



MarMaRa

amU Marseille rare diseases
institute
Aix Marseille Université



Forming a community

Training through and for research

Connecting disciplines

Strengthening partnerships

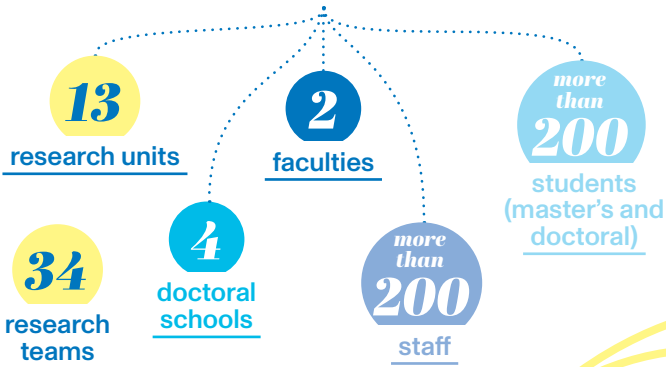
Acting internationally

MarMaRa

Our main objective: 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



- Genetics
- Pathophysiology
- Bioinformatics
- Patient association
- Ethics
- Mediterranean basin

Training through and for research

- ▶ Doctoral training
- ▶ Masters and end-of-thesis grants
- ▶ Summer school

Acting internationally

- ▶ International doctoral programme
- ▶ Working group with the CIVIS alliance, European civic university



Connecting disciplines

Strengthening partnerships

Shared facilities

Incentive calls for projects

Interdisciplinary actions combining

Biology

Humanities and social sciences

Bioinformatics

Physics

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



Watch the institute's animated film



The institute as seen by its members



Meet our executive office



Deputy research director:
Frédérique Magdinier



Director:
Thierry Brue



Project manager:
Marie-Cécile Gaillard



Deputy training director:
Denis Puthier

Discover the entire governance team:
<http://url.univ-amu.fr/institut-marmara-gouvernance>

Focus on

Our '3M' working group (Rare Diseases, Mutation, Mediterranean)

Since September 2021, the MarMaRa Institute has set up an innovative working group with the Institute for changing societies in the Mediterranean (SoMuM) to promote interdisciplinarity around genetic diseases. This working group, called "3M" (Rare Diseases, Mutation, Mediterranean), encourages dialogue between the academic world, institutional stakeholders and those from the voluntary and socio-economic sectors. This dialogue is expressed through seminars on genetics and parentage (2022), clinical trials (2023) and life experience (2024). The scale of its work (Marseille and the surrounding area) is a gateway to broader partnerships with other areas of the Euro-Mediterranean zone, with a view to sharing knowledge and experience.

To find out more, go to: <https://somum.hypotheses.org/category/penser-la-sante>



Contact

Director: **Thierry Brue**
Project manager: **Marie-Cécile Gaillard**

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Training at MarMaRa

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

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

Our ecosystem

150

master's students¹

50

PhD students²

5

key master's

1

interdisciplinary doctoral programme

2

partner faculties

4

partner doctoral schools

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

- ▶ Life and health sciences (ED62)
- ▶ Mathematics and computer sciences (ED184)
- ▶ Engineering sciences (ED353)
- ▶ Physics and science of matter (ED352)

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

Consult all master's degree specialisations:



¹ students on master's specialisations attached to the institute

² PhD students from our partner doctoral schools doing their theses while being part of our research teams

Our key actions

- ▶ A doctoral training programme
- ▶ Laboratory internships for Masters students
- ▶ Support for PhD students
- ▶ Support for scientific training
- ▶ A summer school

2020-2023

63

participants in our doctoral courses

of which **16**

European students

19

laboratory internships

23

end-of-thesis grants

8

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 2022 internship grant

Our international training initiatives

CIVIS alliance

European civic university

An international doctoral programme

Close collaboration with the Mediterranean basin



Contact



Research at MarMaRa

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

photo credit: UNIVFD



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

Research ecosystem

34

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)



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

Our key actions

- ▶ Support for mobility (*call for projects mission*)
 - participation
in scientific
events**
 - inter-
laboratory
collaboration**
- ▶ Strengthening collaboration between the institute's laboratories (incentive action)
- ▶ 3M (Rare Diseases - Mutation - Mediterranean) interdisciplinary working group with the SoMuM institute
- ▶ Funding for junior researchers (*postdoctoral call for projects*) and PhD students (*end-of-thesis call for projects*)

