amU Institut Marseille maladies rares Aix Marseille Université

PHD PROGRAM

2025

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I.GENERAL INFORMATION

A new program

The Marseille Rare Diseases institute - MarMaRa- launches a PhD program for 2025. The program is divided into rare diseases and cross-cutting trainings.

These trainings are opened in priority to PhD students but if the maximum capacity of attendees is not reached, Master students, engineers and researchers are welcomed.

After each training you will receive a certificate of attendance and AMU PhD students can have up to 7h/training validated by their Doctoral School.

RARE DISEASES TRAININGS

- Rare disease Ecosystem (planned for 2026)
- Models and Pre-clinical Development for Rare Diseases (planned for 2026)
- Biomedical ethics (planned for 2026)
- Research data management (planned for 2026)
- Fundamental notions in Medical Genetics (half day - 7 May 2025)

CROSS-CUTTING TRAININGS

- Introduction to microscopy for cell and tissue imaging (1 day - 12 March 2025)
- Statistics for genomics: continuous probability distributions (half day - 11 March 2025)
- Statistics for genomics: discrete probability distributions (1 day - 28 May 2025)
- Workshop on Single cell RNA seq (1 day - 27 May 2025)

II. RARE DISEASES TRAININGS

II.1.Rare Diseases ecosystem

II.2.Models & Pre-clinical Development for Rare Diseases

II.3.Biomedical ethics

II.4.Research data management

II.5.Fundamental notions in Medical Genetics

A HIGHLY RECOMMENDED MODULE !

II.1.RARE DISEASE ECOSYSTEM

COORDINATOR: LAURENT VILLARD

Objectives

This module aims to showcase the actors of the rare disease ecosystem in France and in Europe.

More details

Prerequisites: none.

Langage: English

Duration: 12h (4 half-days)

Date: This training is planned for 2026 registration and details will be shared later

Format: hybrid learning

Program

Speakers(s):

-**Thierry Brue** (AMU - AP HM)
-**Paul Gimenès / Elodie Crépieux** (Alliance Maladies Rares)
-**Diana Désir-Parseille / Daniel Scherman** (Fondation Maladies Rares)
-**Charlotte Rodwell** (Orphanet)
-**Anne-Sophie Lapointe** (DGOS - Ministère des Solidarités et de la Santé)
-**Christel Thauvin-Robinet** (AVIESAN et Centre de Génétique de Dijon)
-**Marion Mathieu / Jean Thimonnier** (Tous Chercheurs)
-**Catherine Barthelemy** (Inserm - GRAM)
-**Daria Julkowska** (EJPRD)
-**Virginie Bros-Facer / Gulcin Gumus** (Eurordis)

Content of the training:

Training sequence 1: Association and foundations
-Alliance Maladies Rares
-Fondation Maladies Rares
-Orphanet

Training sequence 2: French Institutional Organization
-Plan National Maladies Rares (PNMR4).
-Plan France Médecine Génomique 2025 (PFMG 2025).
-Organization of the care (Sectors, Reference Centers, PNDS).

Training sequence 3: Training and Mediation
-Tous Chercheurs.
-Groupe de Réflexion avec les Associations de Malades (GRAM).
-Online ressources.

Training sequence 4: European actors
-European Rare Diseases Research Alliance (ERDERA).
-European Research Networks (ERNs).
-Rare Diseases Europe (Eurordis).

II.2.MODELS AND PRE-CLINICAL DEVELOPMENT FOR RARE DISEASES

COORDINATOR(S): FRANCESCA ROCHAIS & STÉPHANE ZAFFRA

Program

Speakers(s):

-Fabienne Lescroart - Organoids from heart and skeletal muscle to study rare diseases

-Stéphane Bot/Ines Barthélémy - Canine model of Duchenne Muscular Dystrophy

-Laurence Legeai-Mallet - Zebrafish model of Achondroplasia and clinical trials

-Antoine Muchir - Pharmacological approaches for the treatment of LMNA-related cardiomyopathies. Mouse models

-Laurent Villard - Pharmacological approaches for the treatment of genetic related epilepsies - clinical trials

-Marc Bartoli - Gene therapy and mouse models of muscular dystrophies

Content of the training:

-Organoids from heart and skeletal muscle to study rare diseases.

