



**Models for Rare Diseases**

**Partner Platforms Booklet**

**October 2023**

## Phenomin

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

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## TEFOR Paris-Saclay

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<b>Phone</b>	-
<b>Organism(s)</b>	<i>Danio rerio</i>
<b>Applications</b>	<ul style="list-style-type: none"> <li>• Knock-Out</li> <li>• Knock-In</li> <li>• Prime editing for disease models with point mutations</li> </ul>
<b>Technologies</b>	<ul style="list-style-type: none"> <li>• Zebrafish egg injections</li> <li>• Zebrafish husbandry with automatic feeding (AB, inbred, Casper)</li> <li>• CRISPR/Cas9 technology</li> <li>• Prime editing by modified Cas proteins</li> <li>• Targeted integrations of a single transgene by CRISPR-Cas9</li> <li>• PCR for genotyping</li> <li>• CRISPANTS</li> <li>• Phenotyping platform (Whole-mount 3D imaging)</li> </ul>

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## PF iPSC Nantes

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<b>Phone</b>	02 28 08 01 46
<b>Organism(s)</b>	<i>Human</i>
<b>Applications</b>	<ul style="list-style-type: none"> <li>• Production of pluripotent cells from samples of patients with rare diseases, then differentiation into the cell type of interest to mimic the disease</li> </ul>
<b>Technologies</b>	<ul style="list-style-type: none"> <li>• Preparation of PBMC from blood samples</li> <li>• Reprogramming somatic cells of patients by transcription factors</li> </ul>

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**"Caenorhabditis elegans Biology" Facility**

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<b>Phone</b>	04 26 68 82 81
<b>Organism(s)</b>	<i>Caenorhabditis elegans</i>
<b>Applications</b>	<ul style="list-style-type: none"><li>• Knock-Out and Knock-In strain generation</li><li>• Mos-engineered mutant strains (KO, KI, tag) development</li></ul>
<b>Technologies</b>	<ul style="list-style-type: none"><li>• CRISPR/Cas9 technology</li><li>• MosSCI, MosTIC, MosDEL</li></ul>

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## TRIP-Nantes

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<b>Phone</b>	02 40 08 74 15
<b>Organism(s)</b>	<i>Rattus norvegicus</i> (with inbred or outbred strains)
<b>Applications</b>	<ul style="list-style-type: none"> <li>• Generation of transgenic by small or large (BAC) DNA microinjection</li> <li>• Generation of KO and KI by exon exchange with ssODN or by insertion of DNA construct</li> </ul>
<b>Technologies</b>	<ul style="list-style-type: none"> <li>• DNA, ZFNs, TALENs, CRISPR/Cas9, PiggyBac transposon and lentiviral vectors microinjection</li> <li>• Embryo freezing</li> <li>• Q-PCR genotyping (zygosity animals)</li> <li>• Identification of transgenes integration site (LM-PCR)</li> <li>• Electroporation of embryos</li> <li>• Production of animal with conventional health status or with SPF status</li> </ul>

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For any additional information:

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

or

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