



MarMaRa



To bring together all the players in rare diseases on the Aix-Marseille site

There are between 7,000 and 8,000 rare diseases (fewer than 5 people affected out of 10,000), over 80% of which are genetic in origin. These diseases affect around 3 million people in France. However, despite recent successes in diagnosis, care and the development of treatments, 50% of patients still have no molecular diagnosis and less than 5% of patients benefit from treatment. The aim of the Marseille institute for rare diseases (MarMaRa) is to bring together research, medical and educational teams, as well as partners from industry, in cross-disciplinary training and research initiatives.

Forming a community

13 research units

34 research teams

5 doctoral schools

2 faculties

**more than
200 staff**

**more than
200 students
(master's and
doctoral)**



Genetics

Pathophysiology

Bioinformatics

**Patient
association**

Ethics

**Mediterranean
basin**

Therapies

Heredity

Orphan drugs

Biophysics



Bringing disciplines closer together

Shared facilities

Incentive calls for projects

Interdisciplinary actions combining:

Biology

Bioinformatics

Physics

Humanities and social sciences



Training through and for research

Doctoral training

Masters and
end-of-thesis grants

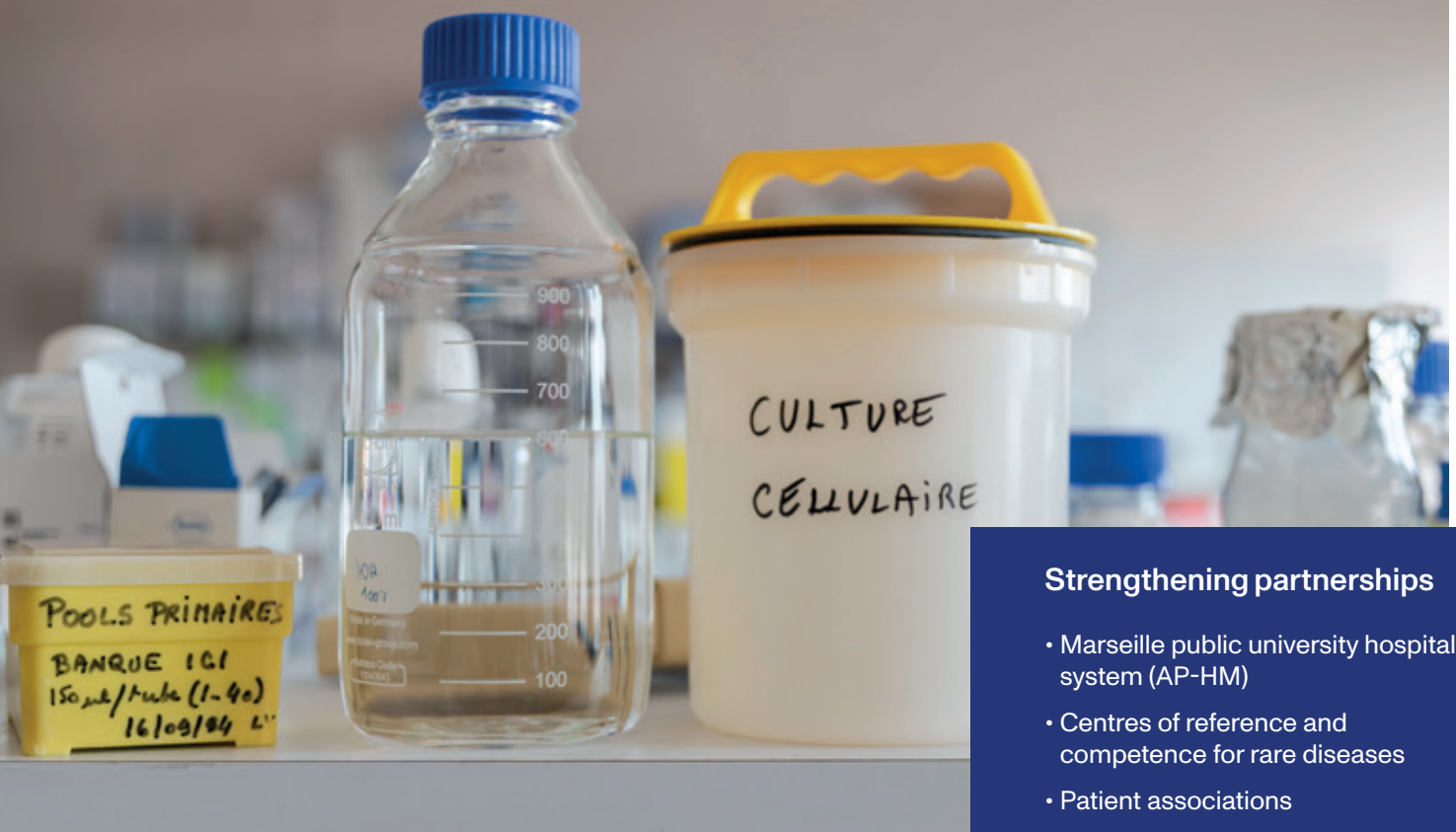
Summer school

Acting internationally

International doctoral programme

International summer school
(Blended Intensive Programme by
the CIVIS alliance)

Working group with the CIVIS
alliance, European civic university



Strengthening partnerships

- Marseille public university hospital system (AP-HM)
- Centres of reference and competence for rare diseases
- Patient associations

Videos



Watch the institute's animated film



The institute as seen by its members



Our executive office

The entire governance team: <https://url.univ-amu.fr/institut-marmara-gouvernance>



Director:
**Frédérique
Magdinier**



Co-Deputy research
director:
Françoise Muscatelli



Co-Deputy training
director:
Denis Puthier



Operations manager
Marie-Cécile Gaillard



Co-Deputy research
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Focus on the RARE TRIPS project *Training and Research Interdisciplinary Platforms*

The MarMaRa institute, awardee of the 2024 RARE TRIPS project, brings together two genomics core facilities on the Aix Marseille territory: TGML (Transcriptomics and Genomics Marseille Luminy) and GBiM (Genomics & Bioinformatics Core Facility).

The project aims to provide researchers and students with access to a genomics technology platform by fostering interactions through immersive workshops and training courses. Its objectives are also to enhance technical expertise and employability while promoting the vertical integration of teaching across different educational levels.