-Canine model of Duchenne Muscular Dystrophy.

-Zebrafish model of Achondroplasia and clinical trials.

-Pharmacological approaches for the treatment of LMNA-related cardiomyopathies. Mouse models.

-Pharmacological approaches for the treatment of genetic related epilepsies - clinical trials.

-Gene therapy and mouse models of muscular dystrophies.

-Industry and drug repositioning.

Objectives

This workshop aims to propose an overview of the different experimental models, including animal and in vitro models, used to study rare diseases. Additionally, preclinical studies utilizing diverse therapeutic approaches (pharmacological and gene transfer approaches) will be presented. Finally, the industrial point of view in the development of therapies for rare diseases will be addressed.

More details

Prerequisites: none

Langage: English

Duration: 10h-12h

Date: This training is planned for 2026 registration and details will be shared later

Format: hybrid learning

II.3.BIOMEDICAL ETHICS

COORDINATOR: ANNAGRAZIA ALTAVILLA

Program

Speakers(s):

-**Perrine MALZAC**, Département de génétique Médicale et Espace de réflexion éthique PACA-Corse, AP-HM / Aix Marseille Univ, CNRS, EFS, ADES, UMR 7268 / Marseille, France

-**Annagrazia ALTAVILLA**, Espace de réflexion éthique PACA-Corse, AP-HM / Réseau européen d'excellence TEDDY

-**Marion MATHIEU**, Tous chercheurs / Aix Marseille Univ, CNRS, EFS, ADES, UMR 7268 / Marseille, France

Content of the training:

-**Ethic and fondamental rights** (2 hours) – English – Annagrazia ALTAVILLA.

-**Clinical research ethics, CPP, CCNE and other ethics committees** (2 hours) – French – Perrine MALZAC.

-**Ethics applied to concrete cases of rare diseases** (2 hours) – French - Perrine MALZAC, Annagrazia ALTAVILLA, Marion MATHIEU.

Objectives

First, we will describe the legal framework and the principles of biomedical ethics. Secondly, we will learn about the structures responsible for producing opinions and mobilizing ethical reflection in France. Finally, we will approach the practice of ethics by learning to reason from concrete examples and with the help of the tools previously described

More details

Prerequisites: none

Langage: English

Duration: 6h

Date: This training is planned for 2026 registration and details will be shared later

Format: hybrid learning

II.4.RESEARCH DATA MANAGEMENT

COORDINATOR: FRANCESCA ROCHAIS

Program

Speakers(s) :

-**Fréderic de Lamotte** - INRAE - IFB, Montpellier agronomie

-**Mouron Philippe** - LID2MS

-**Stéphane Bergamini** - SATT Sud Est - Directeur du Transfert de Technologies

-**Caroline S Donati (Isabelle Gras)** - DGS AMU

Content of the training :

-**Intervention 1:** Introduction; what is data; what is around data; quality implementation (2h).

-**Intervention 2:** FAIR training in data management, metadata, storage reproducibility, accessibility (2h).

-**Intervention 3:** Research data protection (2h).

-**Intervention 4:** Intellectual property - patent filing – (2h).

-**Intervention 5:** Data dissemination; Open Science (2h).

Objectives

This workshop aims to have a global overview of Data Management in Research. It will thus cover a wide area including the secure collection of data, the safe storage and archiving of data, the protection of data and data communication such as open access.

More details

Prerequisites: none

Langage: English

Duration: 12h (two days)

Date: This training is planned for 2026 registration and details will be shared later

Format: hybrid training

Program

Speakers(s):

Mario Abaji - MMG

Content of the training:

-Cells, Genomes, DNA and Genes.

-Types of Genetic Disease.

-Laws of Inheritance.

-Genetic Variation.

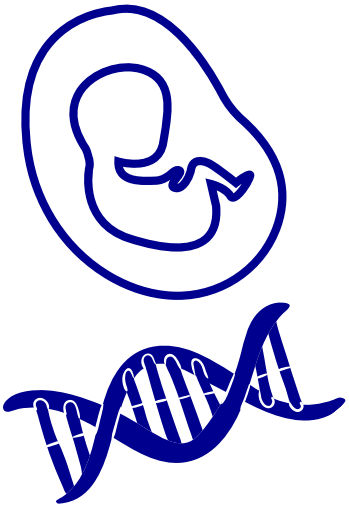
-Diagnosis.

