GIS Institut des maladies rares

Résultats de l'appel d'offres 2004

Nom du coordinateur		Laboratoire du coordinateur	Titre du projet
ADNOT	Serge	Inserm U. 492, Faculté de médecine de Créteil, Creteil	Is lung serotonin synthesis playing a key role in pathogenesis of human primary pulmonary hypertension ?
ARENZANA	Fernando	Département de médecine moléculaire, Institut Pasteur, Paris	The pathogenic mechanisms of the WHIM syndrome: characterisation of the molecular basis and genetic diversity
BACHOUD-LEVI	Anne-Catherine	Inserm U. 421, Hôpital Henri Mondor, Créteil	Predictive biomarkers of Huntington's Disease: extension to presymptomatic patients, to new markers and to brain imaging and infraclinic cognitive tasks
BARDONI	Barbara	IFR 50, CNRS FRE 2720, Faculté de médecine, Nice	Molecular genetics of mental retardation
BELMATOUG	Nadia	Département de médecine interne, Hôpital Beaujon, Clichy	Computer solutions for the epidemiological registries of rare diseases: the Gaucher Disease as a model for a universal solution
BLOCH	Juliette	Département des maladies chroniques et traumatiques, Institut de veille sanitaire, Saint Maurice	Feasibility study of the epidemiological surveillance of rare diseases

BOILEAU	Catherine	Inserm U. 383, Hôpital Necker Enfants Malades, Paris	Research network on rare monogenic hypercholesterolemia
CAILLAUD	Catherine	Inserm U. 567, Faculté de médecine Cochin-Port Royal, Paris	Characterization of the molecular and pathophysiological mechanisms involved in ceroid-lipofuscinoses
CARTIER	Nathalie	Inserm U. 561, Hôpital Saint Vincent de Paul, Paris	Gene Therapy for Metachromatic Leukodystrophy: direct intracerebral targeting of the human ASA gene through stereotaxic injection of recombinant adeno-associated vector
CLEMENT	Annick	Inserm E213, Hôpital Armand Trousseau, Paris	Research network on modifier genes in cystic fibrosis
COLIN	Yves	Inserm Unité 76, Institut National de transfusion sanguine, Paris	Collaborative biological research network for childhood Evans Syndrome (ES) and Auto-Immune Hemolytic Anemia (AIHA)
COPPO	Paul	Service d'hématologie et de thérapie cellulaire, Hôpital Saint Antoine, Paris	Adult Thrombotic Microangiopathies (< 18 year-old): prospective study of specific von Willebrand factor cleaving-protease (ADAMTS13) and complement Factor H. Establishment of a study network
DEUGNIER	Yves	Inserm CIC 0203, CHU Pontchaillou, Rennes	A French network for the management of family screening and research studies on modifier genes in HFE hereditary haemochromatosis
DEYBACH	Jean-Charles	Inserm U. 409 et Centre Français des Porphyries, Hôpital Louis Mourier, Colombes	European Porphyria Initiative (EPI) : European network of platform for clinical and basis research on porphyrias : www.porphyria-europe.com
DOLLFUS	Hélène	Service de génétique médicale, Hôpitaux universitaires de Strasbourg, Strasbourg	Cockayne syndrome and related disorders of DNA repair and transcription: phenotype-genotype correlations - identification of new clinical and molecular entities

