

## DESCRIPTION OF THE PROJECT

### 1. Objectives (1 page maximum, arial 10, 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 addressed questions).

### 2. Strategy and samples included in the study (3 pages maximum)

**Important:** Clearly describe and **JUSTIFY** the strategy, the experimental model and associated technics, and the choice of samples included in the study.

- For Whole exome (WES) and whole genome sequencing (WGS) projects:

Specifically mention the following criteria: modes of inheritance, family/sporadic cases, genetic heterogeneity, etc. in the choice of strategy.

Describe a) samples available and samples that will be analyzed in the project, with precise information regarding clinical phenotyping, pedigree, linkage studies; b) numbers of biological samples, quantity available and the criteria used for their selection for inclusion in the project.

*\* DNA Samples (with quality checked) as well as informed consent from patients **MUST** be available at time of project submission!\**

\* Important issue for WGS projects:

- If you have already performed whole exome studies on the same disease and/or on the selected patients, provide detailed results of these studies (technical aspects: sequencing platform, coverage/depth sequencing, complete process of data analysis, variants interpretation and validation...) and justify the proposed strategy of whole genome sequencing and expected results

- In case whole exome study has not been conducted previously, clearly state why WGS will be more suitable than WES for the present project

- For RNAseq projects:

Specifically mention the following criteria: samples information (material, treatments or genetic modifications prior to RNA isolation, sample abundance, etc.), RNA information and method of isolation, RNA quantification and quality control, library construction, sequencing (format, read length, etc.), RNA-Seq design including replicate number, sequencing depth.

*\* RNA Samples (with quality checked) as well as ethical approvals to biological resources **MUST** be available at time of design validation by the platform!\**

For projects relying on combined approaches, please clearly indicate the part(s) of your project for which you request financial support.

### 3. Sequencing data analysis and interpretation (1,5 page)

Clearly state objectives of bioinformatic analyses, key stages of analyses and tools employed. Indicate how sequencing data analysis and interpretation will be managed. Expertise of the laboratory and its collaborators and bioinformatics resources available will be clearly stated.

For projects not using WES or WGS, please provide a detailed budget (based on a quote provided by a partnering platform) of the sequencing and data analysis.

### 4. Clearly describe the strategy that will be engaged to validate the results and the downstream research you intend to perform (1 page)

Please state collaboration(s) involved.

### 5. Existence of similar and/or competing projects (1/2 page)

Situation of the project in the international competition: priority of the project.