II.5.FUNDAMENTAL NOTIONS IN MEDICAL GENETICS

COORDINATOR: MARIO ABAJI

Objectives

This session provides fundamental information about basic genetics concepts, including cell structure, the molecular & biochemical basis of disease, major types of genetic disease, laws of inheritance, mosaicism, polysomy, polyploïdy, parental digenism, oligogenism and the impact of genetic variation. a particular emphasis will be addressed for diagnosis of rare diseases, diagnostic wandering, diagnostic deadlocks.



More details

Prerequisites: none

Langage: English

Duration: half a day (9h-12h30)

Date: 7th May, 2025

Format: remote learning

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III. CROSS-CUTTING TRAININGS

III.1.Introduction to microscopy for cell & tissue imaging

III.2.Statistics for genomics: continuous probability distributions

III.3.Statistics for genomics: discrete probability distributions

III.4.Workshop on Single-cell RNA-seq

III.1. INTRODUCTION TO MICROSCOPY FOR CELL AND TISSUE IMAGING

COORDINATOR(S) : MANOS MAVRAKIS

Program

Speakers(s):

-**Serge Monneret** (DR CNRS, institut Fresnel)

-**Sophie Brasselet** (DR CNRS, institut Fresnel)

Content of the training:

-The course consists of a morning and an afternoon session of 2.5 hours each, followed by a 2-hour demonstration of cutting-edge microscopy techniques.

- basic notions of optics related to image formation and microscopy.
- principles of fluorescence, two-photon and nonlinear microscopy.
- applications in cell and tissue imaging

Objectives

This course will cover how light-matter interactions lead to the formation of images of cells and tissues in a microscope (eg, diffraction,refraction, scattering), will detail key optics notions that affect image formation (eg, resolution, numerical aperture, aberrations), and provide an overview of the main microscopy techniques used in biology and their applications to cell and tissue imaging.

More details

Prerequisites: none

Langage: English and/or French

Duration: 1 day

Date: 12th March, 2025

Format: face-to-face training, Institut Fresnel, Marseille

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Program

Speakers(s):

-Aïtor Gonzalez - TAGC

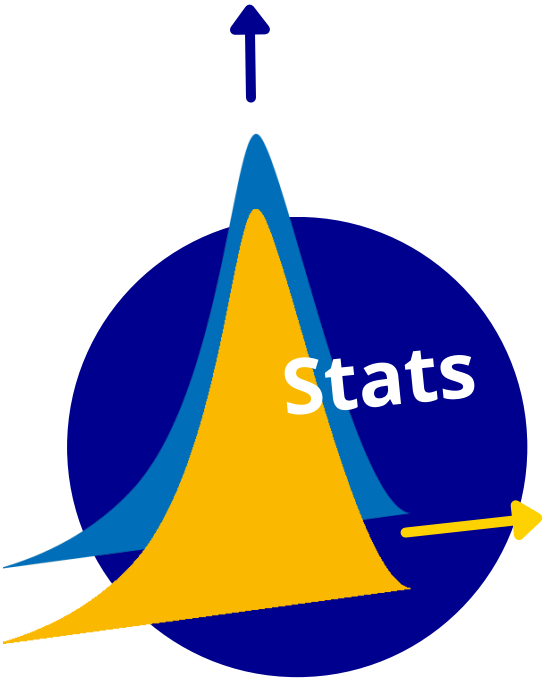
Content of the training:

-**Continuous probability distribution** (normal, Student's t-distribution)

-**Hypothesis testing** (t-test, ANOVA).

III.2.STATISTICS FOR GENOMICS: CONTINUOUS PROBABILITY DISTRIBUTIONS

COORDINATOR: AÏTOR GONZALEZ



Objectives

The objective of this workshop is to propose an introduction to popular statistical methods and tests used in the field of genomics. The course will typically cover continuous probability distribution (e.g. normal, t distribution), and procedures of hypothesis testing (t-test, ANOVA). Examples will be typically taken from the field of biology (e.g. differential gene expression). Practical sections will be performed in R language. However only basic instructions will be used and, thus, no particular skill in informatics should be required.