2020-2023

7

collaborative projects

3

GT3M seminars

3

mobility in laboratories

4

postdoctoral grants

"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, 2022

Contact

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

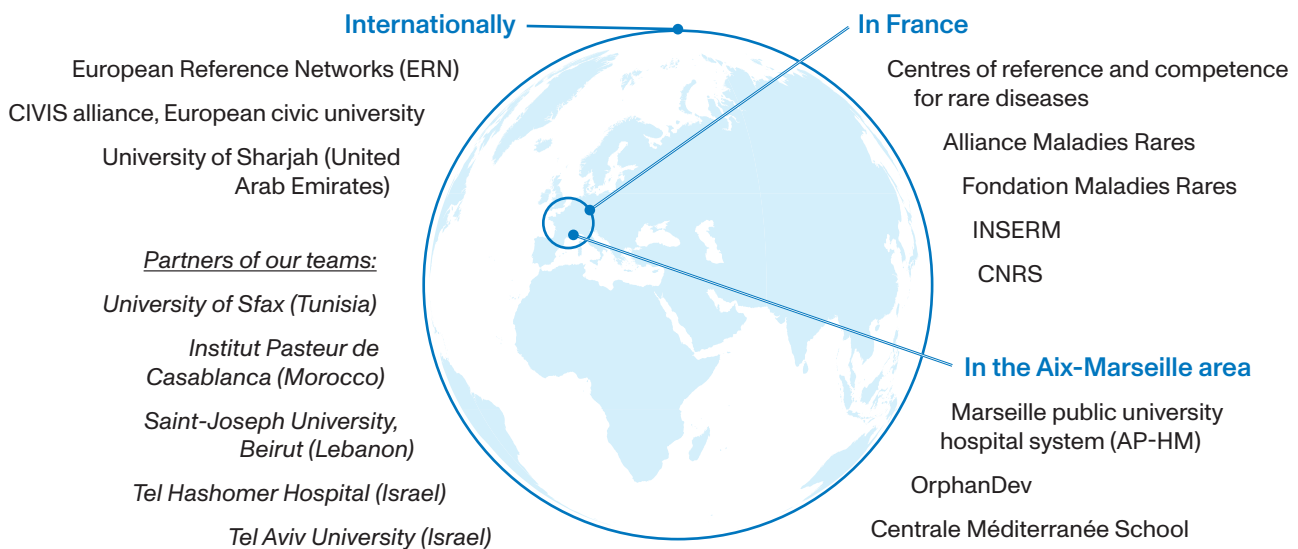
"Our project aims to develop and study new cellular models of LAMA2-RD derived from patient stem cells (hiPSC). This funding has already enabled the team to derive hiPSCs from four patients with different mutations in the LAMA2 gene, and phenotypic analyses of the differentiated muscle cells are underway. 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, 2022

Our key partners include



Our key actions

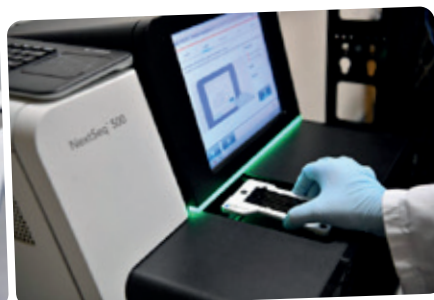
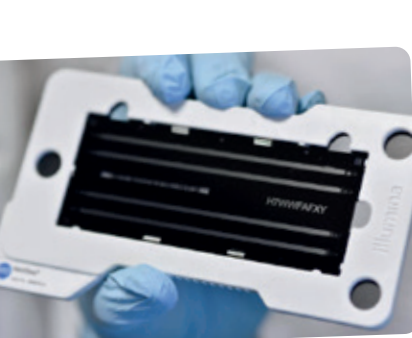
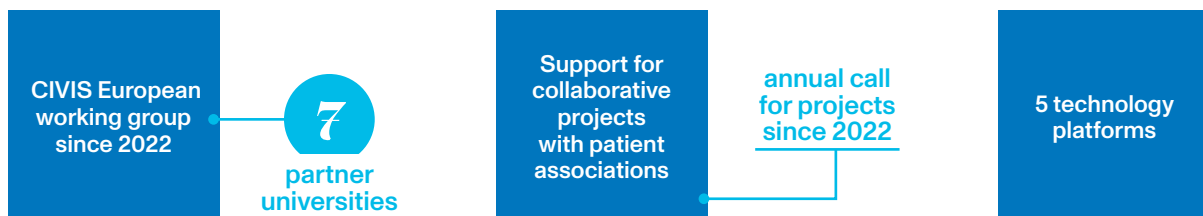