The initiative highlights translational research by offering interdisciplinary training and raising awareness of data management, Open Science, as well as health and safety regulations. It also encourages creativity, critical thinking, scientific rigor, and autonomy.

To learn more, participate in the annual workshop «Bring Your Own Samples: Bulk RNA-seq».



Training at MarMaRa



*Training for, through and in research,
with continuity from master's to doctorate level*



Our training ecosystem

150 master's students*

2 partner faculties

Faculty of medical and
paramedical sciences (SMPM)
Faculty of sciences

50 PhD students*

**5 partner doctoral
schools**

Life sciences (ED 658)
Biomedical research (ED 659)
Mathematics and computer
sciences (ED 184)
Physics and science of
matter (ED 352)
Engineering sciences (ED 353)

5 key master's

Biology and health
Structural biology and genomics
Bioinformatics
Integrative Biology and Physiology
Medical humanities

**1 interdisciplinary
PhD programme**

* These figures include students enrolled in master's specialisations affiliated with the institute and PhD students enrolled in our partner doctoral schools.

Explore our
full range of
training offers:





Our key actions since 2020

A PhD training programme

Funding for lab internships for master's students

Support for PhD students

Support for scientific training

A summer school and a CIVIS Blended Intensive Programme (BIP)

more than 150 participants in our PhD programme

of which 56 European students

35 scholarships for lab internships

36 end-of-thesis grants

16 scientific training grants

"I did my Master 2 bioinformatics internship at the Marseille centre for medical genetics (MMG) in the systems biomedicine team. During this internship, we were able to highlight various molecular processes that are disrupted during premature ageing, such as DNA repair, the cell cycle and communication between cells."



Cécile Beust
M2 Bioinformatics student,
awarded a Master internship grant

Our international training

CIVIS Alliance
European civic university

An international PhD programme

Close collaboration with the Mediterranean basin

Contact

Co-Deputy training directors:
Diane Frankel and Denis Puthier

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Research at MarMaRa



*Bringing together the strengths in rare diseases
of the Aix-Marseille site to consolidate
our international scientific position*



Our research ecosystem

36 teams spread across

13 research units

Health and life sciences

Bio-cultural anthropology, law, ethics and health (ADES)

Centre for study and research on health services and quality of life (CEReSS)

Centre for cardiovascular and nutrition research (C2VN)

Marseille centre for cancer research (CRCM)

Marseille institute for developmental biology (IBDM)

Mediterranean institute for neurobiology (INMED)

Marseille centre for medical genetics (MMG)

