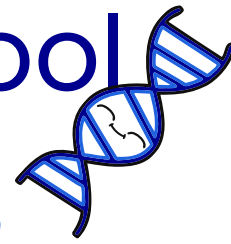




# MarMaRa Summer School

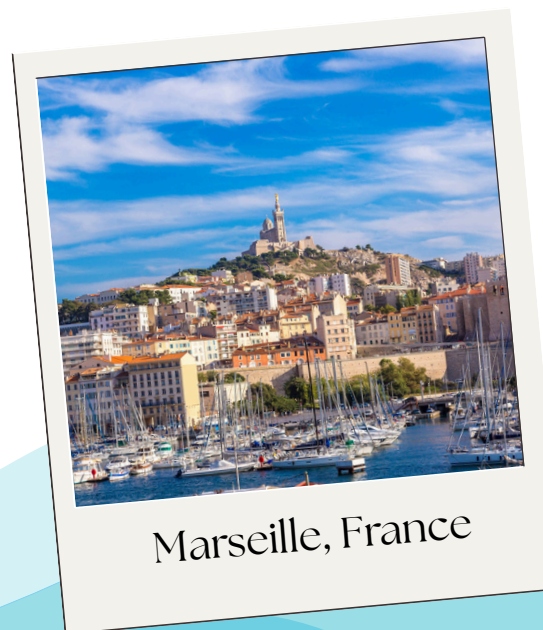


*Blended Intensive Program*

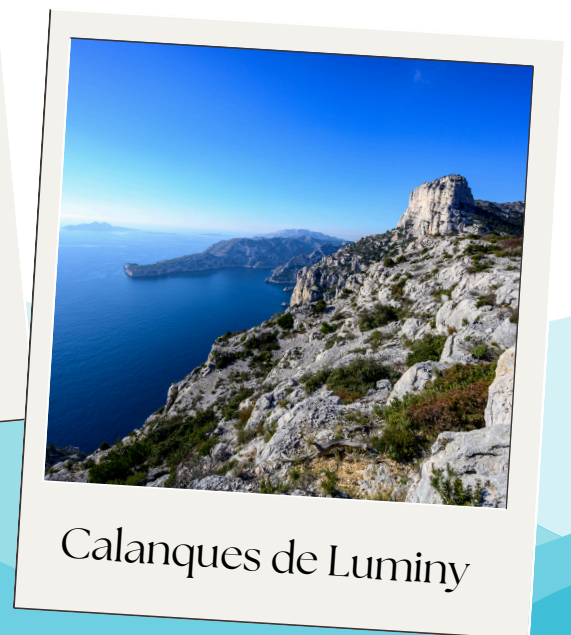
**"Rare Diseases at the Omics era:  
Current tools for frequent challenges"**

**23 - 27 June, 2025**

**Luminy campus**



Marseille, France



Calanques de Luminy



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## MarMaRa Summer School at Hexagone, Luminy Campus

### "Rare Diseases at the Omics era: Current tools for frequent challenges"

New sequencing methods represent a major advance for **the study of rare diseases**. However, it is becoming increasingly important to **integrate data** from multiple sources to improve the **molecular analysis** and **diagnosis** of these diseases.

**The objective** of the "Rare diseases at the Omics era: current tools for frequent challenges" is to present the contribution of sequencing and multi-omics methods to rare disease analyses.

During this summer school, you will discover **rare diseases** and how **sequencing methods**, associated with **multi-omics approaches**, can be used to describe them from a molecular point of view and contribute to the diagnosis and management of patients.

#### Day 1 - Monday 23 June 2025

The school will start with **an introduction on rare diseases** (ecosystem, technological approaches, human and social sciences approach). The afternoon will focus on **variants analysis** and **sequencing technologies**.

#### Day 2 - Tuesday 24 June 2025

The 2nd day is about **single cell and transcriptomics** in practice, the first part will be dedicated to **sequences quality control & alignements** for single cell technologies using **Galaxy**. A second part is dedicated to **cell population heterogeneity analysis** using **Monocle tutorial in R**.

#### Day 3 - Wednesday 25 June 2025

The 3rd day will focus on the advent of **spatial transcriptomics** including an overview of the spatial transcriptomics technologies, an example of a **cardiac development cartography** using **ATLAS**. The afternoon will be focused on **data analysis challenge** and the use of **Tech Visium** and **Merscope** tools.

#### Day 4 - Thursday 26 June 2025

The 4th day will include a first session on **epigenomics dynamic & regulation** (secondary DNA structures, CHIPseq analysis, G-quadruplexes) and the second part will focus on examples of **genome (dys)regulation in Rare Diseases** involving **enhancers** elements, pathologies of **repeated sequences or DNA repair machinery**.