More details

Prerequisites: none

Langage: English

Duration: 7h

Date: 11th March, 2025

Format: remote learning (Zoom)

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Program

Speakers(s):

-Denis Puthier - TAGC

Content of the training:

-Discrete probability distribution (binomial, Poisson, hypergeometric and negative binomial distribution).

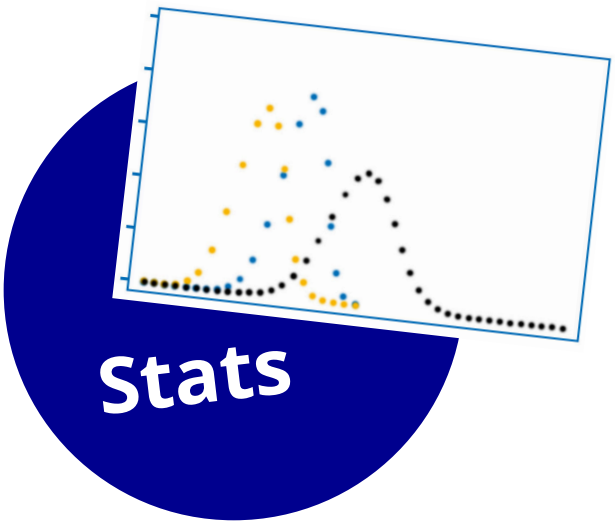
-Hypothesis testing (Fisher's Exact test and hypergeometric test).

III.3.STATISTICS FOR GENOMICS: DISCRETE PROBABILITY DISTRIBUTIONS

COORDINATOR: DENIS PUTHIER

Objectives

The objective of this workshop is to propose an introduction to popular statistical methods and tests used in the field of genomics. The course will typically cover discrete probability distributions (e.g. binomial, hypergeometric and negative binomial distribution), and hypothesis testing (e.g. Fisher's Exact test and hypergeometric test). Examples will be typically taken from the field of biology (e.g. functional enrichment analysis). Practical sections will be performed in R language. However only basic instructions will be used and, thus, no particular skill in informatics should be required.



More details

Prerequisites: none

Langage: English

Duration: 7h

Date: 28th May, 2025

Format: remote learning (Zoom)

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III.4.WORKSHOP ON SINGLE-CELL RNA-SEQ

COORDINATOR: DENIS PUTHIER

Program

Speakers(s):

-**Denis Puthier - TAGC**

Content of the training:

-Galaxy servers.

-Fastq format.

-Quality control of sequencing data.

-Trimming.

-Splice-aware aligners.

-Mapping quality.

-Abundance estimation and differential expression.

Objectives

In this workshop you will learn how to analyse a typical RNA-seq experiment with a known reference genome. The objective will be to cover steps starting from raw data (fastq) to quantification and differential gene expression analysis.
This workshop will essentially focus on the bioinformatic tools and algorithms used to construct a workflow. The workflow will be produced using a Galaxy server so attendees are not supposed to have any programming skills.

More details

Prerequisites: none

Langage: English

Duration: 7h

Date: 27th May, 2025

Format: remote training (Zoom)

Scan me to register

REGISTRATION

Online survey link

If you are interested in one or more of our courses, please, click on the corresponding QR code or link.
More information on:

Institute MarMaRa website - PhD Program

Do not hesitate to contact us if you have any questions:

- **Denis Puthier**, deputy director for education
  denis.puthier@univ-amu.fr
- **Marie-Cécile Gaillard**, project officer
  marie-cecile.gaillard@univ-amu.fr

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Aix-Marseille University
2025

MarMaRa institute

NEW TRAININGS AVAILABLE

PhD PROGRAM

amU MarMaRa Aix Marseille Université

The Marseille Rare Diseases Institute launches its 2025 PhD program
Discover below the first trainings open for registration !

📢 The Marseille Rare Diseases institute - MarMaRa - PhD program 2025

These trainings are opened in priority to PhD students but Master students, engineers and researchers are welcomed. Our trainings are in English (unless all participants are French-speaking) and almost all are remote.
European and international students are welcome!

After each training you will receive a certificate of attendance and PhD students of AMU can have up to 7h/training validated by their Doctoral School.

