn Laporte	Illkirch	Validation and pathophysiological characterization of novel genes for myopathies (MYO-fish)
Olivier Loréal	Rennes	Hereditary aceruloplasminemia: mechanisms involved in the expression of the disease and development of new therapeutic approaches
Grégoire Michaux	Rennes	Towards a model for rare intestinal absorption diseases in C. elegans
Arnaud Monteil	Montpellier	Modeling infantile neuroaxonal dystrophy, a NALCN channel-related disorder, in zebrafish
Véronique Morel	Lyon	Direct access to Nesprin1 variants contribution to Emery Dreifuss Muscular Dystrophy and Autosomal Recessive Cerebellar Ataxia ARCA1
Sophie Nicole	Paris	Neuromuscular excitability disorders in zebrafish: progressive muscle weakness in periodic paralysis and congenital myasthenia
Frédéric Perros	Le Plessis Robinson	Role of KCNK3 in the pathogenesis of pulmonary arterial hypertension
Thomas Pietri	Paris	Characterization of a zebrafish model of Rett syndrome
Isabelle Richard	Evry	Rat model for calpainopathies
Dimitrios Skoufias	Grenoble	Generation of a zebrafish model to study KIF11 motor protein mutations associated with the human Microcephaly-Lymphedema-Chorioretinal-Dysplasia (MLDRC) rare syndrome
Hervé Tricoire	Paris	Generation of new endogenous drosophila models of trinucleotide repeat expansion diseases
Yvon Trottier	Illkirch	A zebrafish model of SCA7 for physiopathological analyses and drug evaluation

Développer de nouveaux traitements

• Appel à projets « Criblage à haut débit de molécules à potentiel thérapeutique »

Responsable du projet	Ville	Titre du projet
Irène Ceballos-Picot	Paris	Identify molecules able to induce HPRT-like activity in HPRT-deficient fibroblasts and dopaminergic neuronal cells as models for Lesch-Nyhan Disease
Hanna Debiec	Paris	Searching for C5b-9 antagonists by high-throughput screening of chemical libraries: Toward new treatment of membranous nephropathy
Thomas Falguières	Paris	Identification of targeting correctors of ABCB4/MDR3 defective mutants by a high throughput screening approach
Olfa Khalfallah	Nice	Search for active molecules on a cell model for Fragile X Syndrome by high throughput screening
Fabrice Lejeune	Lille	Correction of nonsense mutations in genetic diseases
Delphine Meynard	Toulouse	Matriptase-2: identification of pharmacological inhibitors to decrease iron overload in non-transfusion-dependent thalassemia

• Appel à projets « Développement des étapes translationnelles »

Responsable du projet	Ville	Titre du projet
Marie-Anne Colle	Nantes	Gene therapy for pediatric forms of Pompe disease using AAV gene transfer to the CNS: Preclinical feasibility
Philippe Moullier	Nantes	Systemic injection of a recombinant AAV vector encoding for the µdystrophin: Efficiency and global safety
Sibylle Opsahl Vital	Montrouge	Development of a bioengineering treatment for the necrotic pulp of patients with familial rickets: Preclinical approach
Pierre-Louis Tharaux	Paris	Treatment of rapidly progressive glomerulonephritis by powerful inhibitors of HB-EGF and miRNA92a pathway

Améliorer le parcours de vie des personnes malades

• Appel à projets « Sciences humaines et sociales & maladies rares »

Responsable du projet	Ville	Titre du projet
Pascal Antoine	Villeneuve d'Ascq	Impact de trois MAladies GEnétiques Rares : Recherche psychosociale exploratoire et comparative
Drina Candilis Huisman	Paris	Parcours de soins et accession à la parentalité des femmes en situation de handicap moteur atteintes de maladies rares
Philippe Charron	Paris	Test génétique prédictif dans les maladies cardiaques héréditaires : évaluation de l'impact psycho-social et de la prise en charge pluridisciplinaire
Hervé Devilliers	Dijon	Étude qualitative des conséquences des maladies auto-immunes sur la qualité de vie
Alexandre Eusebio	Marseille	Étude transversale des facteurs déterminant la qualité de vie du patient et le fardeau de l'aidant dans la paralysie supranucléaire progressive
Laurence Faivre-Olivier	Dijon	Préférences et représentations face aux technologies de séquençage à haut débit pour la pratique de la génétique médicale. Le cas des anomalies du développement
Damien Léger	Paris	Parcours professionnel des personnes atteintes d'hypersomnies rares

Partager à l'échelle internationale

• Appel à projets conjoint Fondation maladies rares - TWAS

Responsable du projet	Ville	Titre du projet
Sonia Abdelhak	Tunis	Consanguinity and hereditary rare diseases: Challenges and perspectives in post genomics

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