#### Day 5 - Friday 27 June 2025

The last day will overviewed **IPSCs technologies and their transformative impact on Rare Diseases** research through various examples (cardiac chanelopathies using iPSCs, BBB pharmacological approaches in RD...).

#### Contact

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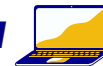
# MarMaRa Summer school

**"Rare Diseases at the Omics era:  
Current tools for frequent challenges"**

**P R O G R A M**

**23–27 June 2025**

**Hexagone auditorium  
informatics room EO.11, TPR1  
Luminy Campus**



**Monday, 23 June, 2025**

## **SESSION I : Introduction to rare diseases (09:15 – 12:00)**

- 09:15–09:45** Introduction to rare diseases, *Frédérique Magdinier*
- 09:45–10:45** The IGPrare project: a collaborative, multidisciplinary and transversal research in the field of rare diseases, *Sandrine de Montgolfier*
- 10:45–11:00 *Break*
- 11:00–12:00** Next Generation Sequencing Technologies: an overview, *Valérie Delague*
- 12:00–13:00 *Lunch Break*

## **SESSION II : Advanced Notion in Medical Genetics (13:00– 17:00)**

- 13:00–13:45** Practical aspects in medical genetics, *Mario Abaji*
- 13:45–14:45** Challenges in rare disease diagnostics, *Svetlana Gorokhova*
- 14:45–15:00 *Break*
- 15:00–17:00** Hands-on session on genetic variant analysis, *Svetlana Gorokhova & Mario Abaji*



## Tuesday, 24 June, 2025

### SESSION III : Single cell transcriptomics (09:15 - 12:00)

**09:15-10:45** **Sequence quality control & alignements (using Galaxy)**, Aarathy Geetha & Denis Puthier 

10:45-11:00 *Break*

**11:00-12:00** **Sequence quality control & alignements (using Galaxy)**, Aarathy Geetha & Denis Puthier 

12:00-13:00 *Lunch break*

### SESSION IV : Single cell transcriptomics (practical) (13:00 - 17:00)

**13:00-14:45** **Cell population heterogeneity analysis using Monocle tutorial (R/RStudio)**, Aarathy Geetha & Denis Puthier 

14:45-15:00 *Break*

**15:00-17:00** **Cell population heterogeneity analysis using Monocle tutorial (R/RStudio)**, Aarathy Geetha & Denis Puthier 

18:00-22:00 *Social Group evening*

## Wednesday, 25 June 2025

### SESSION V: The advent of spatial transcriptomics (09:15 - 12:00)

**09:15-10:00** **Method Overview/ Introduction to spatial transcriptomics technologies**, Peter Krenn (*remote*)

**10:00-10:45** **Constructing a human heart atlas of the first gestational trimester using spatial transcriptomics and single-nucleus RNA-sequencing**, Christopher De Bono

10:45-11:00 *Break*

**11:00-12:00** **Spatial transcriptomics ATLAS online**, Denis Puthier & Peter Krenn 

12:00-13:00 *Lunch break*

### SESSION VI: Spatial transcriptomics using Tech visium (13:00 - 17:00)

**13:00-14:00** **Data analysis challenge in the fields of Spatial Transcriptomics**, Paul Villoutreix

14:00-14:30 *Break*

**14:30-17:00** **Practical session about Spatial Transcriptomics (Visium LD/HD, Merscope)**, Denis Puthier & Paul Villoutreix 



## Thursday, 26 June 2025

### SESSION VII: Epigenome dynamic & regulation (09:15 - 12:00)

**09:15-10:00** Introduction about epigenetics dynamic / Investigation of DNA secondary structures in rare diseases, *Angelika Lahnsteiner*

**10:00-10:45** **ChIP-seq analysis how to analyse epigenetics markers and data,** *Angelika Lahnsteiner*



10:45-11:00 *Break*

**11:00-12:00** **Hands-on exploration of G-quadruplexes (G4s) using R,** *Angelika Lahnsteiner*



12:00-13:00 *Lunch break*

### SESSION VIII: Examples of genome (dys)regulation (13:00 - 17:00)

**13:00-14:15** **Enhancer dysregulation in diseases,** *Aitor Gonzalez*

**14:15-15:30** **Pathology associated to telomeres repeated elements - telomeropathies,** *Stéphane Coulon*

15:30-15:45 *Break*

**15:45-17:00** **Genome integrity (Faconi anemia) DNA repair machinery,** *Christophe Lachaud*

## Friday, 27 June 2025

### SESSION IX: iPSC Technologies and Their Transformative Impact on Rare Disease Research (09:00 - 13:00)

**09:15-10:45** **iPSC applied to rare diseases. Introduction,** *Leslie Caron* & **technologies application facility,** *Ola Hadadeh*

