



19

ANNUAL REPORT OF THE FOUNDATION FOR RARE DISEASES

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EDITORIAL

Advances in research, opening up to Europe and developing links with patient associations were the strong realizations of year 2019.

In 2019, the Foundation for Rare Diseases confirmed its role as a central player in the field of research and rare diseases. The results are there: in eight years, 380 projects have already been financed within the framework of 36 calls for projects, for more than 11.3 million euros allocated! The Foundation's impact is reflected in nearly 150 publications in international journals mentioning our financial support.

The Foundation for Rare Diseases is a unique structure whose mission is to accelerate research according to 4 axes: 1) identify the causes of rare diseases to speed up their diagnosis and thus put an end to the diagnostic wandering of patients and their families, 2) understand the physiopathological mechanisms of these pathologies, 3) facilitate the development of new treatments and, 4) study the specific problems affecting the quality of life of patients affected by a rare pathology and their families, in order to propose solutions, through approaches in human and social sciences, in partnership with associations and clinicians.

In 2019, we maintained our commitment to researchers by launching 4 calls for projects covering these axes. Within this framework, 37 research projects carried out in the laboratories of public research and higher education organizations were selected and funded for a total amount of 1 million euros. It is important to emphasize that this year again, 6 projects in human and social sciences, the Foundation's uniquely original program, were supported for an amount of 350,000 euros.

To promote as quickly as possible the therapeutic avenues resulting from the research, the Foundation has presented 56 therapeutic proofs of concept to drug companies since the creation of its Club POC (Proof Of Concept), a club of members from pharmaceutical industry and biotech. We are continuing to develop this local action with our regional managers who link university and hospital research laboratories with the various pharmaceutical industries and biotechnology start-ups in agreement with the Tech Transfer organizations (Satts, Inserm T, ...).

At the heart of innovation, in 2019 we also held our first co-design workshop "e-health & rare diseases" with the support of the Fondation d'entreprise IRCM. This initiative was designed to provide expertise to "young" digital health projects for the benefit of the care and life journey of people with a rare disease.

At the same time, we have provided increased support to patient associations in raising awareness of the contributions of research and in structuring their research funding actions. This merger was made possible thanks to the recruitment, in January 2019, of a manager dedicated to these functions and in interaction with the Alliance Maladies Rares.

As an extension of our mission, 2019 was a year of information and awareness of rare diseases, particularly during our scientific conference at the Collège de France on May 7, 2019 and during the Congress Rencontres RARE, on November 5 & 6, 2019 in Paris with the presence of Mrs. Agnès BUZYN, Minister of Solidarity and Health.

At the European level, the EJP "European Joint Program on Rare Diseases" consortium, of which the Foundation is a partner, started in January 2019 in collaboration with 130 organizations and more than 100 million euros in budget. We are leaders in several activities of this project coordinated by Inserm, and which will end in 2024.

All these objectives could not be achieved without the support of our private or public partners and the generosity of our donors - committed ambassadors - who help to make the Foundation a real research accelerator. We are proud and honored by their trust, which guides us and gives us the means to act.

We would particularly like to thank the AFM-Telethon, always by our side, whose very important financial support maintained each year is central for the Foundation.

We would also like to salute the conviction of our members of the Board of Directors, of the Scientific Council and of the specialist scientific committees, who provide the guarantee of excellence and independence essential to our actions.

Our ambitions for 2020

In the service of research and health, in 2020 we wish to continue our mission of general interest for research and assistance to patient associations in this field.

The start of 2020 was shaken by a health crisis linked to Covid-19. We wish to maintain our scientific policy by launching 4 to 5 calls for scientific research projects, by continuing our European work within the EJP, by collaborating with the Inter-regional Grouping of Clinical Research and Innovation of the South-West GIRCI Soho, by actively participating in the RHU COSY ("Réseau de Recherche Hospitalo-Universitaire") Cure Over-growth Syndromes) University Hospital Research project.

Our priority is to increase our investment in research, and for this ambitious program, we must increase our financial resources, strengthen our collaborations with our partners and our links with our donors.

Our results must be even more readable. Our mission, the need to support strong and innovative academic research, must be explained.

We will continue to implement promotion actions to report on and clarify our objectives: our scientific conference, the co-design workshop, web broadcasts, and the dissemination of information on all our digital channels. We are working on setting up a new website.

These ambitions mark the work and enthusiasm of the entire team. Our common wish is to perpetuate our actions by combining experience and excellence, while being responsive to the emergencies and needs of our environment, within a Foundation open to life and to the city.

**Prof. Jean-Louis
MANDEL**
President of the
Foundation



**Prof. Daniel
SCHERMAN**
Director of the
Foundation



RARE DISEASES

RARE DISEASES

In the European Union, a disease is designed to be rare when it affects less than one in 2,000 people. In France, for example, a disease is said to be rare if it affects less than 30,000 people.

Rare diseases affect 3 million people in France and nearly 30 million in Europe.

There are between 7,000 and 8,000 rare diseases, 80% of which are genetic, and new ones are defined every week.

Rare diseases are characterized by a great variability of clinical signs from one disease to another, but also from one person to another with the same disease.

Rare diseases can be hidden behind relatively common symptoms, leading to misdiagnosis.

One of the first big difficulties is the lack of diagnosis.

The so-called “diagnosis wandering” generally exceeds five years and generates absences of treatment or delays in care, accidents, and deaths.

A large number of these pathologies are also said to be “orphan” because the people concerned do not benefit from a therapeutic response.



RARE DISEASES PLATFORM

The Foundation for Rare Diseases, based in Paris, is located on the Rare Diseases Platform, a unique resource center in Europe that brings together employees and many volunteers in one place, mobilized to advance the fight against rare diseases and improve the life of sick people and their families :

- **Alliance Maladies Rares**, a French collective association gathering over 230 different rare diseases patient associations.
- **AFM-Téléthon**, at the origin of the creation of the Platform in 2001 and main funder of it.
- **EURORDIS**, European federation which brings together more than 500 patient associations.
- **Foundation for Rare Diseases**, which brings together research and care players (public, private and associative).
- **Maladies Rares Info Services**, an information and support service on rare diseases
- **Orphanet**, the reference portal for rare diseases and orphan drugs.

DR PASCALE BOMONT

INSERM RESEARCHER, WINNER OF A 2019 CONSOLIDATOR GRANT-ERC FROM THE EUROPEAN COUNCIL OF RESEARCH
TRIPLE WINNER OF THE FOUNDATION FOR RARE DISEASES :



- *Murine model project 2015 - Development of an In vivo model for Giant Axonal Neuropathy*
- *Screening project 2015 - Reversing motor deficits in Giant Axonal Neuropathy*
- *Hit-to-lead project 2019 - Hit to Lead discovery for Giant Axonal Neuropathy*

« My interest in rare diseases was born from the start of my career since my PhD thesis in human genetics on rare diseases. I am very happy to have been able to follow the growing role of rare diseases at European level, the dynamics of the French three Rare Diseases National Action Plans, and the creation of the Foundation for Rare Diseases. During my career, I participated in the study of a dozen rare neurological pathologies but I specialized very early on Giant Axon Neuro-pathy (GAN) which is a severe neurodegenerative disease beginning in young children, and which causes loss of mobility and sensitivity, then causes severe "damage" to the brain. After identifying the GAN gene, I could have continued my journey in human genetics and identified genes for other diseases, that was the line I was advised. I preferred the difficult route and I am extremely happy to have clung to understanding this rare disease, and to have dedicated twenty years of research to it.

My goal is to dissect the pathological mechanisms of rare neurodegenerative diseases. The Foundation for Rare Diseases supported me by funding three projects on GAN for my team. The last one hasn't started yet. For the first two, it is the development of a new mouse model and a therapy project on another model, the zebrafish.

Today, we have just revealed that the mutated protein in the patient, gigaxonin, plays an essential role in the recycling of cellular components, and in the birth and the integrity of neurons. This knowledge is crucial for understanding the disorders in GAN patients and developing targeted therapies, but also has an impact on other rare and common diseases, and well beyond the nervous system.

Little funding is dedicated to rare diseases. The AFM-Téléthon and the Fondation Maladies Rares play an essential role, by enabling teams like ours to initiate and lead original projects, and to accelerate discovery until the stages of therapy. My motivation comes from the patients. Knowing that I can help families to understand the disease, to find a treatment, or to alleviate a pain, is phenomenal motivation. »

01

FOUNDATION FOR RARE DISEASES

FOUNDATION FOR RARE DISEASES





THE GOUVERNANCE

The Foundation for Rare Diseases was created on February 7, 2012 by decree of the Ministry of Higher Education and Research, as a flagship measure of the "research" axis of the 2nd National Rare Diseases Plan 2011-14.

The Foundation for Rare Diseases has the french legal status of "Scientific Cooperation Foundation", which is a private non-profit legal person. It is subject to the legal rules relating to foundations recognized as being of public utility.

The Foundation is born from the joint will of 5 major players involved in the management of rare diseases and research:

+ PATIENTS ORGANIZATIONS

+ REPRESENTATIVES OF RESEARCHERS

+ REPRESENTATIVES OF PHYSICIANS



NATHALIE TRICLIN-CONSEIL

PRÉSIDENT OF ALLIANCE MALADIES RARES

« Alliance Maladies Rares is a collective of 230 patient associations, which has never ceased to promote research, which is synonymous with hope for a cure.

Together with the AFM-Telethon, the Alliance Maladies Rares has supported the creation of the Foundation for Rare Diseases in 2012, as part of the 2nd National Rare Diseases Plan. The Foundation promotes research in biomedical sciences and in human and social sciences in the field of rare diseases. As a member of the Board of Directors of the Foundation, the Alliance makes the voice of patient associations to be heard and benefits from actions to raise awareness among its members about the contributions of research and the optimization of its funding. »



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THE BOARD OF DIRECTORS

The Foundation's Board of Directors defines its strategy and actions and oversees their successful implementation. It votes the annual budget and approves the accounts for the closed financial year. It meets twice a year.

The Board is composed of 14 board members representing all the players in research and care: 5 founding members, 8 qualified personalities and 1 elected representative of professors and researchers.

THE 5 FOUNDING MEMBERS



- ✦ Mrs Laurence TIENNOT-HERMENT, AFM-Téléthon
- ✦ Mrs Nathalie TRICLIN-CONSEIL, Alliance Maladies Rares
- ✦ Mr Gilles BLOCH, Inserm
- ✦ Mr Marc PENAUD, The Conference of Managing Directors of Regional and University Hospitals
- ✦ Prof. Manuel TUNON DE LARA, The Conference of The University Presidents

8 QUALIFIED PERSONALITIES

- ✦ Prof. Jean-Louis MANDEL – President
- ✦ Prof. Michel GOOSSENS – Treasurer
- ✦ Prof. Jean-Jacques CASSIMAN
- ✦ Prof. François-Noël GILLY
- ✦ Prof. Emmanuel JACQUEMIN
- ✦ Prof. Karine LAMIRAUD
- ✦ Dr Karim OULD-KACI
- ✦ Prof. Daniel VASMANT

1 ELECTED REPRESENTATIVE OF THE TEACHERS, RESEARCHERS AND TEACHING RESEARCHERS

- ✦ Dr Guy LENAERS : Titulaire
- ✦ Prof. Stéphane BEZIEAU : Suppléant

THE SCIENTIFIC ADVISORY BOARD

The scientific advisory board is composed of renowned French and international physicians and scientists, it defines the main scientific orientations and the modalities of implementation of the annual research program. It is appointed by the Board of Directors for 4 years. It meets twice a year.

