

Open Call for proposals High Throughput sequencing “ Plateforme Mutations ”

The identification of 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. These genes are nevertheless crucial at the scientific level, and their identification is of paramount importance for the development of diagnostic tools, the understanding of the biological processes involved and the development of innovative therapeutics. The GIS Institute for Rare Diseases and IBISA, together with the INSERM Institutes of Genetics and development and Neurosciences, neurology and psychiatry, and Genoscope - Centre National de Séquençage -, have decided to settle a platform for the identification of mutations, referred as to the “ Plateforme Mutations “. This platform will use a combined approach of enrichment by hybridization and high throughput sequencing to identify rare mutations in human pathologies, with a particular focus on monogenic diseases.

The goal of the open call for projects launched by the “ Plateforme Mutations “ is to support research projects that aim at identifying genes involved in diseases whose molecular basis remains unknown.

The projects can be sent all year long. They will be evaluated and selected by the Scientific Committee (SC) of the “ Plateforme Mutations “ with the help of external experts. The SC will meet 4 times a year, April 16, July 9, October 15, and December 15, 2009. Projects should be received by the “Plateforme Mutations” secretariat 4 weeks before the indicated dates, with exception for the first SC meeting for which applications should be received (by E-mail) no later than April 2, 2009. Any project received after these deadlines will be taken into consideration for the next session.

Each selected project will benefit from the expertise of the “ Plateforme Mutations “ for the refinement of the strategy to choose with respect to the objectives of the project and available material.

The costs of the consumables (arrays, fluids...) will be at the research team's expense. Please, consult the budget page (end of application) for information on how to proceed to calculate the cost of your project.

The GIS-Institute for rare diseases, in association with the French Association against Myopathies, may provide a financial support for consumables to the selected projects on rare diseases.

After completion of the submission form:

- 1/ Please, keep one copy for your files
- 2/ Send an electronic copy (word.doc format) to: skoutouzov@gis-maladiesrares.net
- 3/ Send one paper copy to:
Sophie Koutouzov
Gis-Institut des Maladies Rares
Hôpital Broussais
102 rue Didot, 75014 Paris

Informations : pfm@genoscope.cns.fr

Call for proposals

I - Project Identification form

Project title (maximum 120 signs)

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Rare diseases project:

Yes
No

A1-Principal investigator

A principal investigator, responsible for the project, must be designated by the partners if the project involves several partners

Mr, Ms	Last name	First name
.....
Position	
Mail	
Tel		Fax

Laboratory (complete name)				
.....				
N° Unit (Dept) (if any)			
Complete address of the laboratory				
.....				
City		Zip code		Region
Institution (name of the institution with ability to financially administer the project) :				
.....				

Other partners

<i>Mr, Ms</i>	<i>Last name</i>	<i>First name</i>	<i>Laboratory (complete name/address)</i>	<i>Role and contribution</i>

Number of personnel involved in the project (person-months):

Je déclare exactes toutes les informations contenues dans ce document

Visa du Directeur du laboratoire

Lu et approuvé, date et signature du coordinateur du projet

Nom, prénom, date et signature

II – Description of the project

Abstract (Not exceeding 1000 characters)
(objectives, expected results)

1 – Objectives: (1 page maximum, arial 11, single space)

Situate the project in its scientific context with indications 1 /on the targeted disease(s) and available knowledge 2/ acquired or preliminary results obtained by the applicant(s).3/ Objectives. Include 3 general references (reviews, articles directly dealing with the questions addressed by the project...).

2 – Description of samples available for the study (1 page)

Describe a) patients that will be analyzed in this project (family cases / trios / sporadic cases ...), with information on clinical phenotyping; b) numbers of biological samples, quantity of available DNA (please make sure that the DNA has been prepared according to the platform recommendations)

3 – Genes, genomic region, cumulated size

Important: please indicate and justify the size of the regions to be sequenced (number of genes to be sequenced, size of intervals to be sequenced ...) and whether the project involves sequence capture

4 – Existence of similar and/or competing projects (1/2 page)

Situation of the project in the international competition, international coordination; priority of the project

5 – Expertise of the laboratory in analysis, annotation and interpretation of sequences to be produced in this project

6 – Briefly describe the downstream research you intend to perform, including possible collaboration, for which this sequence information is a pre-requisite (1/2 page)

7 – Main publications

List of main publications and/or patents of the last three years with particular attention to those related to the project (from the principal investigator and associated teams)

III – Budget information

1 – Calculate the cost of your project

Before filling in this part, please contact pfm@genoscope.cns.fr to get an estimated cost of your project

In your request, please, clearly indicate:

- Number of samples (patients/controls)
- Size of the region (exons) to be sequenced (cumulative size or regions)
- Reading coverage (X)

Please, indicate here the cost of your project:

2 - Funded projects related to the current application (principal investigator and associated teams) in the last three years (ongoing and achieved)

Name (coordinator or associated team)	Name of the call for projects/ Source/funding amount	Project title	Project period

* Add as many columns as needed

3 - Funding requests UNDER EVALUATION related to the current application (principal investigator and associated teams)

Name (coordinator or associated team)	Name of the call for projects/ Source/funding amount	Project title	Project period

* Add as many columns as needed

3 – FUNDING REQUESTED FROM GIS – Institute for Rare Diseases: