Projets lauréats de l'appel à projets 2013 'Mouse models and rare diseases' - Fondation maladies rares / PHENOMIN		
Porteur du projet	**Laboratoire**	**Titre du projet**
Jeanne AMIEL	Inserm U781, Hôpital Necker-Enfants Malades, Paris	Proposal to create a mouse model of EVE dysplasia by generating Hspa9 knockout mice
Sabine BAILLY	iRTSV, BCI - Inserm U1036, CEA-Grenoble	BMP10 in HHT disease
Marie-Christine CHABOISSIER	Inserm U1091, Institut de Biologie Valrose (iBV), Nice	Analysis of R-spondin1 functions in transdifferentiation and maintenance of the ovary: implications for disorders of sexual development
Delphine DELACOUR	Institut Jacques Monod (IJM), CNRS UMR7592, Paris	Functional characterization of Spint2 in intestinal morphogenesis – Physiopathological repercussions in the pathogenesis of Congenital Tufting Enteropathy (CTE)
Luc DUPUIS	Inserm U1118, Faculté de médecine, Strasbourg	Generation of an inducible model of amyotrophic lateral sclerosis through conditional truncation of fus/als
Claire FRANCASTEL	UMR7216 CNRS/Université Paris7, Epigenetics and Cell Fate, Paris	Creation and epigenetic/phenotypic characterization of a mouse model for the ICF type II syndrome
Fiona FRANCIS	Inserm UMRS 839, UPMC, Institut du Fer à Moulin, Paris	Molecular and cellular causes, and physiopathology of heterotopia
Laurent GOUYA	Inserm U773, Centre de Recherche Biomédicale Bichat-Beaujon, Paris	Antisense oligonucleotide therapeutic strategy in EPP: Development of a humanized mouse model.
Alain HOVNANIAN	Inserm U781, Hôpital Necker-Enfants Malades, Paris	Development of a murine model overexpressing human Kallikrein 14 in the context of Netherton syndrome
Xavier JEUNEMAITRE	Paris Centre de Recherche Cardiovasculaire (PARCC), Inserm UMR970, Paris	Generation of CUL3 delta-ex9 mouse model reproducing a mendelian form of arterial hypertension
Thierry LEVEILLARD	Institut de la Vision/ Département de Génétique, UMR-S 968, Paris	Inactivation of the thioredoxin-like protein RdCVFL encoded by the Nucleoredoxin-like-1 gene: RdCVFL-/- mouse
Alain LILIENBAUM	Université Paris Diderot – Paris 7, Unité de Biologie Fonctionnelle et Adaptative (BFA) - CNRS EAC 4413, Paris	A mouse model for desmin-related myopathies
Antoine MARTINEZ	GReD (Génétique, Reproduction & Développement), CNRS UMR6293, Inserm U1103, Université de Clermont	Pathogenic potential of R1α truncated mutants found in severe forms of Carney complex
Jean-Jacques MERCADIER	Inserm UMR_S 769 – LabEx LERMIT, Signalisation et Physiopathologie Cardiaque, Faculté de Pharmacie, Chatenay-Malabry	Advances in the understanding and treatment of Catecholamine Polymorphic Ventricular Tachycardia (CPVT)
Michael MITCHELL	Inserm – Aix-Marseille Université, UMR_S 910, Génétique médicale et Génomique fonctionnelle, Marseille	Role of the homologue of a human oligozoospermia factor gene, during mouse spermatogenesis
Rima NABBOUT	Inserm U663, Hôpital Necker Enfants malades, Université Paris Descartes, Neurologie Pédiatrique, Epilepsie de l'enfant et plasticité cérébrale, Paris	KI Mouse model for Migrating partial seizures in infancy
Hamid Reza REZVANI	Inserm U1035, Biothérapies des maladies génétiques et cancers, Bordeaux	Role of NADPH oxidase 1 in Xeroderma pigmentosum C
Isabelle RICHARD	Genethon, Dystrophies musculaires, Evry	New model for evaluation of rescue of sarcoglycan mutations
Frédéric SAUDOU	Institut Curie - CNRS UMR 3306 - Inserm U1005, Signaling Neurobiology and Cancer, Orsay	Huntington's disease: modelling huntingtin proteolysis in mouse (proteo-htt)
Michaël SEBBAGH	UMR 1068 Inserm , UMR 7258 CNRS, Institut Paoli-Calmettes, Centre de Recherche en Cancérologie, Marseille	STRADbeta involvement in Peutz-Jeghers syndrome