10:45-11:00 *Break*

**11:00-12:00** **Introduction to cardiac channelopathies, basics and clinical aspects. Phenotypic analysis to diagnosis and principles of therapy using iPSC cardiomyocytes,** *Bogdan Amuzescu*

**12:00-13:00** **Blood brain barrier physiology; Disruption of BBB in RD; BBB in pharmacological approaches targeting rare diseases,** *Beatrice Radu*

13:00-14:00 *Picnic lunch*

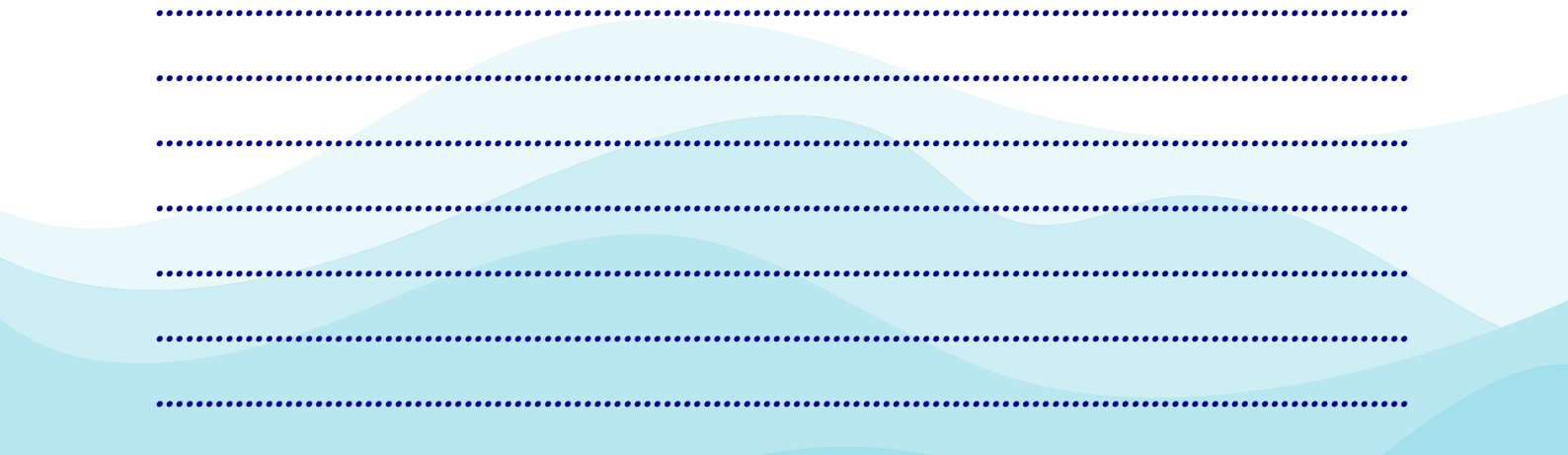
*Social group event "Les calanques" guided hike (14:00 - 18:00)*