Theories and approaches of genomic complexity (TAGC)

Advanced science and technology

Marseille institute for mathematics (I2M)

Institute for research into non-equilibrium phenomena (IRPHE)

Fresnel institute

Mechanics and acoustics laboratory (LMA)

Laboratory for plasma lasers and photonic processes (LP3)

200 staff

Aix Marseille Université
INSERM

Centrale Méditerranée
CNRS
AP-HM



Our key actions since 2020

Support for mobility (call for projects mission):

- participation in scientific events
- inter-laboratory collaboration

Strengthening collaboration between the institute's laboratories (incentive action) and clinical teams of the AP-HM (translational research)

Support for scientific partner events

Funding for junior researchers (postdoctoral call for projects) and PhD students (end-of-thesis call for projects)

17 collaborative projects

6 sponsored scientific events

5 mobilities in laboratories

6 postdoctoral grants

Our main areas of research

Cutting-edge approaches to genomics

Pathophysiology

Development of therapeutic approaches

Bioinformatics

Modelling in mathematics and systems biology

Science and technology in the service of health

Consideration of ethical issues

Contact

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"With funding from the institute, we have put in place a strategy to generate cell models to assess whether genetic and telomere instabilities are present in patients with rare encephalopathies caused by UFMylation deficiency. Depending on future results, we may consider applying for national funding."



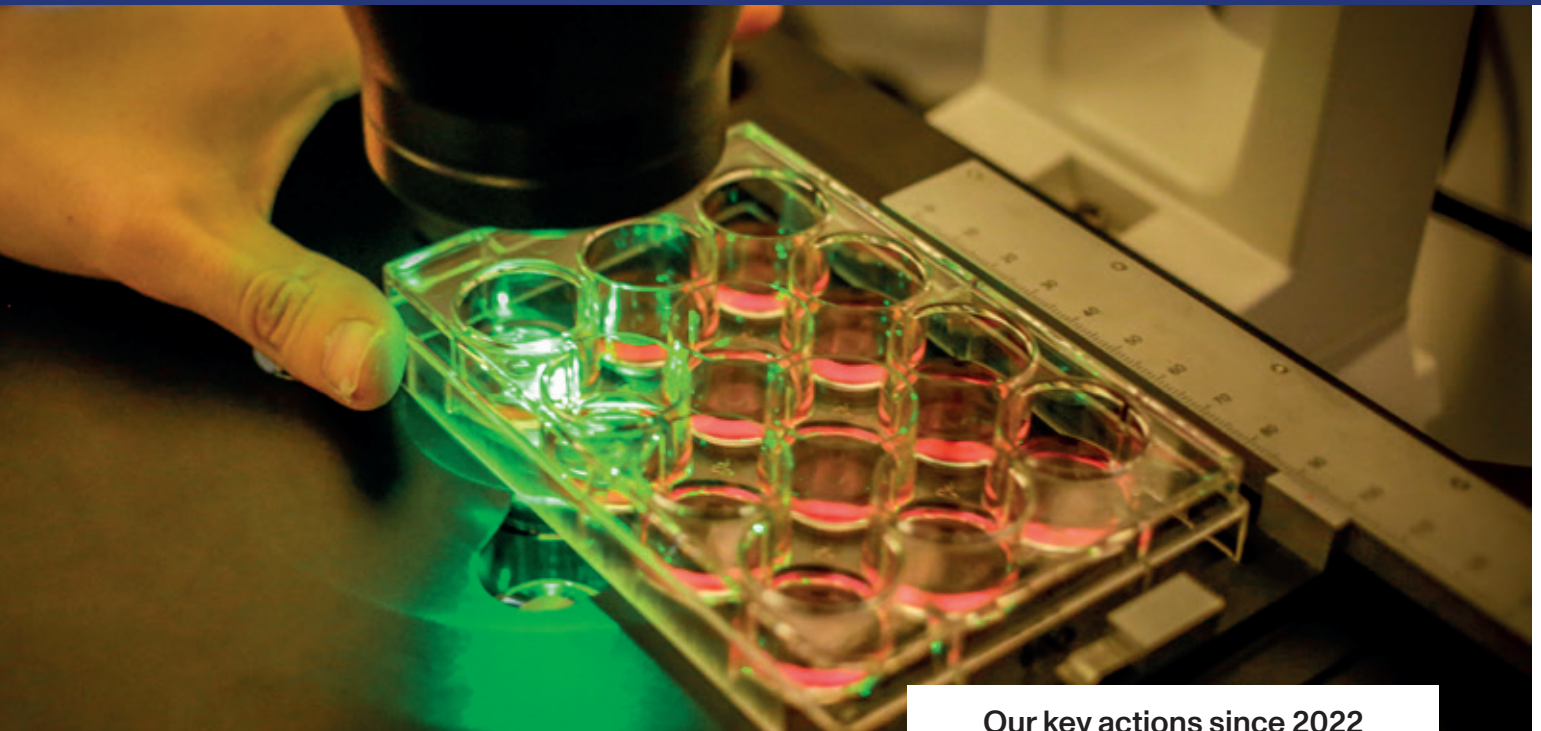
Christophe Lachaud
CRCM laboratory,
awardee of the "incentive action"
call for projects