PRESIDENT : Dr Nathalie CARTIER-LACAVE, Brain and Spinal Cord Institute, Pitié Salpêtrière Hospital, Inserm

Prof. Jacques BECKMANN, Genetics and Clinical Bioinformatics, Swiss Institute of Bioinformatics, University of Lausanne (Switzerland)

Prof. Alexis BRICE, Neurogenetics, Pitié-Salpêtrière Hospital, Pierre and Marie Curie University, Paris

Prof. Patrick EDERY, Pediatric Clinical Genetics, Woman Mother Child Hospital, University of Lyon

Prof. Eric HACHULLA, Internal Medicine, CHRU de Lille, University of Lille 2

Prof. Albert HAGÈGE, Cardiology, Georges Pompidou European Hospital, Paris Descartes University

Prof. Katherine HIGH, Translational Medicine, University of Pennsylvania (USA)

Prof. Marc HUMBERT, Pulmonology, Bicêtre Hospital, Paris-Sud University

Prof. Marie-Laure KOTTLER, Department of Genetics, CHU de Caen

Prof. Pierre LÉVY, Health economics, Paris Dauphine University

Dr Jocelyn LAPORTE, Genetics, Institute of Genetics and Molecular and Cellular Biology, University of Strasbourg

Dr Sandrine MARLIN, Genetic deafness, Necker Enfants Malades Hospital, Paris

Pr Gert MATTHIJS, Genetics, University Hospital of Louvain (Belgium)

Dr Anne-Marie MASQUELIER, Consultant in strategy and development of healthcare companies, Masquelier Conseil

Dr Catherine NGUYEN, Genetics Genomics and Bioinformatics, Paris

Dr Tuan NGUYEN, Hepatology, Goliver Therapeutics, Nantes

Prof. Francesc PALAU, Genetics, Pediatric Institute for Rare Diseases, Barcelona Children's Hospital (Spain)

Prof. Yann PÉREON, Neuromuscular, Nantes University Hospital, Nantes University

Prof. A-Catherine PERROY, Pharmaceutical Law and Economics, University of Lille

Prof. Yves PIRSON, Nephrology, Saint Luc University Clinics, Louvain (Belgium)

Prof. Pierre RONCO, Nephrology, Hôpital Tenon Université P&M Curie, Paris

Prof. Damien SANLAVILLE, Genetics, Woman Mother Child Hospital, University of Lyon

Prof. Alain TAIEB, Dermatology, Saint André and Pellegrin-enfants Hospitals, University of Bordeaux

Prof. Maïté TAUBER, Pediatrics, Toulouse University Hospital, Toulouse University

Dr Laurent VILLARD, Genetics, CHU de La Timone, Aix Marseille University

Mr Michel CERBELLE, guest of the Board, representative of the Alliance Maladies Rares.

THE TEAM

The Foundation for Rare Diseases is composed of a team of professionals dedicated to supporting and funding research. Based in Paris and at the heart of inter-hospital-university regions, it works every day in close proximity with research teams and all the players in its environment to bring together expertise and promote links.

DIRECTOR

Prof. Daniel SCHERMAN

1- SCIENTIFIC POLICY MANAGER

Dr Ingrid ZWAENEPOEL

2- HEAD OF ADMINISTRATION OF RESEARCH

Mrs Diana DÉsir-PARSEILLE

3- RESEARCH MANAGER AT ASSOCIATIONS

Dr Lyne VALENTINO

4- ADMINISTRATIVE MANAGER

Mrs Nouara BENAÏ

5- HEAD OF THE COMMUNICATION DEPARTMENT AND PARTNERSHIP - Mrs Anne-Sophie BLANCHER

6- COMMUNICATION AND FUNDRAISING OFFICER

Mr Yannis HAJJI

REGIONAL MANAGERS

7- EAST - Dr Laura BENKEMOUN

8- ÎLE-DE-FRANCE - Mrs Célia MERCIER

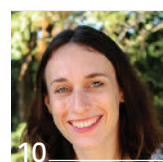
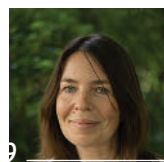
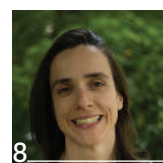
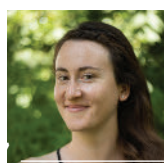
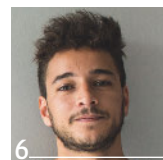
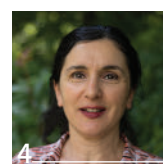
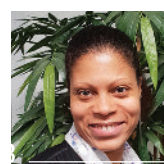
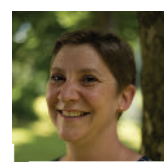
9- NORTH-WEST - Dr Anne-Sophie YRIBARREN

10- WEST - Mrs Gaëlle DOMBU SMEETS

11- RHÔNE-ALPES AUVERGNE - Dr Christine FÉTRO

12- SOUTH MEDITERRANEAN - Mrs Roseline FAVRESSE

13- SOUTH-WEST - Dr Emilie BONNAUD



THE MISSION

The Foundation for Rare Diseases has a mission of of public interest : **to accelerate research** on all rare diseases and is mobilized to:

IDENTIFY THE CAUSE OF RARE DISEASES AND HELP IN DIAGNOSIS

The Foundation for Rare Diseases supports french research teams in the stages essential to understanding rare diseases with the aim of facilitating their diagnosis. The most promising research is selected through calls for projects on the basis of criteria of scientific excellence.

ITS RESULTS SINCE 2012

- + 221 granted research projects
- + 100 news genes identified
- + 3 000 exomes
- + 800 transcriptomes
- + 400 genomes

4M€

CONTRIBUTE TO THE DEVELOPMENT OF NEW TREATMENTS

The Foundation facilitates access to the latest technological advances and associated skills, in partnership with cutting-edge technological platforms, in order to develop new therapeutic approaches.

Thanks to its team, the Foundation detects promising projects and supports research teams in the various stages of therapeutic development.

ITS RESULTS SINCE 2012

- + 113 granted research projects
- + 84 models of rare diseases
- + 15 high throughput screening
- + 14 translational studies

3,9M€

IMPROVING PATIENTS' LIFE PATH

For the first time in the field of rare diseases in France, in 2012, the Foundation launched a call for projects to support joint research between doctors, researchers in the human and social sciences and patient associations. The aim is to assess the repercussions of rare diseases for those affected and their families and to suggest adaptations of practices.

ITS RESULTS SINCE 2012

- + 45 granted collaborative projects gathering
- + 153 research teams
- + 59 patient associations involved

3,4M€



BILAN SINCE 2012

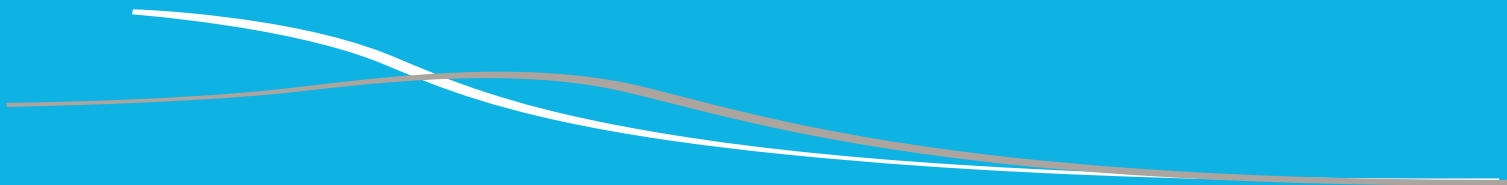
- + 36 calls for projects
- + 379 granted research projects
- + 1 159 submitted applications
- + Reasonable success rate of 33%
- + 150 publications

11,3M€

02

FOUNDATION FOR RARE DISEASES

THE FOUNDATION'S ACTIONS





The Foundation for Rare Diseases is the only national structure to drive research into all rare diseases.

It finances, coordinates and supports the best research projects in terms of diagnosis, treatment and improvement of daily life in order to give hope to patients and their families. To respond to these missions, the Foundation has implemented 3 types of actions:

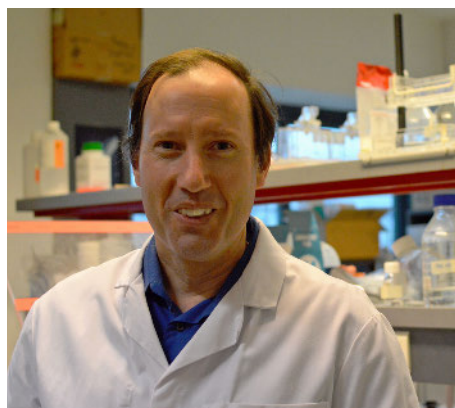
- ✦ FINANCE SCIENTIFIC RESEARCH PROJECTS
- ✦ SUPPORT THE DIFFERENT ACTORS IN THE FIELD
- ✦ INFORM AND TRAIN

DR FABRICE LEJEUNE

2013 WINNER OF THE FOUNDATION FOR RARE DISEASES CALL*

Inserm researcher in the CANcerHeterogeneity, Plasticity and Resistance to THERapies laboratory at the Lille Biology Institute.

The research team led by Dr Lejeune has shown that one of the active ingredients contained in the fungus *Lepista inversa* has restorative properties which make it possible to correct certain genetic mutations, known as "nonsense" mutations. The results are published in Nature Communications.



« I'm working on a type of mutation called a 'nonsense' mutation which is a very serious form of mutation. It leads to the premature termination of the synthesis of proteins - essential for the function of the gene - which malfunction and can no longer fulfill their role in the body. This genetic defect leads to the onset of clinical symptoms of the disease.

The advantage of this mutation is that the gene is still present in the patient's cells but it is no longer expressed. We have now obtained a very effective molecule which allows this gene to be re-expressed and allows us to achieve a level of correction of the mutation which is compatible with a therapeutic benefit. This discovery is hopeful, it is a big step forward for research, reinforced by the very low or even absence of toxicity of the molecule. This data is essential when we want to develop a treatment. Now we still have steps to take before arriving at a real therapeutic strategy and being able to offer a drug. »

* This project, laureate of the Foundation's 2013 screening call for projects, is now supported as part of the "Proof of concept" support".

02

FOUNDATION FOR RARE DISEASES

THE FOUNDATION'S ACTIONS

FINANCING SCIENTIFIC PROJECTS

1- CALLS FOR PROJECTS

2- CALLS FOR PROJECTS IN PARTNERSHIP

3- SUPPORTED RESEARCH PROJECTS

5- OUT OF CALLS FOR PROJECTS

FINANCING SCIENTIFIC PROJECTS

The Foundation for Rare Diseases pursues an active scientific policy to bring out great discoveries by funding academic research projects and innovations in the fields of biology, chemistry and the human and social sciences.

1- 2019 CALLS FOR PROJECTS



The Foundation launches each year 4 to 6 calls for projects to fund the winning teams and facilitate researchers' access to cutting-edge technologies.

PROCESS FOR SELECTING FUNDED PROJECTS

The most promising research works are selected on the basis of criteria of scientific excellence.

Depending on the themes, the selection of projects is carried out in one step (evaluation on a complete application) or in two steps (preselection on a letter of intent then evaluation on a complete application), according to the schedule of calls for projects established with the Scientific Board.

