



ANNUAL SYMPOSIUM

*Artificial Intelligence:
what future for Rare Diseases ?*



**19 & 20 May
2025**

**Grisoli
Amphitheater**
Faculty of Medical &
Paramedical Sciences
TIMONE campus

**Free but mandatory
registration**



1st day | 19 MAY 2025

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

09:30-10:00 **MarMaRa news**, Frédérique Magdinier (RST) ; Denis Puthier, Deputy Director for Education; Françoise Muscatelli & Laurent Fasano co-Deputy Directors for Research

Opening lecture

10:00-10:50 ERDERA: Powering the Future of Rare Disease Research – Data, Collaboration, Innovation, [Daria Julkowska](#), IT GGB INSERM

Session I - Data integration, genomic medicine & AI

Moderator:

11:00-11:30 **AI and data integration to study Rare Genetic Diseases**, [Anaïs Baudot](#), MMG

11:30-12:00 **Language models and rare diseases: how artificial intelligence contributes to diagnosis and patient care**, [Marie de Tayrac](#), CHU Ponchaillou, Rennes

12:00-12:30 **Title TBD**, [Julien Thevenon](#), CHU de Grenoble

12:30-14:00 **Lunch break - Poster session**

Interdisciplinary lecture

14:00-14:50 *L'IA est-elle un risque pour l'Homme ?* [Marius Bertolucci](#), IMPGT
[presentation in french]

Session II - Throwback: incentive action projects

Moderator:

15:00-15:15 **Dissecting how retinoic acid signaling controls myocardial progenitor and neural crest cell contributions to the developing heart**, [Christopher De Bono](#), MMG

15:15-15:30 **Epigenetic modifications in two types of laminopathies: lipodystrophies and progeroid syndromes**, [Camille Desgrouas](#), C2VN & [Diane Frankel](#), MMG

15:30-15:45 **Development of a novel gene therapy targeting adipose tissue leptin production to treat Rett syndrome**, [Jean-Luc Gaiarsa](#), INMED

15:45-16:00 **Structure-function relationship of Purkinje fibres in ventricular arrhythmias**, [Lucile Miquerol](#), IBDM

16:00-17:30 **POSTER SESSION**

PROGRAM

2nd day | 20 MAY 2025

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

Session III - Real World Data, Numeric application & IA tools

Moderator: Sarra Kraiem, OrphanDev

- 09:30-10:00 **The French Rare Diseases National Registry, from healthcare data collection to secondary reuse for research**, Alban Lermine, BNDMR
- 10:00-10:30 **Contribution of AI in Enhancing Rare Diseases Knowledge Bases: 2 Use Cases**, Bruno Sarfati, Tekkare
- 10:30-11:00 **Toward Data-Driven Medicine: Quarto as a Key Tool for Generating RWE in Rare Diseases**, Vincent Montero, OrphanDev

Round Table

Moderator: Sarra Kraiem, OrphanDev

11:15-12:00 *Regards croisés sur l'IA dans le champ des maladies rares : quelles évolutions et perspectives pour les patients*, François Antonini, AP-HM ; Xénia Proton de la Chapelle, AFSO, AtmosR ; Solange Roumengous, BNDMR [in french]

12:00-14:00 **Lunch break - Poster session**

Session IV - AI for medical imaging

Moderator:

- 14:00-14:20 **Quantitative MRI of neuromuscular disorders: The issue of images segmentation**, David Bendahan, CRMBM
- 14:20-14:40 **AI & preclinical magnetic resonance imaging applications on rodent models for cardiomyopathies and cerebral malaria studies**, Emilien Royer, CRMBM

Session V - Shorts talks selected - young researchers

Moderator:

- 15:15-15:30 1. *Single-cell morphometrics reveals T-box gene-dependent patterns of epithelial tension in the Second Heart field*, Paul Villoutreix, MMG
- 15:30-15:45 2. *Demonstration of prominent role of hiPSC-derived endothelial cells in Response to Pro-Inflammatory Cytokines in a cellular model of ICI-Induced Myocarditis*, Isaure Firoaguer, C2VN
- 15:45-16:00 3. *Impact of maternal obesity on early neural development in neonatal offspring*, Mélanie Brosset-Heckel, INMED
- 16:00-16:15 4. *Mapping the brain activity involved in the first suckling in wild-type newborn mice and in a mouse model of Prader-Willi syndrome*, Marie-Sophie Alifrangis, INMED
- 16:15-16:30 5. *Optimization and characterization of functional cardiac output in mouse gastruloids*, Srivatsava Viswanadha, MMG, IBDM
- 16:30-16:45 6. *VisuelAIclassification, a tool to share your visual skills without coding. Test on a new hematology dataset*, Théo Charnay, C2VN

17:00-17:15

Awards & Conclusions