MarMaRa partnerships



*Strengthening and developing special links with
academic and institutional players and the
socio-economic and cultural world*



Our key actions since 2022

CIVIS European working group
> 7 partner universities

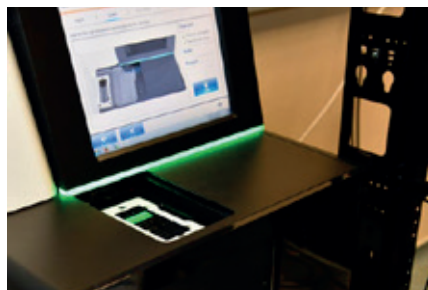
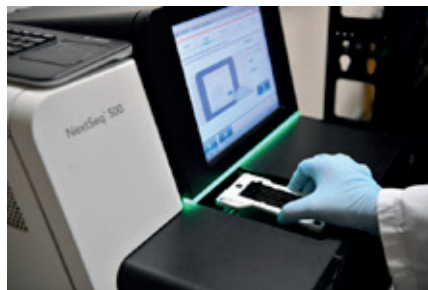
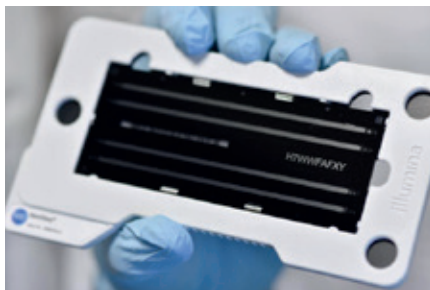
BIP CIVIS «Rare diseases at
the omics era: current tools
for frequent challenges»

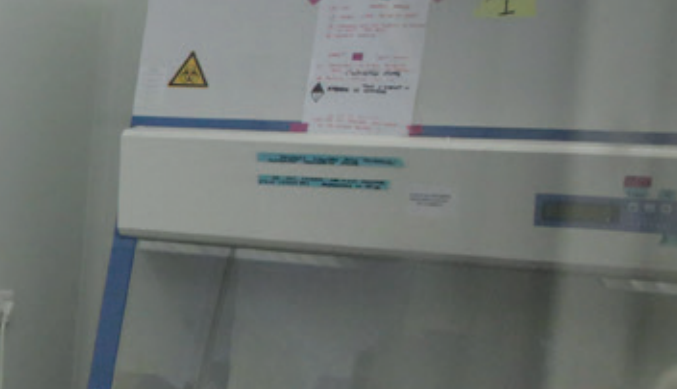
Joint working group on
participatory research (RPART) with
other interdisciplinary institutes

Genomics platforme
project (RARE TRIP)

Support for collaborative projects
with patient associations
> annual call for projects

5 technology
platforms





Our key partners include

Internationally

- European Reference Networks (ERN)
- CIVIS alliance, European civic university
- University of Sharjah (United Arab Emirates)

Partners of our teams:

- University of Sfax (Tunisia)
- Institut Pasteur de Casablanca (Morocco)
- Saint-Joseph University, Beirut (Lebanon)
- Tel Hashomer Hospital (Israel)
- Tel Aviv University (Israel)

In France

- Centres of reference and competence for rare diseases
- Alliance Maladies Rares
- Fondation Maladies Rares
- INSERM
- CNRS
- Rare diseases associations

In the Aix-Marseille area

- Marseille public university hospital system (AP-HM)
- OrphanDev
- Centrale Méditerranée
- Marseille polytech

"Our project aims to develop and study new cellular models of LAMA2-RD derived from patient stem cells (hiPSC). This funding has already enabled the team to derive hiPSCs from four patients with different mutations in the LAMA2 gene, and phenotypic analyses of the differentiated muscle cells are underway. They will help to identify the pathological mechanisms of LAMA2-RD in humans and new therapeutic targets.

