



“Models for Rare Diseases”

Partnering platforms booklet

November 2024



Marseille Stem Cell (MaSC)

Localization	Faculty of Medicine 5 th floor - green wing 27 boulevard Jean Moulin 13005 Marseille
Website	https://www.marseille-medical-genetics.org/fr/plateforme-de-reprogrammation-et-differentiation-cellulaire/
Contact	Natacha Broucqsault mmg-plateforme-masc-contact@univ-amu.fr 04 91 31 48 89
Organism(s)	<i>Human</i>
Applications	<ul style="list-style-type: none">• Reprogramming of somatic cells (mainly PBMCs and fibroblasts) into induced pluripotent stem cells. Expertise in rare diseases
Technologies	<ul style="list-style-type: none">• Reprogramming by Sendai Virus transduction• RNA reprogramming (applicable only for fibroblasts)• Episomal reprogramming (applicable only for fibroblasts)• Quality control• Cell banking

Platform director e-mail: Natacha.BROUCQSAULT@univ-amu.fr

Phenomin

Localization	Institut Clinique de la Souris (ICS, Illkirch), Centre for Immunophenomics (CIPHE, Marseille)
Website	http://www.phenomin.fr
Contacts	contact@phenomin.fr
Phone	-
Organism(s)	<i>Mus musculus</i> <i>Rattus norvegicus</i>
Applications	<ul style="list-style-type: none"> • Any kind of mutations from knock-out to full length humanization in mouse • Knock-out, small knock-in and conditional knock-out in rats • Cell engineering • Derivation of ES cells from mutant mouse line • Molecular model validation • Colony expansion and cryopreservation • Phenotypic characterization • Preclinical studies
Technologies	<ul style="list-style-type: none"> • CRISPR/Cas9 technology and/or ES-based methods • PCR, qPCR and digital PCR genotyping • Phenotyping platforms (behavior & cognition, cardiovascular, metabolism, immune-profiling, clinical chemistry & hematology, anatomo-pathology)

Platform director e-mail: herault@iqbmc.fr



TEFOR Paris-Saclay

Localization	TEFOR Paris-Saclay, Institut des Neurosciences, UAR2010 CNRS UPS, Saclay
Website	http://celphedia.eu http://tefor.net
Contacts	services@tefor.net
Phone	-
Organism(s)	<i>Danio rerio</i>
Applications	<ul style="list-style-type: none"> • Knock-Out • Knock-In • Prime editing for disease models with point mutations
Technologies	<ul style="list-style-type: none"> • Zebrafish egg injections • Zebrafish husbandry with automatic feeding (AB, inbred, Casper) • CRISPR/Cas9 technology • Prime editing by modified Cas proteins • Targeted integrations of a single transgene by CRISPR-Cas9 • PCR for genotyping • CRISPANTS • Phenotyping platform (Whole-mount 3D imaging)

Platform director e-mail: johanna.djian@cnrs.fr



PF iPSC Nantes

Localization	Nantes Université, CHU Nantes, Inserm, CNRS, BioCore
Website	https://sfrsante.univ-nantes.fr/plates-formes/modeles-cellulaires-et-geniques/plate-forme-ipsc-nantes-cellules-souches-pluripotentes-induites-1101640.kjsp?RH=1232979193469
Contacts	Laurent David laurent.david@univ-nantes.fr pf-ipsc@univ-nantes.fr
Phone	02 28 08 01 46
Organism(s)	<i>Human</i>
Applications	<ul style="list-style-type: none"> • Production of pluripotent cells from samples of patients with rare diseases, then differentiation into the cell type of interest to mimic the disease
Technologies	<ul style="list-style-type: none"> • Preparation of PBMC from blood samples • Reprogramming somatic cells of patients by transcription factors

Platform director e-mail: laurent.david@univ-nantes.fr

"Caenorhabditis elegans Biology" Facility

Localization	CNRS UAR3453, Université Claude Bernard - Lyon I, Faculté de Médecine et de Pharmacie, 8 avenue Rockefeller, 69008 Lyon
Website	https://www.excitingworms.eu/SEGiCel/
Contacts	Thomas Boulin thomas.boulin@univ-lyon1.fr
Phone	04 26 68 82 81
Organism(s)	<i>Caenorhabditis elegans</i>
Applications	<ul style="list-style-type: none">• Knock-Out and Knock-In strain generation• Mos-engineered mutant strains (KO, KI, tag) development
Technologies	<ul style="list-style-type: none">• CRISPR/Cas9 technology• MosSCI, MosTIC, MosDEL

Platform director e-mail: thomas.boulin@univ-lyon1.fr

TRIP-Nantes

Localization	INSERM UMR 1064, 30, boulevard Jean Monnet, 44093 Nantes BioCore
Website	https://sfrsante.univ-nantes.fr/fr/plateformes/modeles-in-vivo-experimentation-preclinique/plate-forme-trip-gce
Contacts	Ignacio Anegon : ignacio.Anegon@univ-nantes.fr Séverine Ménoret : severine.menoret@univ-nantes.fr
Phone	02 40 08 74 15
Organism(s)	<i>Rattus norvegicus</i> (with inbred or outbred strains)
Applications	<ul style="list-style-type: none"> • Generation of transgenic by small or large (BAC) DNA microinjection • Generation of KO and KI by exon exchange with ssODN or by insertion of DNA construct
Technologies	<ul style="list-style-type: none"> • DNA, ZFNs, TALENs, CRISPR/Cas9, PiggyBac transposon and lentiviral vectors microinjection • Embryo freezing • Q-PCR genotyping (zygosity animals) • Identification of transgenes integration site (LM-PCR) • Electroporation of embryos • Production of animal with conventional health status or with SPF status

Platform directors e-mail: matthieu.giraud@inserm.fr / jerome.Jullien@univ-nantes.fr

For any additional information:

- visit the website: <https://fondation-maladiesrares.org/plateformes-partenariats/>

or

- send an email to: aap-bio@fondation-maladiesrares.com