- 1- Each application is evaluated by at least two independent experts.
- 2- On the basis of the external expert evaluations, specialized committees made up of members of the Foundation's Scientific Council and experts of each research field meet to assess and select the application submitted to the Foundation's calls for projects.

‘HIGH-THROUGHPUT SEQUENCING’

MISSION : TO HELP IN DIAGNOSIS

Decipher the molecular bases of rare diseases in order to allow a diagnosis for each disease.

SINCE 2016 :

Broadening of the contours of the call for projects (exome, genome, epigenome, transcriptome).

- ✦ 6 calls for projects.
- ✦ 80 projects supported out of 195 file proposals.
- ✦ Average funding per project of 25,000 euros.

RESULTS IN 2019

1 calls for projects

- ✦ 14 projects funded out of 32 applications.
- ✦ 250,000 euros allocated.
- ✦ Partnership : Imagine Institute, IGBMC, CNRGH, Integrigen and Eurofins genomics

Extended to other platforms for specific projects based on innovative technologies (IBENS (Paris), UCA-Genomix (Nice)).

List of the 14 granted projects in 2019 p66

‘DEVELOPMENT OF EXPERIMENTAL MODELS ’

MISSION : HELP TO DEVELOP NEW TREATMENTS

To elucidate the pathophysiological mechanisms and explore evidence of therapeutic principle.

SINCE 2013 :

- ✦ 5 calls for projects.
- ✦ 79 projects supported out of 162 file proposals.
- ✦ Average funding of 4,000 to 60,000 euros depending on the project.

RESULTS IN 2019

1 call for projects

In 2019, a unique call for projects for the development of animal models and iPS-type cellular models.

- ✦ 15 projects granted out of 35 applications.
- ✦ 330,000 euros allocated.

List of 15 projects funded in 2019 p68



‘HIGH THROUGHPUT SCREENING ’

MISSION : HELP TO DEVELOP NEW TREATMENTS

Identify molecules with therapeutic potential based on the development and validation of a miniaturized high throughput screening test.

SINCE 2013 :

- ✦ 4 calls for projects.
- ✦ 15 projects supported out of 70 applications.
- ✦ Average funding per project of 25,000 euros.

RESULTS IN 2019

In 2019, in order to capitalize on the results of high-throughput screening projects, the Foundation provided additional support to projects aimed at confirming and optimizing “hits” resulting from the Foundation's laureate screening projects.

- ✦ 2 'hit to lead' projects supported on 3 projects presented.
- ✦ 50,000 euros allocated.
- ✦ Partnerships with 6 platforms : PCBIS, Strasbourg - CMBA, Grenoble - ARIADNE, Lille - Criblage, CEA Saclay - Criblage, Institut de la Vision, Biophenics, Institut Curie.

List of 2 granted projects in 2019 p67

"HUMANITIES AND SOCIAL SCIENCES"

MISSION : IMPROVING PATIENTS' LIFE PATH

Analyze the individual and social consequences of rare diseases, with a view to improving the life course of the sick person and those around them.

SINCE 2013 :

- ✦ 7 calls for projects.
- ✦ 45 projects supported.
- ✦ 73 associations involved.
- ✦ Average funding per project of 100,000 euros (until 2019).

The call for projects in human and social sciences led by the Fondation benefits from the support of the IRCEM corporate foundation* (until 2018) and the Caisse Nationale de Solidarité pour l'Autonomie (CNSA).

RESULTS IN 2019

1 call for projects

Maximum budget 80,000 euros.

- ✦ 6 projects funded in 2019 out of 31 files presented.
- ✦ 20 research teams and 6 associations supported.
- ✦ 350,000 euros allocated.

List of 6 projects funded in 2019 p70



* Translation from "Fondation d'entreprise IRCEM"

2- CALLS FOR PROJECTS IN PARTNERSHIP



Regional partnership



**European
programme**



150,000 euros
allocated

WITH THE GIRCI SOHO (Inter-regional Group for Clinical Research and Innovation in the South-West and Overseas Hospital)

MISSION :

Enable health professionals from the GIRCI inter-region (Nouvelle-Aquitaine, Occitanie and Overseas) to better understand and assess rare diseases and cancers through feasibility studies / pilot clinical studies. It corresponds to the fourth edition of the Inter-regional Thematic Call for Projects "APITHEM" by GIRCI SOHO, the thema of which varies from one edition to another.

RESULTS IN 2019

- ✦ 9 laureates projects out of 40 applications submitted: 8 concern the study of rare diseases and 1 the study of rare cancer.
- ✦ Attributed to hospital practitioners from the GIRCI SOHO inter-region.
- ✦ 50,000 euros from the Foundation out of the 450,000 euros allocated in total.
- ✦ Consolidation of collaborative networks between expert centers and teams involved within the establishments.

In the longer term, these studies may allow the emergence of larger clinical research projects.



CALL FOR EXPRESSIONS OF INTEREST WITH SATT OCCITANIE

In January 2019, the two Technology Transfer Acceleration Companies in the Occitanie region (SATT TTT in Toulouse and SATT AxLR in Montpellier) joined forces with the Fondation Maladies Rares to promote the emergence and maturation of innovative projects in the field of rare diseases.

MISSION :

Detect emerging projects with high potential for innovation and technology transfer concerning diagnostic or prognostic approaches, medical devices, preventive or therapeutic approaches for all rare diseases, including rare cancers..

RESULTS IN 2019

- ✦ 6 laureate projects out of 13 applications.
- ✦ Subjects covered: development of new gene therapies, innovative strategies in the treatment of rare and severe neurological disorders, development of new active molecules to treat T-cell non-Hodgkin lymphoma and small-cell lung cancer, or development an original prognostic tool at the service of antenatal medicine.

The six selected projects are now in a support phase from the SATTs where experts assess their development potential and the possibility of benefiting from maturation funding from their respective SATTs.

EUROPEAN JOINT PROGRAMME « RARE DISEASES » EJP-RD

The Foundation for Rare Diseases is a partner of EJP-RD, a European program for the coordination of means and resources for research in rare diseases (European Joint Program Cofund on Rare Diseases). The project coordinated by Inserm, began in January 2019 for a period of 5 years and brings together 130 organizations from 35 different countries.



To learn more about the EJP RD : <https://www.ejprarediseases.org>



In 2019, the Foundation was involved in 3 activities : :

THE CO-FUNDING BY THE FOUNDATION OF FOUR SELECTED RESEARCH PROJECTS UNDER THE 2019 EJP RD TRANSNATIONAL CALL FOR PROJECTS

The Foundation participated in the co-financing of four research projects selected as part of the EJP RD transnational call for projects. This call for projects focused on the theme **"Research projects to accelerate diagnosis and / or explore the progression and mechanisms of rare diseases"**. The Foundation will also co-fund the 2020 transnational call for projects to the sum of 100,000 euros.

RESULTS IN 2019

- ✦ 220 pre-proposals submitted / 217 eligible proposals.
- ✦ 52 preselected projects / 22 supported projects.
- ✦ 23 french teams funded.
- ✦ 100,000 euros from the Foundation.

List of 4 projects funded (5 teams) in 2019 p71

Find out more : <https://www.ejprarediseases.org/index.php/open-call-jtc2020/>

THE DEVELOPMENT OF AN ONLINE COURSE ON RARE DISEASE RESEARCH (WP16 - "E-LEARNING ACADEMIC COURSE ON RD RESEARCH")

The Foundation for Rare Diseases coordinates this working group which brings together 17 partner organizations. The objective is to offer the **1st multidisciplinary and transversal course available online** on research in rare diseases. In 2019, significant comparison and census work was carried out and two major surveys conducted to assess the needs of the European "rare diseases" community in terms of online training.

- ✦ Targets: students (medicine / research) and / or health professionals.
- ✦ Language: English
- ✦ Three priority themes: diagnostic research, therapies personalized innovations and translational research.

To find out more about this consultation and investigation work : <https://www.ejprarediseases.org/index.php/training-and-empowerment/e-learning/>

THE DEFINITION OF A CALL FOR INNOVATIVE PROJECTS ON RESEARCH CHALLENGES FOR RARE DISEASES (WP8 - "RARE DISEASES RESEARCH CHALLENGE")

The innovative call "Research challenges for rare diseases" aims to facilitate and finance collaborations between industrial partners, academics, SMEs and patient organizations in order to solve research challenges specific to rare diseases.

- ✦ 2 round tables with manufacturers under the coordination of Eurordis.
- ✦ 4 challenges have been identified which will be the subject of the call for projects:
 - Development of a non-invasive tool to measure the mobility of patients with rare diseases on a daily basis (industrial sponsors: *Chiesi and CSL Behring*).
 - Characterization of mobility challenges related to rare bone disorders in a real life environment (industrial sponsor: *Ipsen*).
 - Development of an intranasal delivery system for biological drugs to newborns (industrial sponsor: *Chiesi*).
 - Development of a preclinical test to detect the instability of repeated expansions of microsatellites (industrial sponsors: *Cydan and Pfizer*).

A networking event was organized by the Foundation in early March 2020 in Paris between potential candidates (universities, SMEs, patient associations) and industrial partners involved in defining the challenges.

- ✦ Call for projects launched in the second quarter of 2020.

To know more : <https://www.ejprarediseases.org/index.php/fundings-and-calls/rare-diseases-challenges/>

3- SUPPORTED RESEARCH PROJECTS OUT OF CALLS FOR PROJECTS

The Foundation for Rare Diseases joins forces with partners: patient associations, private companies, medical insurance and social protection groups, foundations, in order to finance and scientifically support projects of excellence in rare diseases outside the field of its calls for projects, in particular concerning devices and e-health.

COM-ATAXIA DIGITAL TOOLS FOR COMMUNICATION OF PATIENTS WITH FRIEDREICH ATAXIA

FINANCED BY THE IRCEM corporate foundation

Friedreich's Ataxia (FA) is a rare handicapping poly-neurodegenerative disease of genetic origin. Its prevalence is of the order of 1 for 50,000 people in Europe. This represents around 1,500 sick people in France.

The disease is caused by an abnormality in the X25 gene, located on chromosome 9. The X25 gene normally codes for a protein called frataxin. The precise role of this protein is not yet precisely known, but it seems essential for the proper functioning of cells.

FA results in the progressive development of motor disorders (slow and imprecise gestures, hyper fatigability, falls, loss of walking, etc.) and sensory disorders (reduction in visual acuity, hearing loss, etc.). There is no treatment that can prevent, slow down or even stabilize the progression of the disease.

The COM-ATAXIE project originated from a strong demand from patients and their entourage and from the desire to help ataxic people to communicate for as long as possible.

Its aim is to identify compensation tools and technical aids useful for maintaining the communication skills of people with Friedreich's ataxia.

It offers an innovative research approach at the intersection of themes e-health and rare diseases.

OVERGROWTH SYNDROMES: IMPROVEMENT OF THE HEALTH STATUS OF PATIENTS CARRIED OUT BY PROFESSOR GUILLAUME CANAUD

IRCEM corporate foundation GRANT

Overgrowth diseases, in which part of a child's body grows in an anarchic and uncontrolled manner, represent a sometimes insurmountable obstacle to a child's development and his passage to adulthood and independence, when they do not affect his life expectancy.

Prof. Guillaume Canaud's team (Institut Necker Enfants Malades, INSERM U1151) has made an exceptional therapeutic breakthrough through the use of a drug that blocks an intracellular signaling pathway called PIK3CA.

The grant from the IRCEM corporate foundation will help to carry out "Omics"-type experiments to better decipher the pathophysiology of the overgrowth syndrome linked to PIK3CA (PROS patients) and to identify new therapeutic targets in the event if appearance of drug resistance occurs. This should allow the long-term treatment of young patients, who must be treated throughout their growth, and ensure their passage into adulthood. **This research project also aims to extend and generalize the concepts and treatments obtained so far to other overgrowth diseases.**

A MEDICO-ECONOMIC STUDY carried out by the Cemka Eval firm

FINANCED BY THE IRCEM corporate foundation

This study aims to assess the economic and social impact of certain rare pathologies. It is deployed around two components :

- **quantitative component:** an analysis of data from the National Health Data System for 5 rare pathologies: SMA (spinal muscular atrophy), Rett, Marfan, Sickle cell disease and Willebrand.
- **qualitative aspect:** a survey of patients and / or their families to describe the consumption of resources not covered by health insurance. Three rare pathologies will be included: SMA, Rett Syndrome, Marfan Syndrome.

A report on the quantitative part is being finalized.

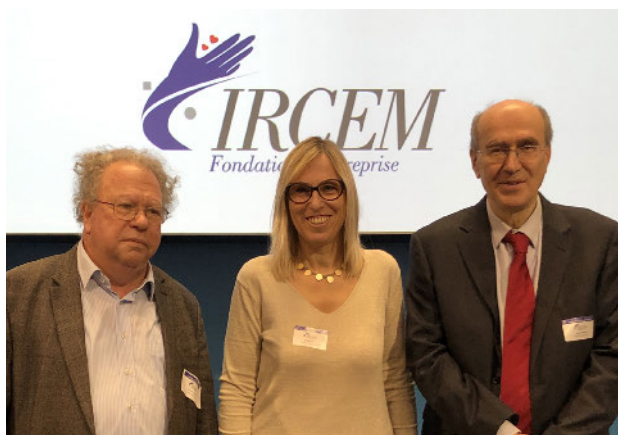
The recruitment of patients and families by clinicians and associations has started in 2020.



DR NATHALIE COULON

PRESIDENT OF THE IRCEM CORPORATE FOUNDATION

« The IRCEM Group is the medical insurance and social protection group employees in family jobs at home. In 2013, The IRCEM Group chose to invest funds in the creation of a corporate foundation whose purpose is to support actions which allow, at all ages of life, to live better within the family (prevention, fight against dependency, home care) and improve the life course of patients and their caregivers. The IRCEM corporate foundation primarily targets support for children with rare diseases, on the one hand, and patients with Parkinson's disease and their family caregivers, on the other.



Very early, a partnership with the Foundation for Rare Diseases was set up, in particular through its call for projects in human and social sciences, which made it possible to finance support projects for patients and their families. The involved teams of researchers and associations were able to present their studies and results at two specific conferences in 2016 and 2019.

Today, the collaboration still revolves around the funding of research projects in social and human sciences, but has also been enriched with new axes: projects, in particular carried by associations, promoting the development of new care and support solutions. for the benefit of children, adolescents and their families, but also the joint exploration of new themes such as e-health in rare diseases. »

02

FOUNDATION FOR RARE DISEASES

THE FOUNDATION'S ACTIONS

SUPPORTING THE DIFFERENT ACTORS IN THE FIELD

1- RESEARCH TEAMS

2- ASSOCIATIONS

SUPPORTING THE DIFFERENT ACTORS IN THE FIELD

1- RESEARCH TEAMS

The Foundation for Rare Diseases supports research teams in the development of their project at all stages without distinction with regard to their nature or the targeted pathology.

Its field of action covers the field of translational research and brings together the expertise necessary for the continuity between basic research and clinical research. This promotion aid increases the value of research and development results, thus promoting the translation of discoveries into industrial applications.

This support by the Foundation is free-of-charge and does not imply any sharing of intellectual property. It covers the different following aspects:

INTELLECTUAL PROTECTION

In order to reach the market and benefit the patient, any innovation must be the subject of an often long and expensive development. Consequently, it is essential to protect the invention so as to have the exclusive rights.

The Foundation for Rare Diseases helps to contact the Intellectual Property structures, to establish an invention declaration, to patent an innovation and to transfer the innovation.

MEDICAL AND REGULATORY ASSISTANCE

The aim of the research is to find an active drug candidate in a given pathology. The development of this drug candidate into a drug then goes through stages framed by strict and complex regulations..

The Foundation helps to assess the relevance of submitting a file according to the stage of development of the project, to find service providers specializing in regulatory affairs and to establish a relationship with regulatory agencies.

FINANCE RESEARCH

Funding for calls for projects carried by the Foundation is complementary to other sources of funding for pilot or seed programs, whose completion and validation may prove to be fundamental for the further deployment of larger size development steps.

The Foundation assist scientists in identifying the most relevant among its calls for projects or also in identifying other funding sources, depending on the nature of the project (AFM, FRM, ANR, E-Rare, PHRC, PRTS, etc.). She also helps in drafting the application proposals.

CONNECTING SCIENTISTS TO

- academic and private partners, depending on the identified needs: patent / intellectual property experts, CRO (contract research organization), formulation experts, etc.
- drug manufacturers or investment funds to promote research results, in particular through the Club POC (Proof of concept / evidence of therapeutic principle).

This activity is carried out by 7 regional managers who are in the field in close proximity to the research teams. Depending on your location, you can contact this expert responsible for your region by email.

All information : <https://fondation-maladiesrares.org/>

THE PROOF-OF-CONCEPT CLUB



In 2017, the Foundation created its Club for the promotion of research: the Club POC (Proof Of Concept) which connects academic research teams with industrial partners.

This Club is a mechanism for promoting research for the development of new treatments, outside of our calls for projects.

It brings together Pharma and Biotech Industry, as well as venture capital wishing to strengthen their positioning on rare diseases. In this context, the Foundation initially selects sufficiently mature academic projects. It then presents them, with the agreement of the researchers and their Technology Transfer organization, to industrial members of the Club. In case of an expression of interest, our team then organizes meetings between all the players under the cover of confidentiality agreements.

Members

- Drug companies.
- Big, middle-sized, biotechs.
- Venture capital organizations

Sessions

Two sessions per year since its creation in 2017.

Results

- ✦ 148 projects supported by the Foundation out of 254 identified.
- ✦ 56 projects presented to Club POC.
- ✦ 32 links made, i.e. 60% interest from manufacturers.
- ✦ 1 partnership agreement signed in April 2018 between the company Lysogene. Dr Hervé MOINE and SATT Conectus

2- ASSOCIATIONS

The Foundation for Rare Diseases advises and supports patient associations in the structuring and implementation of their actions in favor of research.concerned associations

36

supported associations

ISABELLE COSTE, AUTOUR DU BPAN

« Less than two years ago, my husband and I founded the association “Autour du BPAN” (around BPAN - beta-propeller Protein Associated Neurodegeneration). We immediately wanted to stimulate and support medical research dedicated to the rare disease called BPAN.

The world of research was foreign to us. It was therefore natural that I turned to the Foundation for Rare Diseases.

I had the pleasure of learning that a service dedicated to supporting associations had just been created. I was received very quickly by the manager, Mrs. Lyne VALENTINO.

Over the months, thanks to her benevolence and her explanations, the workings of the research became clearer. Seven months later, following her advice, Autour du BPAN launched its first call for projects. Two research programs have been selected, one of which is co-funded by the Foundation for Rare Diseases. A real relationship and trust has been established.

The Foundation in the person of Lyne traveled to the Loire during the conference we were organizing. The Foundation therefore has a real close relationship with associations. This is really very appreciable. Today we continue to work together and it is a real pleasure. Thank you so much to the professionalism of the Foundation for Rare Diseases! »



1- ASSIST THE ASSOCIATIONS WITH THE IDENTIFICATION OF TEAMS AND RESEARCH PROJECTS

The Foundation, through its knowledge of the various players in the field, helps in the search for scientific partners for the development of associative projects.

Supported associations

- ✦ French Association against Peripheral Neuropathies (AFNP)
- ✦ Algodystrophy France (algodystrophy, chronic regional pain syndrome)
- ✦ HPN France (paroxysmal nocturnal hemoglobinuria, idiopathic aplastic anemia) *Joint call for projects Association / Foundation*
- ✦ CHAMP-1 France (intellectual disability linked to an anomaly of the CHAMP-1 gene)
- ✦ APEHDia - Association Parents of children with diaphragmatic hernia (of the dome) *Joint call for projects Association / Foundation*
- ✦ Fructos'Amis pour la vie, French Association of fructose intolerants (congenital fructosemia)
- ✦ Association des sclerodermiques de France (scleroderma)
- ✦ ASPASC - Association of People with Currarino Syndrome

2- SCIENTIFIC MONITORING AND ASSISTANCE FOR THE VALUATION OF RESEARCH PROJECTS SUPPORTED BY PATIENT ASSOCIATIONS

The Foundation facilitates relations with researchers and helps monitor the progress of achievements. It analyzes and popularizes the results of research, it provides support for the promotion of projects.

Supported associations

- ✦ AFAF - French Association of Friedreich's Ataxia - With the support of the IRCM corporate foundation
- ✦ Xtraordinary (Mental handicap linked to the X chromosome)
- ✦ OLY - Open Eyes (Hereditary Optic Neuropathies)
- ✦ Association MED13L syndrome (intellectual disability linked to an anomaly MED13L or MED13 genes)
- ✦ Autour du BPAN (neurodegeneration linked to an anomaly of the WDR45 gene)
- ✦ Ewen Life - For a life like any other with a rare disease (all rare diseases)
- ✦ ALBI - Association for the fight against inflammatory diseases of the liver and bile ducts

XTRAORDINAIRE

« Xtraordinaire is an association bringing together families affected by neurodevelopmental disorders linked to the X chromosome. It represents around ten very rare diseases and is organized into committees by disease in order to support families and research programs.

Website : <http://xtraordinaire.org/index.php>

For several years, the Creatine Transporter Deficit Commission (SLC6A8) has been following and financially supporting the work of Dr Aloïse MABONDZO, head of the Neurovascular Pharmacology Team at CEA in Saclay.

In 2019, while therapeutic trials are being planned on the drug candidate developed by the CEA, the merger with the Foundation for Rare Diseases proved to be very rich. In mutual respect and trust, the project was presented to them to assess its strengths, difficulties, and legal, technical and human needs.

The benevolence, availability, understanding and responsiveness of our contacts at the Foundation are essential to advise us and allow the CEA to succeed in its research. »



3- INFORM ASSOCIATIONS ABOUT RESEARCH INTERVENTIONS

The Foundation for Rare Diseases communicates and disseminates information on the scientific approach methodology and the organization of research. It raises awareness of the challenges of research in rare diseases and the complexity of drug development.

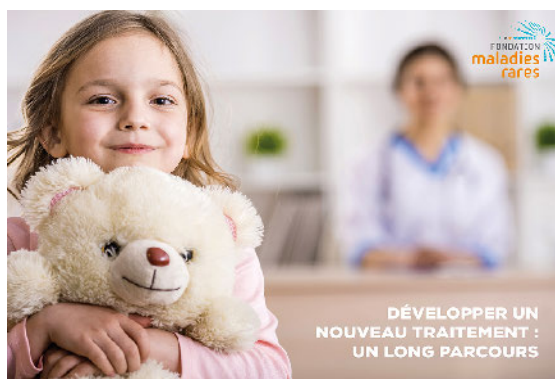
This translates into 3 types of actions:

INTERVENTIONS

- Annual day of the DEFISCIENCES sector (March 2019)
- Board of Directors of the association “Connaître les Syndromes Cérébelleux” (Knowing about Cerebellar Syndromes (Paris on 09/14/19)
- Research day of the association Around BPAN (Roanne on 09/28/19)
- Family day of the Francophone association of Glycogenoses (Rouen on 10/26/19)
- BRAIN-TEAM association day (Paris on 11/26/2019)

PUBLICATIONS

Conception of a popular science article on the process of new drug development and therapeutic advances in Angelman syndrome.