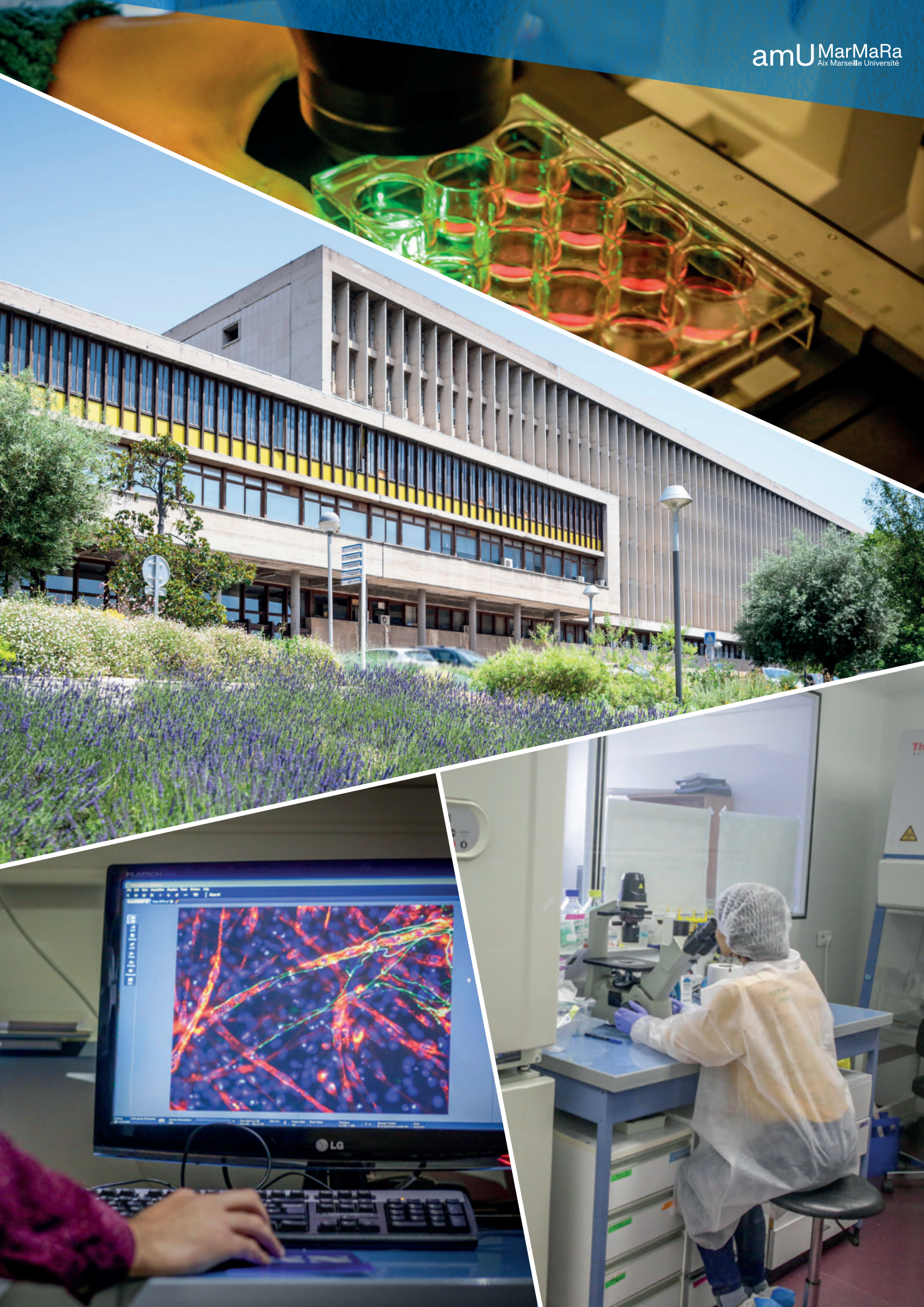
photo credit: TGML TAGC

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



The Aix Marseille Université institutes are coordinated by the Amidex foundation



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