DONADIEU	Jean	Service d'hémato oncologie pédiatrique, Hôpital Trousseau, Paris	Phenotypic and genotypic analysis of patients included in the French register of severe chronic neutropenia, and functional study of interactions between neutrophil elastase and the CXCR4/SDF1 complex related to ELA2 mutations
DURANDY	Anne	Inserm U. 429, Hôpital Necker Enfants Malades, Paris	Molecular delineation of hyper-IgM syndromes
ESTOURNET-MATHIAUD	Brigitte	Service de pédiatrie, Hôpital Raymond Poincaré, Garches	French Congenital Muscular Dystrophies Network (CMD network): Natural history, carrying out of phenotypic and molecular characterisation and therapeutical approaches of CMDs with main interest in Ulrich's syndrome and collagen 6 abnormalities
FERRY	Nicolas	CIC-Inserm 4 Biothérapies hépatiques, CHU Hôtel Dieu, Nantes	Gene therapy for Crigler-Najjar disease. New strategies to circumvent immune response
GEISSMANN	Frédéric	Inserm équipe Avenir, Laboratoire d'anatomie pathologique, Hôpital Necker Enfants Malades, Paris	Development of a murine model for langerhans cell histocytosis using binary knock-in mice for selective targetting of activated langerhans cells
GUICHENEY	Pascale	Inserm U. 582, Institut de Myologie, Paris	Network on Brugada syndrome: phenotype-genotype assessment : Establishment of a web-based database
HUMBERT	Marc	UPRES EA 2705, Service de pneumologie et réanimation respiratoire, Hôpital Antoine Béclère, Clamart	The French Network on Pulmonary Arterial Hypertension
JONDEAU	Guillaume	Consultation Multidisciplinaire Marfan, Hôpital Ambroise Paré, Boulogne	Network on Marfan syndrome and non syndromic overlapping diseases
LAVAU	Catherine	CNRS UPR 9051, Hôpital Saint-Louis, Paris	Novel therapeutic targets in MLL leukemias: analysis of the potential role of FLT3 and other tyrosine kinases in a murine model

LEGEAI-MALLET	Laurence	Inserm U. 383, Hôpital Necker Enfants Malades, Paris	Specific tyrosine kinase inhibitors for the management of Achondroplasia
LEMARCHAND	Patricia	Inserm U. 533, Institut du Thorax, Nantes	Cell therapy for familial cardiomyopathy
LYONNET	Stanislas	Inserm U. 393, Hôpital Necker Enfants Malades, Paris	Hirschsprung disease and related neurocristopathies: Genetics, molecular bases and functional genomics
MAZURIER	Claudine	Laboratoire français du fractionnement et des biotechnologies, Lille	Preclinical development of a human plasma-derived factor H concentrate for the treatment of patients affected with atypical haemolytic uraemic syndrome
PECKER	Françoise	Inserm U. 581, Hôpital Henri Mondor, Créteil	The role of tumor necrosis factor (TNF) in Emery-Dreifuss muscular dystrophy and other laminopathies affecting striated muscles.: N-acetylcysteine as a potential anti-TNF therapy.
PUCCIO	Hélène	IGBMC, Strasbourg	Finding therapeutics for Friedreich Ataxia: A large-scale pharmacological screen on a novel cellular model
REYNIER	Pascal	EA3862, Laboratoire de biochimie et biologie moléculaire, CHU d'Angers, Angers	Mitochondrial diseases network - Evaluation of high-throughput sequencing microarray technology in mtDNA diseases
RODIEN	Patrice	Inserm E 0018, CHU d'Angers, Angers	Resistance to thyroid hormones: Clinical network and design of new therapeutics
ROLLING	Fabienne	Inserm U. 649, CHU Hôtel-Dieu, Nantes	Evaluation of a recombinant Adeno-Associated Vector serotype 4 (rAAV-4) vector for the gene replacement therapy of a canine model of leber congenital amaurosis (LCA): the Briard dog rpe65-/-

RONCO	Pierre	Inserm U. 489, Hôpital Tenon, Paris	Pathogenesis of renal and nonrenal diseases induced by fetomaternal allo-immunization : The model of neutral endopeptidase deficiency
RUGGIERO	Florence	UMR5086 CNRS, Institut de biologie et chimie des protéines, Lyon	Molecular mechanisms involved in classical Ehlers-Danlos syndrome caused by mutations in collagen V genes
SALOMON	Rémi	Inserm U. 574, Hôpital Necker Enfants Malades, Paris	Nephronophthisis and medullary cystic kidney disease network
STEVANIN	Giovanni	Inserm U. 289, Hôpital de la Pitié-Salpêtrière, Paris	Clinico-genetic analysis of complex and severe forms of autosomal recessive neuropathies
VILLARD	Laurent	Inserm U. 491, Faculté de médecine de la Timone, Marseille	Network for the study of abnormal cortical development
ZENNARO	Maria-Christina	Inserm U. 36, Collège de France, Paris	Type I pseudohypoaldosteronism network (PHA1NET): Genetic mechanisms and cardiovascular complications