PERSONALIZED ADVICE TO ASSOCIATIONS

Analysis of the association's research activities.

Advice for the implementation of projects (bibliographic review, linking, etc.).

Supported associations

- ✦ French Association of Ectodermal Dysplasies
- ✦ Atour du BPAN (neurodegeneration linked to an anomaly of the WDR45 gene)
- ✦ AMS-ARAMISE (Multiple system atrophy)
- ✦ ACO2 Gene Association (Optic neuropathy linked to the mutation of the Aco2 gene)
- ✦ CRPS algodystrophy
- ✦ ASAP for Children (Farber's disease, SMA-PME)
- ✦ ATMM - Association Tanguy Moya Moya (Moya-moya Syndrome)
- ✦ BBS - Bardet-Biedl Association (Bardet-Biedl Syndrome)
- ✦ BPAN France (Neurodegeneration linked to the WDR45 gene)
- ✦ Know the cerebellar syndromes (spinocerebellar ataxias and associated syndromes)
- ✦ DEBRA France (Epidermolysis bullosa)
- ✦ Everest d'Ernest (Duplication of chromosome 15)
- ✦ FFAMH - French Federation of Associations of Hematochromatosis Patients (Hemochromatosis)
- ✦ Fragile X France (Fragile X Syndrome and associated diseases)
- ✦ LHF Espoir (Lymphohistiocytosis)
- ✦ Vaincre la PRR (Recurrent Respiratory Papillomatosis).

THE COLLABORATION WITH THE ALLIANCE MALADIES RARES

- ✦ Participation in the "Research and Treatments" and "Diagnosis" working groups of the Alliance Maladies Rares
- ✦ Participation to events
 - Intervention during the "Drug development" information day (July 2019)
 - IWeb meeting animation (webinars):
 - "Launch an AAP" (April 2019)
 - "Leading a scientific council" (July 2019)
- ✦ Animation of workshops at Fall Universities (October 2019)
 - "Research on rare diseases at European level"
 - "Young researchers"
 - "Explaining research to patients"

Alliance
maladies rares
UNIS POUR VAINCRE



Created on February 24, 2000, Alliance Maladies Rares (association law 1901, recognized of public utility) brings together some 230 patient associations. She is the voice of 3 million patients affected by around 7,000 rare diseases. The Alliance covers the national territory with its 12 regional delegations.

Website : <https://www.alliance-maladies-rares.org>

02

FONDATION MALADIES RARES

THE FOUNDATION'S ACTIONS

INFORM, TRAIN

- 1- THE ANNUAL SCIENTIFIC COLLOQUIUM
- 2- REGIONAL CONFERENCES
- 3- THE FIRST WEB-SHOW
- 4- THE FIRST CO-DESIGN WORKSHOP
- 5- THE CONGRESS RENCONTRES "RARE"
- 6- THE MEETING "CAFÉS-DÉBATS"

INFORM, TRAIN

The events organized by the Foundation aim to disseminate good practices, share knowledge and provide the best possible information on all rare diseases.

1- THE ANNUAL SCIENTIFIC COLLOQUIUM



On May 7, 2019, the Foundation organized a scientific day at the “Collège de France” with the objectives of presenting the results of the laureate projects, exchanging on best practices in the conduct of these projects and on sharing experiences, to encourage collaboration between actors and to encourage the emergence of new research projects.

For the opening of this day, we had the pleasure of welcoming Mr. Philippe Berta, Member of Parliament for Gard, Geneticist, President of the National Assembly study group on rare diseases

- ✦ 28 speakers.
- ✦ 350 participants from academia, clinic, industry and associations.
- ✦ 40 posters presented.
- ✦ Two sponsoring partners : Sanofi, Institut Roche.

Discover the posters, the videos of the sessions and the presentations on the site : <https://evenements-fondation-maladiesrares.org/event/colloquescientifique/>




SANOFI GENZYME


InstitutRoche

2- REGIONAL CONFERENCES

Rare Diseases DAY, MONTPELLIER

JUNE 11, 2019

- ✦ Co-organized with the Institute of Functional Genomics..
- ✦ 130 participants.
- ✦ A partner : Alnylam.

Find out more :

<https://evenements-fondation-maladiesrares.org/event/journee-recherche-maladies-rares-montpellier/>



Rare Diseases DAY, BORDEAUX

MARCH 8, 2019

- ✦ Co-organized with the Bordeaux University Hospital.
- ✦ 100 participants.
- ✦ A partner : Biogen.

Find out more :

<https://evenements-fondation-maladiesrares.org/event/journee-sur-les-maladies-rares-de-la-recherche-aux-traitements/>

3- THE FIRST WEB-SHOW

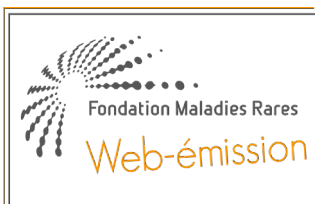
The Foundation offers a new biannual meeting around key topics in the field of rare diseases with the support of the Groupama Foundation 3“Vaincre les maladies rare” a 20-30 minute discussion between experts to follow live.

Speakers :

- ✦ Prof. Agnès BLOCH-ZUPAN, professor (PU-PH) in oral biology at the dental surgery faculty of the University of Strasbourg and hospital practitioner, member of the CRM R O-Rares.
- ✦ Mr Hervé SERVY, computer engineer.
- ✦ Dr Vincent VARLET, emergency doctor, MBA and graduate of INSEAD

To view on our Youtube channel : <https://youtu.be/6vIK9-hYA3A>

It will be followed by 3 new editions in 2020.



4- THE CO-DESIGN WORKSHOP "E-HEALTH & RARE DISEASES"

The Foundation for Rare Diseases has launched a new action to support innovation in e-health with the setting up of e-health and rare diseases co-design workshops.

The workshop is aiming to **support new innovative digital health projects** for the benefit of the care and life path of people with a rare disease.

The first edition took place on June 27, 2019 at **Paris Biotech santé**, an incubator for start-up and SMEs in the field of biology, which made its premises available free of charge.

This day brought together many experts around new digital projects for rare diseases. It ended with the interventions of Ms. Pauline D'ORGEVAL, santebd.org and SecondAvis.com and Mr. David SAINATI, MedAppcare.

- ✦ A financial partner: the IRCEM corporate foundation.
- ✦ 5 projects supported during this 2019 edition.
- ✦ Online, the video of the day : <https://youtu.be/X6kwPvWmo3M>



2019 EDITION PROJECTS

- AI-DENT (Prof. Agnès BLOCH-ZUPAN, CRMR O-Rares, Strasbourg)
- AnDD-e-rare (Prof. David GENEVIÈVE, CRMR Developmental anomalies and malformation syndromes, AnDDI-Rares sector, Montpellier)
- QUALVI (Mrs. Sophie BERNICHTEIN, BRAIN-TEAM health sector, Paris)
- TELE-SLA (Dr Véronique DANIEL, SLA Center, Lille)
- TUTOTEC'H (Mrs Géraldine MERRET, AFM-Téléthon, Evry and Bordeaux)

5- The Congress RENCONTRES "RARE"



The Foundation was honored to carry the 6th edition of the Rencontres RARE, on November 5 and 6, 2019 at the Cité des Sciences et de l'Industrie, in Paris.

These two days brought together the entire rare disease community: public decision-makers, patient representatives, health and research professionals, pharmaceutical, medical device and health technology manufacturers.

The objective of these Congress was to assess the state of research and care in this sector and to define new ambitions to come in order to provide solutions for the management of these serious, chronic and disabling diseases.

- ✦ 600 participants.
- ✦ 60 speakers in the plenary room.
- ✦ 15 speakers on the Agora space.
- ✦ Presence of the Minister of Solidarity and Health, Mrs. Agnès BUZYN.
- ✦ 95 posters on display.
- ✦ 50 industrial partners.



Discover the event: program, partners, videos, interventions on the website : <https://www.rareparis.com/2019>

6- THE MEETING "CAFÉ-DÉBATS"

The LEEM Rare Diseases Committee (LEEM : Les Entreprises du Médicament – Union of the French Pharmaceuticals Companies) and the Foundation organize the "Cafés-débats" to raise awareness among politicians, institutions and authorities of the issues specific to rare diseases



- ✦ January 2019 : "Bioethics, rare diseases and diagnosis".
- ✦ May 2019 : "The role of Europe in responding to the challenges of rare diseases? Implications for the 3 million French patients?".

03

FOUNDATION FOR RARE DISEASES

THE FOUNDATION'S PARTNERS





THE FOUNDATION'S TEAM WOULD LIKE TO THANK ITS PARTNERS,
SPONSORS AND DONORS WHO TRUST US SINCE 2012,
BY SUPPORTING RESEARCH ON RARE DISEASES.

Since its creation, the Foundation for Rare Diseases has endeavored to develop its social missions by creating close and personalized partnerships with professional circles and civil society. It benefited from the financial support of its founding members and its patrons, numerous and faithful donors, families and friends of people affected by rare diseases.

This year again, the AFM-Telethon was a decisive support for the Foundation with a grant of 1,500,000 euros.

AFM TÉLÉTHON



The AFM-Telethon (French Myopathy Association – Telethon) is an organization of patients and relatives of patients, whose objective is to overcome neuromuscular diseases, which are progressive and severely disabling rare diseases. It plays an important role as a patient support association, as well as a patient advocacy organization.

To do this, AFM-Telethon is pursuing a strategy of general interest that benefits all rare diseases. The great popular mobilization of the Telethon that AFM-Telethon organizes every first weekend of December has contributed to the recognition of rare diseases as a public health issue and has enabled it to become a major player in biomedical research in France and in the world.

AFM-Téléthon supports around 200 research teams per year and clinical trials concerning genetic diseases of vision, blood, brain, immune system, liver, muscle, etc.

Through its laboratories (Généthon, Istem, Institut de Myologie), it is also an atypical patient association capable of designing, producing and testing its own innovative therapy drugs. In addition, AFM-Téléthon innovates in the social field, with the implementation of a model of local support for neuromuscular patients and families, from access to diagnosis and care to the implementation of patient life project.

As a Founding Member of the Foundation for Rare Diseases, AFM-Téléthon has provided decisive support for the Foundation's actions since its creation in 2012.

CHARITY EVENTS

GALA "LES ÉTOILES RARES"

A new edition of the charity event "Les Étoiles Rares", initiated by the MBA Wine Marketing & Management students from the INSEEC business school in Bordeaux, took place in May 2019.

The organizing team wanted to renew this evening with a gala dinner associated with a wine auction in support of the Foundation and for the benefit of research on rare diseases.

This event, sponsored by the French rugby international Baptiste SERIN, took place at the Château Pape Clément, which had the generosity to open its doors to us to welcome "Les Étoiles Rares".