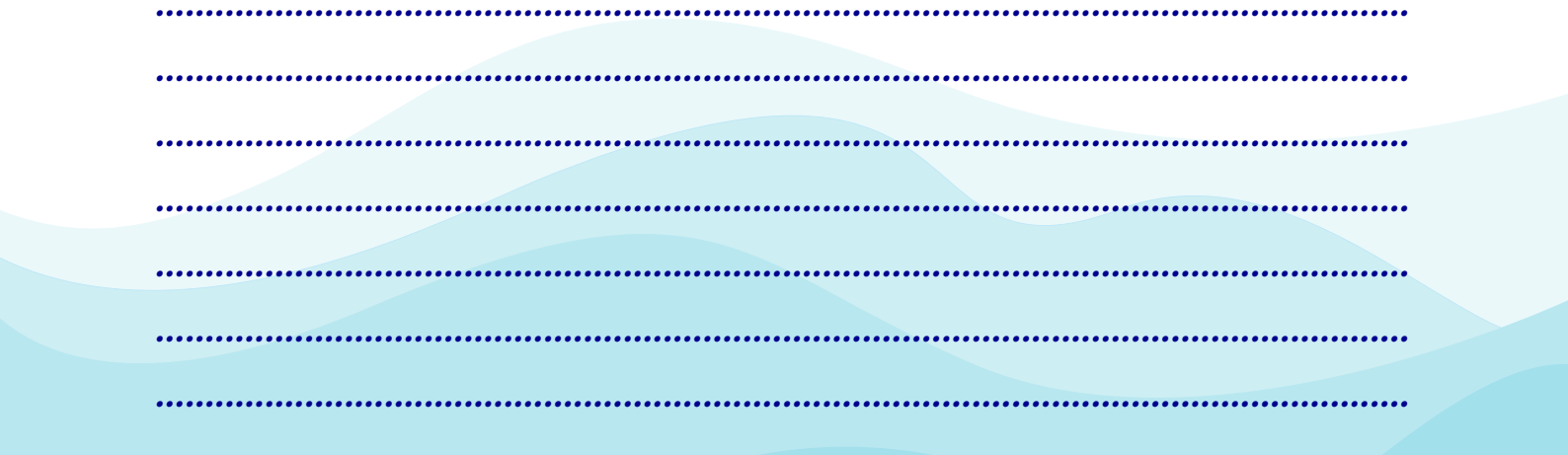


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# MarMaRa Summer school

## TRAVEL & LOCATION



### > Marseille Provence airport

**Bus shuttle L91**  **everyday**

Airport <> Marseille Gare Saint Charles (train station)



### > Luminy campus



**Métro M2**  **everyday**

Gare St Charles > Métro rond point du Prado

Métro Rond point du Prado > Gare St Charles



**Bus B1**  **everyday (limited service during WE)**

Métro Rond point du Prado/Castellane > Luminy PN les calanques

Luminy PN les calanques > Métro Rond Point du Prado/Castellane



**Bus 21 JET**  **Monday <> Friday only**

Métro rond point du Prado > Luminy faculté

Luminy faculté > Métro rond point du Prado



**Bus 24**  **everyday (limited service during WE)**

Métro Ste Marguerite Dromel <> Luminy PN les calanques

Luminy PN les calanques <> Métro Ste Marguerite



- Public Transportation tickets will be provided to all participants
- All the bus/metro timetables/frequency on the RTM website (<https://www.rtm.fr/horaires>) [in French]





# ACCOMODATION INFORMATIONS

## ✓ CHECK IN

**from Sunday 22nd June 2025, PM**

arrival to Luminy campus BUS stop/ Students residency

 Dinner, CROUS restaurant (7:30–8:30PM)

## **MONDAY 23rd – FRIDAY 27th June**

 **Breakfast, CROUS restaurant, 8–9AM**

 **Lunch, CROUS restaurant, self service, 12AM–1PM**  
*except 28/06 > picnic lunch outside*

 **Dinner, CROUS restaurant, self service, 7PM**  
*except 24/06 > social evening in city center (dinner not included)*

## ✓ CHECK OUT

**by Saturday 28th June 2025, 11 AM**

Luminy campus, students residency

 Breakfast, CROUS restaurant (7–8AM)

<https://sciences.univ-amu.fr/en/sites-campuses/marseille-site-luminy>



### Contact

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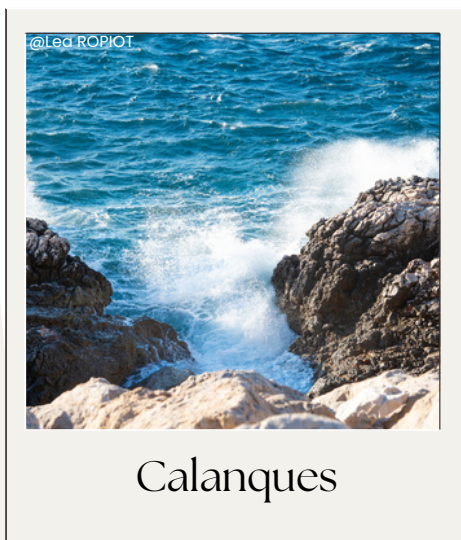
# SOCIAL ACTIVITIES

## Hiking to the Belvedere, calanque de Sugiton, Luminy



**Friday 27 June 2PM ~6PM**

*Luminy campus parking*



- Good walking shoes
- 2L water minimum per person  
*(no water in the park)*
- Sunscreen
- Backpack
- No swimming
- Sunglasses



## Quiz room Marseille

- *Departure to the city center (bus) 5:45PM*
- *Drinks & dinner 7PM*

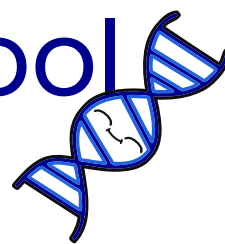


**Tuesday 24 June, 8:30PM**  
*9 Rue Dragon, 13006 Marseille*





# MarMaRa Summer School



*Blended Intensive Program*

**"Rare Diseases at the Omics era:  
Current tools for frequent challenges"**



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