



## Call 2013 - May: 'High throughput sequencing and Rare Diseases'

Understanding the molecular basis of human diseases of genetic origin is far from being achieved. It remains, in particular, to identify the genes that are the most rarely involved. Identification of these genes is of paramount importance to elucidate the pathogenic mechanisms of the diseases and for the development of diagnostic tools and of innovative therapeutics.

In 2012, the **Rare Diseases Foundation ("Fondation maladies rares")**, a new Foundation dedicated to promote research on rare diseases, was created. This Foundation is a key feature of the second French National Rare Disease Plan (2011-2014). **Fondation maladies rares** operates with public and private sources of funding to stimulate, coordinate and fund fundamental, clinical and translational research. Founders include the French Muscular Dystrophy Association (Association Française contre les Myopathies – AFM-Téléthon), the Rare Diseases Alliance, the French National Institute of Health and Medical Research (Inserm), the Conference of General Directors of the French University Hospitals and the Conference of Presidents of French Universities. The Foundation will play a pivotal role to cover all domains of research in rare diseases, with a strong emphasis on translational research.

The Foundation promotes and supports research on rare diseases through the launch of specific calls for projects. Thus, it continues and extends the actions previously initiated by the GIS-Institute for Rare Diseases - today embedded in the Rare Diseases Foundation - in particular those facilitating access for researchers to strategic technology platforms. All the scientific activities of the Foundation are administrated by its administrative board, composed by founders and qualified personalities. The Foundation benefits from the guidance of a Scientific Committee composed of leading medical specialists and scientists in the field of rare diseases.

## Objectives of the call

**The goal of the open call for proposals is to support research projects aimed at identifying – by the use of exome sequencing - genes involved in rare diseases whose molecular basis remains unknown or incompletely known.**

- PROPOSAL SUBMISSION

Projects will be evaluated by external referees and selected by the **Scientific Committee of Fondation maladies rares**. The selection results will be communicated by mail to the principal investigator. - **Please notify in the proposal any possible conflict of interest** -

- \* Applicants **resubmitting projects** are required to provide a detailed answer to the comments provided by the **Scientific Committee of Fondation maladies rares** at the previous session and highlight changes in the revised version.
- \* Applicants, who were **principal investigators** or **partners** in a project or **whose team** was **previously funded** by the GIS-Institute for rare diseases or by Fondation maladies rares since 2009 are required to provide a **detailed report on the results and impacts of all projects funded**. For **ongoing projects**, a detailed progress and / or preliminary data **report** is required.
- \* Please fill out **one submission form per independent project**.

**If these items are not fully answered, the submitted project will not be considered for funding.**

## CALL DATE FOR 2013-May:

Call launch: 2013, May 22<sup>th</sup>. Deadline for submission: 2013, June 20<sup>th</sup>.

After completion of the submission form:

1/ Please, keep one copy for your files

2/ Send an electronic copy

(Document title: Name of applicant-HTS-RD, in a word.doc format) to:

[ngs@fondation-maladiesrares.com](mailto:ngs@fondation-maladiesrares.com)

3/ Send one signed paper copy to:

Fondation maladies rares

Appel à Projets 'Séquençage à haut débit et maladies rares'

Plateforme Maladies Rares, 96 rue Didot, 75014 Paris

- FUNDING

Fondation maladies rares provides **financial support** for exome sequencing and bio-informatics analyses provided by the sequencing platforms acting as partners of the foundation. Funding cannot include equipment or personal costs. **The foundation may financially support either partially or in total, any project submitted to the call. Co-fundings must be indicated for each project.**

- ACCESS TO PLATFORMS

**Fondation maladies rares determines, for each selected project, the platform on which the project will be performed.** This will allow optimization of the handling of projects by each platform, according to its capacity and calendar, with the aim to deliver the results in the shortest possible time.

- DNA SAMPLES

**DNA samples** (quality checked) **MUST be available at time of project submission** and sent to the platform **within one month** after reception of the approval letter from the Foundation.

In case DNA samples should not be ready, the project would be reviewed at the next call for project.

It must be stressed that **in no case the number of DNA samples can be smaller** than that specified in the original application, except if decided by the Scientific Committee.

- SHARED DATABASE OF RARE VARIATIONS

Applicants submitting projects agree that **the genomic data obtained** through funding from Fondation maladies rares **will be released after anonymization into the new national shared database of rare variations.**

- COMMUNICATION

The title of the selected projects and name of their principal investigator will be published on the web site of Fondation maladies rares (<http://fondation-maladiesrares.org>).

**Acknowledgement Policy:** It is required that projects funded by the “Fondation maladies rares” be acknowledged in all publications and communications. Please send us the reference(s) of the publication(s).