The gastronomic chef Thibaut SERVAS was, as in the first edition, the initiator of the success of this evening, offering an "All Cacao" menu designed by four hands with the Bordeaux chocolate maker Hasnaa FERREIRA.

This dinner, associated with the auction of prestigious wines and spirits, has been a great success and raised 20,000 euros.

We warmly thank all the actors involved in the success of this event.



SERVICE ORGANIZATION

MOBILIZATION OF THE KIWANIS DISTRICT FRANCE-MONACO CLUB

The Kiwanis District France-Monaco has been involved for more than five years alongside the Rare Disease Foundation to fight against rare childhood diseases. They raise awareness and publicize the cause of rare diseases throughout the country with the organization of theatrical performances, film screenings, chocolate fairs, auctions, golf tournaments, etc.



Thanks to this support and to its rare disease ambassador, Mr. Patrick MAUREL, 147,595 euros have been allocated to research projects since 2015.

In 2019, nearly 30 clubs took action for a total of 43,248 euros.

Three projects benefited from this support in 2019:

- Dr STUDER (Nice) - Identification of the genetic pathways leading to hereditary congenital facial paralysis and associated hearing loss;
- Dr CHASSAING (Toulouse) - Analysis of regulatory sequences of genes involved in anophthalmia-microphthalmia;
- Dr LESCA (Lyon) - Search for genes involved in West syndrome.

EXAMPLE OF A CHARITABLE EVENT "PIÈCES ROUGES" BY THE KIWANIS OF VAR WEST REGION

Between February and August 2019, the second edition of the "Pièces rouges" took place, an initiative that raised 4,200 euros.

Kiwanis piggy banks were placed at traders and supermarkets in a territory stretching from Bandol to Toulon.

We warmly thank all the participants as well as all the volunteers for this wonderful operation, from the collection of boxes to the counting and until the deposit at the bank.



UTA JOULIAN

Treasurer of Kiwanis Club of La Seyne-Six-Fours-Saint-Mandrier, delegate for rare diseases and initiator of the operation "Pièces rouges".

« This action was initiated two years ago, and it allows Kiwanis to support the Rare Disease Foundation in the fight against rare pediatric diseases, by researching new diagnoses and new treatments. (...) »

As we (the Kiwanis clubs) are very attached to the actions of the Rare Disease Foundation, we have embarked on the unprecedented collection of red coins. It is not always easy to raise money that is intended for help outside the department for research projects. (...) The warm welcome among traders and the generosity of donors are a real driving force for me to continue. This action is a good way to communicate about the Foundation as well as the vocation of Kiwanis clubs. »

THE INTERNATIONAL ORDER OF ANYSETIERS



In the Middle Ages, numerous Corporations gathered people exercising the same profession. They formed very powerful Brotherhoods. The Corporation des Anysetiers was made up of doctors and apothecaries who provided patients with their knowledge of the properties of anise, to which therapeutic virtues were attributed.

Ancestor of pharmacists, the Corporation des Anysetiers is now reviving in the form of the International Order of Anysetiers, a "Service Organization". Through a large number of events organized for the benefit of social needs, the Order is dedicated to many causes, in particular those concerning childhood.

The Foundation was supported in 2019 by numerous Anysetiers Commanderies, in particular in the southwest region. Prof. Daniel SCHERMAN gave several lectures on rare diseases, which were an opportunity to educate the audience on research in this field and to have the honor to receive the funds collected by the various Commanderies. In 2019, the Commanderies of Landes, Béarn-Bigorre, Guyenne Occitane and Haut Languedoc stood out in their support for the Foundation for Rare Diseases. Several actions are planned for 2020, the implementation of which will depend on the evolution of the health situation.

SPORTING SOLIDARITY CHALLENGES

ULTRA TRAIL DU MONT-BLANC®

The UTMB® is a world famous and anticipated sporting event that takes place at the end of August in Chamonix. It is made up of several ultra-endurance mountain races around Mont-Blanc.

In 2019, the Foundation for Rare Diseases was fortunate enough to once again be one of the solidarity causes selected by the UTMB® organization, thus allowing runners to give a human and solidarity dimension to their sporting challenge by running with the Foundation's colors.

The number of runners who can join one of the causes is limited and defined by the organization. **In August 2019, 29 runners chose to wear a Foundation for Rare Diseases "solidarity bib". We are very proud of this recognition and mobilization.**

Through their participation, these volunteers are great ambassadors ! Each runner commits to collecting a minimum of 2.000 euros from sponsors - both companies and private individuals - to finance their race-bib and for the benefit of the chosen cause. In 2019, 58,000 euros were paid directly to the Foundation to finance and accelerate research in rare diseases.

We thank them as well as the partner companies for their commitment!

In August 2020, 25 runners tackle the challenge by wearing a solidarity race-bib of the Foundation. These solidarity bibs are postponed to 2021, 2022 or 2023 following the health crisis.



The website : fondation-maladiesrares.org/utmb

The Facebook page : [defi.utm](https://www.facebook.com/defi.utm)

OUR MAJOR UTMB'S PARTNERS EN 2019



THE SOLIDARITY RACE "J'OFFRE UNE COURSE!"

The 4th edition of the solidarity event “J’offre une course” took place at the Joffre High School in Montpellier in March 2019. The organizers, Mr Laurent LAVAL and Mr Olivier SIERRA were once again able to mobilize their students and the teaching staff of establishment for the benefit of the Foundation and thus support research.

This year, more than 800 participants gathered to take part in this magnificent day of solidarity: at the start 130 teams of 5 runners (4 students and 1 adult) for this relay race. For each km being run, the runners raised funds from their relatives or partner companies for the benefit of the Foundation.

For this edition, two Foundation's employees took part by joining a team of runners : Mrs. Roseline FAVRESSE, South-Mediterranean regional manager and Dr Émilie BONNAUD, South-West regional manager.

The Foundation for Rare Diseases is honored by this renewed commitment and pleased with the visibility given to rare diseases.



SUPPORT THE FOUNDATION

GIVE WITH FULL TRUST

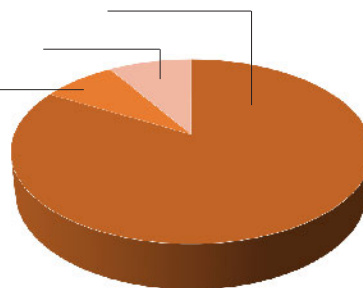
The Foundation for Rare Diseases ensures the transparency of the use of the support of its sponsors and patrons through internal control procedures and the certification of its accounts by an auditor.

To fulfill its mission of general interest, the Foundation needs the financial support of individuals, companies and other foundations. Thanks to these donations, the Foundation will be able to go even faster and further in scientific discoveries and in supporting sick people and their families.

By choosing to support the Foundation, its partners offer patients and their families not only hope for a better life but above all solutions to improve their daily lives.

HOW ARE THE COLLECTED RESOURCES USED?

83,6% Support and funding of research *
8,6% Operating and communication costs
7,8% Fundraising costs



To make a donation, you have different possibilities:

- send a check payable to the Foundation for Rare Diseases,
- make an online donation using a secure form,
- or make a bank transfer (account number of the Foundation online).

We are at your disposal to think together about solutions adapted to your expectations.

Contact : Anne-Sophie Blancher
Head of Communication and Partnership Department
anne-sophie.blancher@fondation-maladiesrares.com
01.58.14.22.87

Foundation for Rare Diseases / Fondation Maladies Rares
Plateforme maladies rares - 96, rue Didot, 75014 Paris

WHAT ARE THE APPLICABLE TAX DEDUCTIONS?

The Foundation for Rare Diseases is authorized to receive donations from companies and private individuals under the fiscal regulations governing patronage. It can also be the beneficiary of bequests, donations and life insurance exempt from inheritance and transfer taxes.

FOR PRIVATE INDIVIDUALS

INCOME TAX

66% tax reduction of the amount of the donation, within the limit of 20% of income tax.*

For every 100€ donated, your real cost is 34€ after tax rebates

SOLIDARITY TAX ON WEALTH

75% tax reduction of the amount of the donation, within the limit of 50.000 of wealth tax.

For every 1,000€ donated, your real cost is 250€ after tax rebates

FOR COMPANIES

CORPORATION TAX

60% tax reduction of the amount of the donation, within the limit of 0.5% of turnover* and 10,000 euros or 5 per thousand of turnover.

For every 10,000€ donated, your real cost is 4,000€ after tax rebates

* Includes in-kind donations entirely earmarked for research.

** For IS and IR, the excess can be carried forward over 5 years.

04

FOUNDATION FOR RARE DISEASES

THE FINANCIAL REPORT





A WORD FROM THE TREASURER

In 2019, despite an unfavorable economic climate and the loss of certain institutional funding, the Foundation for Rare Diseases succeeded in maintaining its research funding activity in a sustained manner, in accordance with its commitments under its general interest mission for support research on rare diseases. This was made possible thanks to the renewed support of the AFM-Telethon (founding member), and of many other contributions collected thanks to the dynamism of our team and the generosity of the support and individual donors, medicare insurance organizations, solidarity sports and pharmaceutical industries.

In 2019, the Foundation has devoted 1.18 million euros to funding research projects, in line with previous years, thus supporting 37 projects selected via committees of independent scientific experts through 4 national "calls for projects".

In addition, we are financing 4 projects selected through a European call for projects, and 4 additional promising projects validated by external experts, but not specifically falling within the thematic framework of our national calls for projects.

Our strong and original commitment in the field of human and social sciences aimed at improving the living conditions of patients and their loved ones has continued thanks to the faithful support of the Caisse Nationale de Solidarité pour l'Autonomie (CNSA).

To this direct investment allocated to the best French research teams, we must also add the investment in the field by the 7 regional managers of the Foundation who support researchers involved in rare diseases.

The POC "Proof of Concept" Club aimed at accelerating the development of new drugs by connecting academic researchers with the pharmaceutical and biotechnological industries continued its activity, and the financial support of manufacturers for this field work provided by our regional managers generated more than 110,000 euros for the Foundation.

As a pledge of the Foundation's future and development, for the first year, we were able to benefit from European funds amounting to 270,000 euros as part of our participation in the "European Joint Program on Rare Diseases".

At the same time, the restructuring of our communication tools and our fundraising approaches have allowed us to increase our income from sports sponsorship. We have continued to reduce our overheads and, remarkably, we managed to reduce our personnel costs by 90,000 euros compared to 2018, while maintaining our level of activity.

These management efforts, without compromising our funding to research on the 7,000 rare diseases, have made it possible to maintain the accounting deficit of our 2019 budget at 154,000 euros, therefore at a level comparable to that of the previous year and less than 50% than in 2017.

In 2020, our efforts will be even more strongly aimed at increasing our resources in order to equilibrate this balance sheet, while maintaining very substantial support for research. We have already obtained significant national funding for the next 5 years, as part of our participation in the "Cure Overgrowth Syndrome" COSY RHU (University Hospital Research Network).

We extend our warm and friendly thanks to the members of our Board of Directors and of our Scientific Council, not to mention the many partners and patrons who continue to trust us.

Prof. Michel GOOSSENS
Treasurer of the Foundation for Rare Diseases



STATUTORY AUDITOR'S REPORT

As part of the mission to present the annual accounts of Foudation for rare diseases for the financial year 01/01/2019 to 12/31/2019, and in accordance with the terms of our engagement letter, we performed the due diligence required by the presentation standard defined by the Order of Chartered Accountants.