Our priority for our small association, LAMA2 France, is to be able to give practical support to researchers working directly on this muscular dystrophy. Project co-funding is an effective way for us to invest, while enabling us to monitor research projects in practice and improve the information we provide to patients and their families. So this is a win-win partnership!"



Céline Damon

President of the LAMA2-France association, awardee of the "laboratory/association collaborative action" call for projects

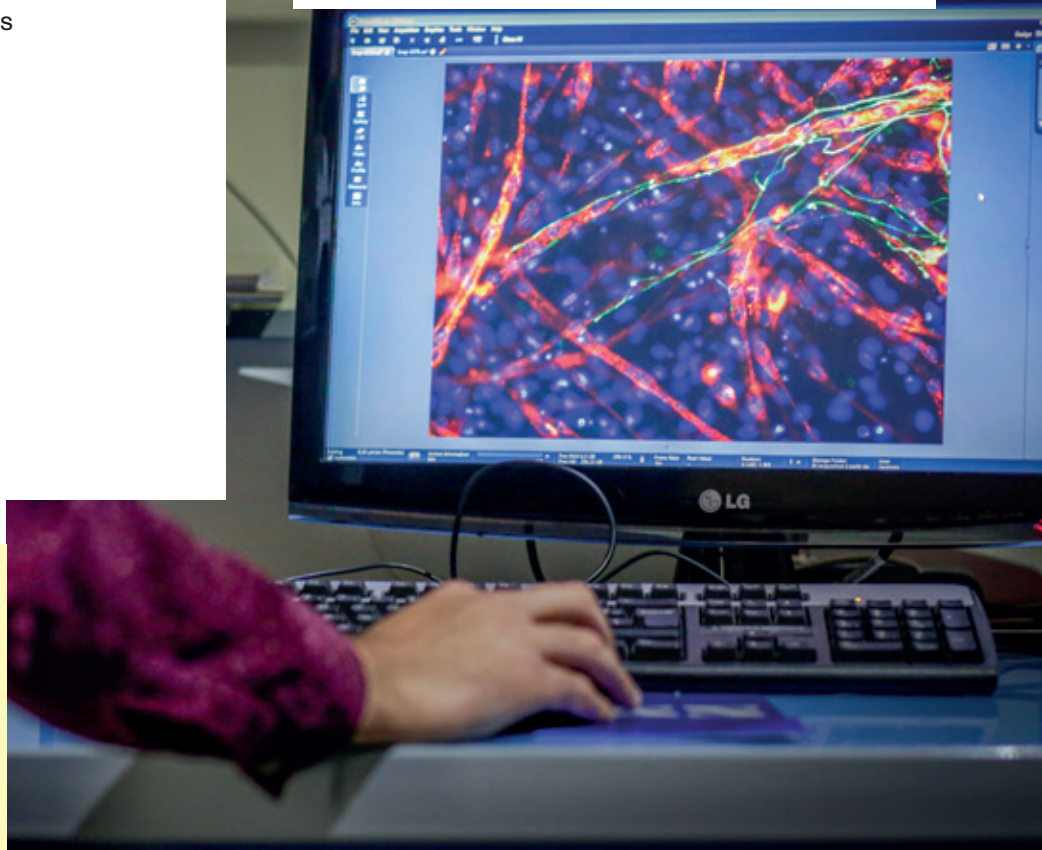
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The MarMaRa institute
brings together teams
on 7 university and
scientific sites

Nord site



Saint-Charles site



Main site: Timone



Paoli-Calmettes site



Château-Gombert site



Saint-Jérôme site



Luminy site



The Aix Marseille Université institutes are coordinated by the Amidex foundation

amidex Aix
Marseille
Université



Aix Marseille Université institutes: facilitators to strengthen the research-training link, interaction with the economic and cultural world, interdisciplinarity and internationalisation around twenty or so strategic fields of research. Their core unifying role: training a new generation of academics to meet the scientific and societal challenges of our time!

The Aix-Marseille Excellence Initiative, a consortium between Aix Marseille Université and 8 partners

amU Aix
Marseille
Université



Inserm



Centrale
Méditerranée

sciencespo.aix

Hôpitaux
Universitaires
de Marseille

ap
hm
INSTITUT PAOLI-CALMETTES
unCancer Marseille