Projets lauréats de l'appel à projets 2014 'Mouse models and rare diseases' - Fondation maladies rares / PHENOMIN		
Porteur du projet	**Laboratoire**	**Titre du projet**
Johann Bohm	IGBMC, Dpt. Médecine translationnelle, CNRS Université de Strasbourg UMR 7104 - INSERM U964, Illkirch	First mammalian model for tubular aggregate myopathy and Stormorken syndrome
Pascale Bomont	INSERM U1051, Institut des Neurosciences de Montpellier	Development of an In vivo model for Giant Axonal Neuropathy
Jamel Chelly	IGBMC, Dpt. Médecine translationnelle, CNRS Université de Strasbourg UMR 7104 - INSERM U964, Illkirch	Understanding NEDD4L-related MCD (Malformations of Cortical Development) through investigations of a Knock-In mouse model
Hélène Dollfus	INSERM U1112, Laboratoire de Génétique Médicale, Faculté de Médecine, Strasbourg	Mouse modeling of a missense mutation in the essentiel gene *PIK3R4* (VPS15) responsible for a ciliopathy-like disease
Julie Dumonceaux	Centre de Recherche en Myologie UM76 UPMC - U974 INSERM - FRE 3617 CNRS – Institut de Myologie, Paris	FacioScapuloHumeral Dystrophy (FSHD): targeting two alternative *Fat1* exons with one mouse
Christian Hamel	INSERM U1051, Institut des Neurosciences de Montpellier	*Impg1* gene Knock-Out, a mouse model for human vitelliform macular dystrophy and retinitis pigmentosa
Alain Hovnanian	INSERM U1163, Institut Imagine, Hôpital Necker-Enfants Malades, Paris	Generation of a humanized mouse model for Recessive Dystrophic Epidermolysis Bullosa harbouring a recurrent *COL7A1* mutation
Sébastien Lacroix-Desmazes	INSERM UMRS1138, Centre de Recherche des Cordeliers, 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	INSERM U1163, Institut Imagine, Hôpital Necker-Enfants Malades, Paris	SMAD4 and Myhre syndrome
Delphine Meynard	INSERM U1043, Centre de Physiopathologie de Toulouse-Purpan (CPTP), Toulouse	Is matriptase-2 involved in iron homeostasis regulation and anemia exclusively through the hepatocytes?
Véronique Paquis	Département de Génétique Médicale, IRCAN, UMR 7284 INSERM U1081, Université de Nice Sophia Antipolis, Nice	CHCHD10^{S59L} mouse model: how mitochondrial dysfunction promotes motor neuron disease?
Frédérique René	INSERM U1118, Faculté de Médecine, Strasbourg	Generation of an ALS-FTD mouse model based on a conditional *CHMP2B* intron 5 mutant Knock-In
Frédéric Rieux-Laucat	INSERM U1163, Institut Imagine, Hôpital Necker-Enfants Malades, Paris	Role of LRBA in the control of the immune response: implication in primary immunodeficiencies