Pursuant to the mission entrusted to us by the Board of Directors, we have audited the annual accounts of the Foundation for Rare Diseases relating to the financial year ended 12/31/2019, as attached. to this report. These accounts were closed by the treasurer on June 1, 2020 on the basis of the information available on that date in an evolving context of the health crisis linked to Covid-19.

We certify that the annual accounts are, with regard to french accounting rules and principles, regular and sincere and give a true picture of the results of operations for the past financial year as well as of the financial situation and of the Foundation's assets at the end of the year.

Extract from the 2019 Financial Report. Paris, June 9, 2020.

ACCOUNTING RULES AND METHODS

The annual accounts for the year on 12/31/2019 have been drawn up and presented in accordance with the provisions of CRC regulation 99-01 relating to the procedures for drawing up the annual accounts of associations. The accounting conventions have been applied with sincerity while respecting the principle of prudence, in accordance with the basic assumptions:

- sustainability concern,
- consistency of accounting methods from one financial year to the next,
- independence of financial years.

and in accordance with the general rules for drawing up and presenting annual accounts.

The basic method used for the valuation of items entered in the accounts is historical cost method.

Only significant information is expressed. Unless otherwise specified, the amounts are expressed in euros.

TANGIBLE AND INTANGIBLE ASSETS

Tangible and intangible fixed assets are valued at their acquisition cost for assets acquired for valuable consideration, at their production cost for assets produced by the company, at their market value for assets acquired free of charge and by way of payment exchange.

The cost of an asset is its purchase price, including customs duties and non-recoverable taxes, after deduction of rebates, trade rebates and cash discounts of all directly attributable costs incurred to bring the asset into use in place and in working order for the intended use. Transfer rights, fees or commissions and legal costs related to the acquisition are attached to this acquisition cost. All costs which are not part of the acquisition price of the asset and which cannot be directly linked to the costs made necessary to put the asset in place and in a state of operation in accordance with its intended use, are accounted as charges

DEPRECIATION

Amortization for depreciation is calculated on a straight-line basis according to the expected life:

- Concessions, software and patents: 5 years
- Office equipment: 5 to 10 years
- Computer equipment: 3 years
- Furniture: 10 years

The depreciation period used for simplicity is the period of use for goods that cannot be originally broken down.

The Foundation has assessed at the closing date, taking into account the internal and external information at its disposal, the existence of signs showing that the assets may have significantly lost value.

RECEIVABLES

Receivables are valued at their nominal value. Depreciation is applied when the inventory value is lower than the book value.

EXCEPTIONAL PRODUCTS AND CHARGES

Exceptional income and expenses take into account items unrelated to the normal activity of the Foundation.

DONATIONS IN KIND

During the year, the Foundation benefited from in-kind donations that could be assessed at 104.1 k€:

- Grant Thornton: 9.6 k€; skills sponsorship
- Other service providers (Rare Stars and conferences): 6.7 k€; skills sponsorship
- Other providers (Rare Stars): 4.4 k€; sponsorship in kind
- Experts for the Foundation: 66.6 k€; contribution in kind for the expertise of scientific projects and participation in selection committees.
- CNRGH (Centre National de Recherche en Génomique Humaine): 16.8 k€; for a call for high throughput sequencing projects.

SOCIAL MISSIONS

The social mission of the Rare Disease Foundation as defined in article 1 of its statutes is to promote and accelerate research on rare diseases.

The implementation of its mission is divided into 6 areas of intervention:

- Linking of research and care actors;
- Facilitating access to resources essential for research: expertise, technologies, financing;
- Facilitation of the collection of clinical and biological data;
- Help in setting up the early phases of clinical trials;
- Stimulation of research in human and social sciences;
- Contribution to national and international policy on rare diseases.

The actions of the Foundation focus on the achievement of 3 major objectives:

- Understanding rare diseases;
- Help develop new treatments;
- Improve the life course of people with rare diseases.

ATTRIBUTION PRINCIPLES - USES OF FUNDS FOR SOCIAL MISSIONS

RESOURCES COLLECTED FROM THE PUBLIC

Donations from individuals are entirely allocated to social missions. In the event that the donor wishes his donation to be geared towards a group of specific diseases, he is informed by mail that his request has been taken into account and will also be informed of the dedicated action carried out.

PERSONNEL COSTS

The head of science policy and the head of research administration are in charge of implementing science policy and managing the Foundation's calls for projects. Their activities are entirely dedicated to carrying out the Foundation's social missions.

The regional managers of the Foundation are deployed in the French inter-university hospital regions and their main missions are :

- Implement the Foundation's strategy and carry out all the actions necessary to facilitate research activities on rare diseases in the inter-region;
- Strengthen synergy between players in the field of rare diseases (rare disease reference centers, research teams, patient associations, private players, patrons, etc.) and facilitate the implementation of research projects on rare diseases.

80% of their activity is dedicated to carrying out the Foundation's social missions.

Management invests 20% of its working time in the deployment and monitoring of programs.

55% of the communications manager's activity is dedicated to social missions.

The communications and fundraising manager carries out 40% of his activity in social missions.

MISSION AND DUTY TRAVEL COSTS

Travel and mission expenses of the people involved in the missions are taken into account according to the allocation percentage previously described.

The mandates of the experts of the Scientific Council and of the various selection bodies for calls for projects are completely free.

Only the costs related to their trips are reimbursed on the basis of actual costs and are assigned to social missions.

OTHER EXPENSES

The other expenses are allocated, according to their use, to the activities of social missions.

COSTS DISTRIBUTION KEYS

The first key concerns the distribution of personnel expenses which include gross salaries, social and fiscal charges and assignable travel costs. These expenses are allocated according to the percentages described above.

The second key concerns travel and non-assignable mission costs which are distributed, according to the weight of each position in relation to the payroll, under the following headings:

- Realization of social missions
- Fundraising actions
- The rest is allocated to operating costs

SUBSEQUENT EVENT

As of January 30, 2020, the WHO declared a state of international public health emergency. Due to the evolution of COVID-19, France announced containment measures dated March 17, 2020. COVID 19 has no impact on the fiscal year ended December 31, 2019.

Due to these containment measures, the Foundation's activity is disrupted in 2020. Given the evolution of the situation on the closing date of the accounts, we are not able to accurately apprehend all the elements that could impact the accounts for the 2020 financial year.

RETIREMENT COMMITMENT

No commitment has been recorded in terms of pensions, supplementary pensions and similar indemnities.

The retirement commitment was valued at December 31, 2017 for an amount of 46.4 K euros.

The assumptions used were as follows:

- Retirement age: 65
- Turnover rate: 1%
- Annual salary evolution: 1%
- Nature of retirement: voluntary departure
- Social security charges rate: 45%

BALANCE SHEET

Assets (in €)	31/12/2018	31/12/2019
<i>Gross investments</i>	74 101	76 015
<i>Depreciation</i>	(43 278)	(49 665)
Net investments	30 823	26 351
Receivables	388 464	2 425 944
Investment securities	244 473	243 986
Availability	5 218 264	3 626 698
Prepaid expenses	4 183	14 701
TOTAL ASSETS	5 886 207	6 337 680

Liabilities (in €)	31/12/2018	31/12/2019
Initial endowment (non expendable)	1 000 000	1 000 000
Reserves	3 176 763	3 051 795
<i>Results</i>	(124 968)	(154 166)
Payables	1 606 637	2 005 374
Social, fiscal and other debts	227 775	247 677
Dedicated funds	-	187 000
TOTAL LIABILITIES	5 886 207	6 337 680

Any discrepancies noted between the total and the detail of the items correspond to discrepancies related to rounding.

INCOME STATEMENT

In €	31/12/2018	31/12/2019
Reversals of provisions and depreciation, transfer of charges	-	-
Others products	2 428 287	2 997 493
Exploitation product	2 428 287	2 997 493
Other purchases and external expenses	1 217 002	1 589 558
Taxes other and payments	84 403	74 895
Salaries and treatments	882 471	817 430
Social charges	367 201	351 536
Operating allocations on fixed assets: depreciation charges	32 151	33 234
Other expenses	3	125 000
Exploitation charges	2 583 232	2 991 654
Operating result	(154 945)	5 839
other interests and similar products	36 033	22 265
Net income on sales of marketable securities	-	-
bottom line	36 033	22 265
Exceptional products	-	10 000
Extraordinary charges	-	225
Income taxes	6 056	5 046
Carry forward of unused resources from previous years commitments to be made from allocated resources	-	187 000
THE RESULT OF THE EXERCISE	(124 968)	(154 166)

CHANGE IN FIXED ASSETS

In €€	Gross value at the start of the 2019 financial year	Increase	Decrease	Gross value at the end of the 2019 financial year
Headquarters , research and development costs	16 899	7 501	-	24 400
Total intangible assets	16 899	7 501	-	24 400
Office equipment, computers, furniture	26 380	4 966	6 081	25 265
Total tangible fixed assets	26 380	4 966	6 081	25 265
GENERAL TOTAL	43 278	12 467	6 081	49 665

CHANGE IN AMORTIZATION

In €€	Cumulation at the start of fiscal year 2019	Increase	Decreases	Cumulation at the end of fiscal year 2019
Establishment, research and development costs	37 506			37 506
Total intangible assets	37 506			37 506
Office equipment, computers, furniture	36 595	7 995	6 081	38 509
Total tangible fixed assets	36 595	7 995	6 081	38 509
GENERAL TOTAL	74 101	7 995	6 081	76 015

ACCRUED INCOME

In €€	31/12/2018	31/12/2019
Customers, invoices to be established	370 350	2 320 813
GENERAL TOTAL	370 350	2 320 813

ACCRUED EXPENSES

In €€	31/12/2018	31/12/2019
Supplier invoices not yet received	968 656	1 596 612
Trade payables and related accounts	968 656	1 596 612
Provision for paid vacation	64 287	85 204
Charges on provision for paid vacation	35 490	37 455
Continuing education	8 301	10 382
Payroll taxes	6 052	4 543
Statement of accrued charges	43 609	48 655
Debts customers have to establish	-	-
Social and tax debts	-	-
GENERAL TOTAL	1 126 394	1 782 851

THE USE OF RESOURCES STATEMENT

USES	Resources use statement 2019 (in €)	Allocation by use of collected resources to the public	Collected resources (in €)	Resources collected income statement	Monitoring of collected resources
1 - SOCIAL MISSIONS	2 357 606		1 - COLLECTED RESOURCES TO THE PUBLIC	116 800	-
1.1. Made in France - Actions carried out directly - Payments to other organizations operating in France	2 357 606 2 357 606	116 800	1.1. Donations and bequests collected - Unallocated manual donations - Assigned manual donations - Bequests and other unallocated gifts - Bequests and other donations affected	116 800 116 800	- 116 800
1.2. Made abroad - Actions carried out directly - Payments to a central body or other bodies	- -		1.2. Other products related to the public appeal for generosity		
2 - FUNDRAISING COSTS	244 371		2 - OTHER PRIVATE FUNDS	2 025 285	
2.1. Charity appeal fees public	244 371		3 - SUBVEBTIONS AND OTHER PUBLIC CONTRIBUTION		
2.2. Research costs of other private funds			4 - AUTRES PRODUCTS	742 393	
2.3. Expenses related to the search for subsidies and other public assistance					
3 - OPERATING COSTS	249 181				
I - TOTAL USE FOR THE FINANCIAL YEAR RECORDED IN THE INCOME STATEMENT	2 851 158		I - FINANCIAL YEAR RECORDED IN THE INCOME STATEMENT	2 884 478	
II - ALLOCATIONS TO PROVISIONS	486		II - RETURN OF PROVISIONS		
III - COMMITMENTS TO BE MADE ON	187 000		III - CARRY-OVER OF UNUSED ALLOCATED RESOURCES FROM PREVIOUS FINANCIAL YEARS		
			IV - CHANGE IN DEDICATED FUNDS COLLECTED FROM THE PUBLIC		
IV - RESOURCES SURPLUS FROM THE FINANCIAL YEAR			V - INSUFFICIENT RESOURCES FOR THE EXERCISE	154 166	
GENERAL TOTAL	3 038 644	116 800	GENERAL TOTAL	3 038 644	116 800
V - Share of purchases of gross fixed assets for the year financed by resources collected from the public					
VI - Neutralization of depreciation allowances for fixed assets financed from the first application of the regulation by resources collected from the public					
VII - Total uses financed by resources collected from the public		116 800	VI - Total uses financed by resources collected from the public		116 800
			BALANCE OF RESOURCES COLLECTED FROM THE PUBLIC NOT ALLOCATED AND UNUSED AT THE END OF THE YEAR		-

