



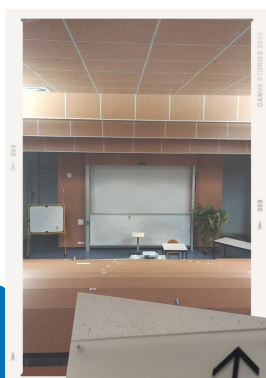
Institut
Marseille
Maladies rares
Aix-Marseille Université

MarMaRa Summer School

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

11 - 13 July, 2022

Online registration (free) : columbo.univ-amu.fr/index.php/664611?lang=fr



 Polytech, Luminy, Marseille



The Calanques of Marseille





The 1st MarMaRa Summer school at Polytech, 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.

Who can apply ?

The school is open **to all, free of charge, with priority** to students and young researchers (Master, PhD students, engineers, etc).

What will you learn ?

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 11 July:

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

Day 2 – Tuesday 12 July:

The 2nd day will open with a session on **single cell and transcriptomics** (experimental & bioinformatics of single cell technologies; single cell application to cardiac development analysis). **A practical session** is planned for the afternoon (limited places).

Day 3 – Wednesday 13 July:

The last day will include a session on **epigenetics** (pathologies of repeated sequences, single-cell epigenomics, omics data integration).

The Summer School will end with **a hike in the Calanques**, easily accessible from the Luminy campus.

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

11-13 July 2022

Jacques Derrien amphitheater , Polytech, Luminy Campus

Monday, July 11, 2022

8:30-09:00 **Welcome coffee**

09:00-09:15 **Introduction**
Denis Puthier, Deputy Director for Education

SESSION I : Introduction to rare diseases (09:15 - 13:45)

Moderator & organizer : Marc Bartoli

09:15-10:00 **Introduction to rare diseases**, Thierry Brue

10:00-11:00 **Next Generation Sequencing Technologies: an overview**, Valérie Delague

11:00-11:15 **Coffee break**

11:15-12:15 **The IGPrare project: a collaborative, multidisciplinary and transversal research in the field of rare diseases**, Marion Mathieu

12:15-13:45 **Lunch break**

SESSION II : Variants, sequencing technologies (13:45 - 17:00)

Moderator : Martin Krahn

13:45-14:30 **How to resolve diagnostic deadlocks**, Laurence Faivre

14:30-15:30 **Functional annotation and analysis of variants**, Valérie Delague

15:30-15:45 **Break**

15:45-16:30 **Diagnosing rare diseases using high-throughput sequencing**, Svetlana Gorokhova

16:30-16:45 **End of the 1st day**, Denis Puthier

Tuesday, July 12, 2022

SESSION III : Single cell, transcriptomics (09:00 - 17:00)

Moderators : Sandrine Marquet & Valérie Delague

- 09:00-10:30 Introduction to single-cell RNA-seq methods and data analysis,**
Pierre Milpied & Lionel Spinelli
- 10:30-10:45 Coffee break**
- 10:45-11:30 Studying the earliest steps of cardiac development at the single cell level,**
Fabienne Lescroart
- 11:30-12:15 Human cell atlas. - Application of spatial transcriptomics in cardiac development,** Stéphane Zaffran
- 12:15-13:45 Lunch break**
- 13:45-15:30 Single cell technologies practical (session I),**
Pierre Milpied, Lionel Spinelli, Denis Puthier, Céline Chevalier
- 15:30-15:45 Break**
- 15:45-17:00 Single cell technologies practical (session II),**
Pierre Milpied, Lionel Spinelli, Denis Puthier, Céline Chevalier

Wednesday, July 13, 2022

SESSION IV : Epigenetics (09:00 - 17:00)

Moderator & organizer : Denis Puthier

- 09:00-10:00 Mendelian disorders of the epigenetic machinery,** Julien Van Gils
- 10:00-10:45 Pathology associated with repeated sequences: Facio Scapulo Humeral Dystrophy, the "perfect model",** Jérôme Robin
- 10:45-11:00 Coffee break**
- 11:00-12:00 Studying the role of enhancer dysregulation in diseases,** Salvatore Spicuglia
- 12:00-13:30 Lunch break**
- 13:30-14:30 Studying epigenetics landmarks with single-cell techniques,** Christophe Becavin
- 14:30-15:30 OMICS data integration,** Anaïs Baudot
- 15:30-16:00 General discussion**
- 15:30-17:00 Social event, a hike in the Calanques**