- 11/03/25 Statistics for genomics: continuous probability distributions (remote)
- 12/03/25 Introduction to microscopy for cell and tissue imaging (face-to-face)
- 07/05/25 Fundamental notions in medical genetics (remote)
- 27/05/25 Single cell RNA-seq analysis (remote)
- 28/05/25 Statistics for genomics: discrete probability distributions (remote)

[Registration open] Statistics for genomics: continuous probability distributions

The objective of this workshop is to propose an introduction to popular statistical methods and tests used in the field of genomics. The course will typically cover continuous probability distribution (e.g. normal, t distribution) and procedures of hypothesis testing (t-test, ANOVA). Examples will be typically taken from the field of biology (e.g. differential gene expression). Practical sections will be performed in R language. However only basic instructions will be used and, thus, no particular skill in informatics should be required.

📍 Where ? remote learning (Zoom)

📅 When ? **Tuesday 11th March, 2025**

📌 Registration: https://amuform.univ-amu.fr/stat-for-genomics-continuous2025

[Registration open] Introduction to microscopy for cell and tissue imaging

This course will cover how light-matter interactions lead to the formation of images of cells and tissues in a microscope (eg,diffraction, refraction, scattering), will detail key optics notions that affect image formation (eg resolution, numerical aperture, aberrations), and provide an overview of the main microscopy techniques used in biology and their applications to cell and tissue imaging.

📍 Where ? face-to-face, Institut Fresnel,
52 Avenue Escadrille Normandie Niemen, 13013 Marseille

📅 When ? **Wednesday 12th March, 2025**

📌 Registration: https://amuform.univ-amu.fr/introduction-to-microscopy2025

[Registration open] Fundamental notions in Medical Genetics

This session provides fundamental information about basic genetics concepts, including cell structure, the molecular & biochemical basis of disease, major types of genetic disease, laws of inheritance, mosaicism, polysomy, polyploid, parental digenism, oligogenism and the impact of genetic variation. A particular emphasis will be addressed for diagnosis of rare diseases, diagnostic wandering, diagnostic deadlocks.

📍 Where ? remote learning (Zoom)

📅 When ? **Wednesday 7th May, 2025**

📌 Registration: https://amuform.univ-amu.fr/fundamental-genetics2025

[Registration open] Statistics for genomics: discrete probability distributions

The objective of this workshop is to propose an introduction to popular statistical methods and tests used in the field of genomics. The course will typically cover discrete probability distributions (e.g. binomial, hypergeometric and negative binomial distribution), and hypothesis testing (e.g. Fisher's Exact test and hypergeometric test). Examples will be typically taken from the field of biology (e.g. functional enrichment analysis). Practical sections will be performed in R language. However only basics instructions will be used, and, thus, no particular skill in informatics should be required.

📍 Where ? remote learning (Zoom)

📅 When ? **Wednesday 28th May, 2025**

📌 Registration: https://amuform.univ-amu.fr/stat-for-genomics-discrete2025

[Registration open] Workshop on Single-cell RNA-seq

In this training, participants will learn about the fundamentals of single-cell analysis using Seurat. Beginning with pre-processing steps, attendees will learn techniques used for quality control, normalization, and data scaling to ensure robust downstream analyses. They will then explore dimensionality reduction methods and clustering to visualize complex cellular landscapes. Finally, they will learn about differential gene expression analysis, unravelling the molecular signatures that define cellular diversity and response to experimental stimuli. The tutorial will be conducted within the R/RStudio environment. While prior familiarity with the language is advantageous, participants can also focus on understanding the diverse steps involved in the analysis process.

📍 Where ? remote learning (Zoom)

📅 When ? **Tuesday 27th May, 2025**

📌 Registration: https://amuform.univ-amu.fr/single-cell-RNAseq2025

If you have any question, please contact us:

- Denis Puthier, Deputy director for education (denis.puthier@univ-amu.fr)
- Marie-Cécile Gaillard, Project officer (marie-cecile.gaillard@univ-amu.fr)

And stay tuned ! Other trainings will be available in 2025-2026 ...

Rare diseases Ecosystem in EU
Therapeutic and Molecular approaches used in Rare Diseases
Biomedical ethics
Research data managment