VALUATION OF CONTRIBUTIONS IN KIND (in €)

Social missions	83 431		Volunteering		
Fundraising costs			Benefits	104 164	
Charges	20 733		Donations in kind		
Total	104 164			104 164	

05

FOUNDATION FOR RARE DISEASES

PROJECTS FUNDED BY THE FOUNDATION





CALL FOR PROJECTS "GENOMICS: HIGH THROUGHPUT SEQUENCING & RARE DISEASES"

RESPONSABLE DU PROJET	LABORATOIRE	TITRE DU PROJET
Sandrine BARBAUX	DRC Institut Cochin	Genetic causes of a rare form of infertility, the sertoli cell only syndrome
Nadine CERF-BENSUSSAN	Laboratory Intestinal Immunity , INSERM UMR 1163 & Institut Imagine Institut National pour la Santé et la Recherche médicale	Whole genome sequencing in Very Early Onset- IBD and related intestinal disorders
Arnaud DUPUIS	Recherche Etablissement Français du Sang Grand Est - Site de Strasbourg	Looking for gene(s) responsible for non syndromic platelet delta storage pool deficiency.
Carole ESCARTIN	CEA	Microglial cells: the third element for mutant Huntingtin clearance in Huntington's disease ?
Céline GAUCHER	Institut Cochin U1016 Université Paris Descartes	High-throughput sequencing for non syndromic dental hard tissues anomalies: ExoDent
Suzanne LESAGE	INSERM U 1127 Pitié-Salpêtrière	Whole genome sequencing in a North African consanguineous family with an early-onset Parkinson's disease
Anne LETESSIER	DRC Institut Cochin	Molecular analysis of Meier-Gorlin Syndrome type 1: consequences of ORC1 mutation on chromatin organization and gene expression
Caroline MICHOT	Imagine Institute	Dissection of molecular bases of myhre syndrome : identification of new genes in pre-screened patients
Isabelle PERRAULT	Genetic ophthalmology Institut des Maladies Génétiques Rares	IGenetic deciphering of new syndrome associating early and severe retinal dystrophy and sensorineural hearing loss.
Sadia SAEED	Université de Lille 2	Identification of new genes and rare variants implicated in monogenic severe obesity in children from a consanguineous population.
Michèle STUDER	Institut de Biologie Valrose (iBV) Université de Nice Sophia Antipolis	Unravelling the Genetic Pathways Leading to Hereditary Congenital Facial Palsy and Associated Hearing Loss

FOLLOWING - CALL FOR PROJECTS "GENOMICS: HIGH THROUGHPUT SEQUENCING & RARE DISEASES"

RESPONSABLE DU PROJET	LABORATOIRE	TITRE DU PROJET
Piotr TOPILKO	Developmental Biology Institut de Biologie de l'Ecole Normale Supérieure (IBENS)	Identification of markers of the cells at the origin of cutaneous Neurofibromatosis type 1 tumours and deciphering the molecular mechanisms responsible for malignant transformation of plexiform neurofibromas.
Aurélien TRIMOUILLE	Génétique Médicale CHU de Bordeaux	Further delineation of molecular bases of Oculo-Auriculo-Vertebral Spectrum.
Clémence VANLERBERGHE	Clinical genetics Centre hospitalier Universitaire de Lille	Deciphering the genetic basis of fibular hypoplasia/agenesis

CHEMICAL HITS OPTIMIZATION

RESPONSABLE DU PROJET	LABORATOIRE	TITRE DU PROJET
Pascale BOMONT	Institut des Neurosciences de Montpellier (INM)INSERM U1051	Hit to Lead discovery for Giant Axonal Neuropathy
Yvon TROTTIER	Institut de Génétique et de Biologie Moléculaire et Cellulaire - IGBMC Translational Medicine and Neurogenetics UMR 7104 CNRS/UdS, Inserm U1258 Fundamental and pathophysiological mechanisms implicated in recessive ataxia	Hit-to-lead development of amyloid aggregation modulators in Huntington's disease and pioneering a new synergic strategy to potentiate their use in therapy

CALLS FOR PROJECTS : "MODELS OF RARE DISEASE"

RESPONSABLE DU PROJET	LABORATOIRE	TITRE DU PROJET
Daniel ABERDAM	INSERM U976, Hôpital Saint-Louis, Paris	Production d'iPSC dérivées de patients atteints d'aniridie, maladie oculaire rare due à des mutations non sens sur le gène PAX6
Fabrice ANTIGNY	INSERM U999, Hopital Marie Lannelongue, Le Plessis-Robinson	Role of Orai1 in the pathogenesis of pulmonary arterial hypertension
Nadia BAHI-BUISSON	INSERM U 1163, Institut Imagine, Paris	Investigating the cellular basis of DYNC1H1 related malformations of cortical development and spinal muscular Atrophy with lower limb predominance using targeted differentiation of human iPS cells
Marc BAUD'HUIN	INSERM UMRS_1238, Université de Nantes	RIBObone
Thierry BIENVENU	INSERM U1266, Institut de Psychiatrie et de Neurosciences de Paris	Development of a new mouse model of anorexia nervosa, a knock-in mice model carrying a missense variant in the cholecystokinin A receptor (CCKAR)
Thomas BOULIN	CNRS UMR5310, INSERM U1217, Université Lyon 1, Institut NeuroMyo- Gène, Lyon	Modeling disease-causing mutations of Neurobeachin/NBEA in the nematode <i>Caenorhabditis elegans</i>
Valérie DUPE	Institut de Génétique et Développement de Rennes IGDR UMR6290 CNRS Uni- versité de Rennes	Assessment of functional relevance of synonymous variants in Shh identified in patients with holoprosencephaly
Olivier GOUREAU	INSERM UMRS_968, UPMC, Institut de la Vision, Paris	Modeling retinitis pigmentosa using retinal organoids derived from patient-specific induced pluripotent stem cells
Laurence LEGEAI-MALLET	INSERM U 1163, Institut Imagine, Paris	Zebrafish model of X-linked hypophosphatemia

FOLLOWING - CALLS FOR PROJECTS : "MODELS OF RARE DISEASE"

RESPONSABLE DU PROJET	LABORATOIRE	TITRE DU PROJET
Philippe LORY	Institut Génomique Fonctionnelle, Département Neuroscience, Montpellier	A mouse model of Childhood Cerebellar Atrophy (ChCA) is required to decipher the pathogenic mechanism and to design a therapeutic option
Massimo MANTEGAZZA	Institut de Pharmacologie Moléculaire et Cellulaire IPMC UMR7275, Nice	Knock-in mouse model for studying and treating negative dominant SCN2A mutations: a novel pathological mechanism specific of autism mutants.
Grégoire MICHAUX	Institut de Génétique et Développement de Rennes UMR6290 CNRS Université de Rennes	Role of V0-ATPase in intestinal absorption and Microvillus Inclusion Disease
Christelle PEYRON	Centre de Recherche en Neurosciences de Lyon INSERM U1028, CNRS UMR5292, Univ Lyon	Development of an hypocretin-KO rat model of narcolepsy type 1
Frederic RELAIX	Institut Mondor de Recherche Biomédicale Equipe 10, Paris	Accurate preclinical modeling and treatment proof-of-principle for Duchenne Muscular Dystrophy
Hamid-Reza REZVANI	INSERM U1035 BMGIC Université Bordeaux	Modelling of Pigmentary abnormalities in xeroderma pigmentosum type C

CALL FOR PROJECTS "HUMAN AND SOCIAL SCIENCES"

RESPONSABLE DU PROJET	LABORATOIRE	TITRE DU PROJET
Nathalie ANGEARD	Université Paris Descartes	Entraînement à la Théorie de l'Esprit en Réalité Virtuelle dans la forme infantile de la Dystrophie Myotonique de Type 1 (DM1)
Pascal ANTOINE	Université Lille 3 UMR CNRS 9193 SCALAB -équipe DEEP	Dyade : Dynamique de Couple dans la Maladie de Huntington
Alexandra FOUBERT- SAMIER	BORDEAUX Centre INSERM psychogéron- tologie,)épidémiolo- gie,psychologie de la santé	Qualité de Vie des patients atteints d'une Atrophie Multi Systématisée et de leurs aidants
Marcela GARGIULO	UNIVERSITE Paris Descartes institut de psychologie Laboratoire de Psychologie, Psychopathologie, Psychanalyse	Projet : PARENTALITE et Amyotrophie Spinale Etude multidimensionnelle de la surcharge des parents . Étude Franco-chilienne
Jean-Pierre POULAIN	Université Toulouse Jean Jaurès Sociologie CERTOP SANTAL	Socialisations des Pratiques Alimentaires des enfants avec un syndrome de Prader-Willi - SoPAP
Amélie ROCHET- CAPELLAN	Université Grenoble-Alpes Sciences du langage GIPSA-lab - Equipe de recherche inter- disciplinaire sur la parole et la communication	ParticipAAction : Un corpus participatif de la Communication Augmentée et Alternative des personnes avec un syndrome neurogénétique rare pour la comprendre et l'améliorer

CALL FOR PROJECTS "JTC 2019 ON RARE DISEASES WITHIN EUROPEAN JOINT PROGRAMME ON RARE DISEASES (EJP RD) "

32 FUNDING BODIES AND AGENCIES (INCLUDING THE RARE DISEASES FOUNDATION) JOINTLY WITH THE EUROPEAN COMMISSION

RESPONSABLE DU PROJET	LABORATOIRE	TITRE DU PROJET
Agnès RÖTIG	Institut Imagine, INSERM U1163	Mitochondrial Disorders: from a global registry to medical genomics, toward clinical trials
Hélène CAVÉ	Département de Génétique, CHU Hôpital Robert Debré and INSERM UMR1131 - Institut de recherche Saint-Louis, IRSL - Université de Paris	European network on Noonan syndrome and related disorders
Armelle YART	Institute for Metabolic and Cardiovascular Diseases INSERM U1048	European network on Noonan syndrome and related disorders
Hélène DOLLFUS	Strasbourg University, Medical Genetics Research Laboratory INSERM unit (U1112), Faculté de Médecine	Solving missing heritability in inherited retinal diseases using integrated omics and gene editing in human cellular and animal models
Lydie DA COSTA	Service d'Hématologie Biologique Robert Debré Hospital	The European Ribosomopathy Consortium



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