FONDAZIONE

CALL FOR RESEARCH PROJECTS - 2019

FULL EVALUATION GUIDELINES - COMMITTEE MEMBERS

According to the Telethon mission, this call is in support of research projects focusing on **diseases of proven genetic origin,** either monogenic or polygenic forms.

For **multifactorial diseases** the following applies:

- Studies focused on **monogenic or polygenic forms** of multifactorial diseases are **ELIGIBLE**
- Studies focused on the **identification of genetic risk factors** (e.g. SNPs or other predisposing variants) for multifactorial diseases are **NOT ELIGIBLE**.

APPLICATION'S SECTIONS AND EVALUATION PHASES

The **Full Application** comprises two parts: the **Core Project** and the **Supplementary Contents** (details are available in the *2019 Call for Applications* and *Guidelines*).

The Core Project is the Application's part evaluated during the triage phase. **During the Full Evaluation, all parts of the Application** (Core Project plus Supplementary Contents) **are made available to the Committee members**.

FULL EVALUATION INSTRUCTIONS

REVIEWERS' ROLE - Each application is reviewed and scored by three Committee members (hereon "Reviewers"). The **primary** reviewer is responsible for the project's presentation during the plenary review session. **Primary and secondary** reviewers have to provide written comments, whereas **tertiary** one is not requested to (but may provide written comments, if he/she wants to).

EVALUATION – The Reviewers are requested to fill in the "Full Project Evaluation" available in *TETRA - Telethon Projects Managements system portal* at https://projects.telethon.it accessible through personal login and password.

In support of their evaluation, Reviewers will be provided with written comments by External Reviewers, who are chosen *ad hoc* for each Application by Telethon Research Program Managers.

Scores

Each project requires **two scores**:

- **Scientific Merit** (is the proposed research excellent?)

  Relative weight: 90%

  Score range: from **1.0 (poor)** to **5.0 (outstanding)** by 0.1 unit increments.

Full Evaluation Scoring Scale		
Score	**Value**	**Description**
4.6 - 5.0	Outstanding	No concerns
4.0 - 4.5	Excellent	No substantial issues need discussion
3.0 - 3.9	Good	Only one or a few addressable concerns
2.0 - 2.9	Average	Several concerns in one or more Aims
1.0 - 1.9	Poor	Major concerns in one or more Aims

- **Impact on patients** (how close to therapeutic development or to any other potential impact on patients are the proposed studies?)

  Relative weight: 10%

  Scores and scoring criteria (on the basis of the proposed research activity):
  Score=5: Clinical Trials. Therapeutic clinical trials, palliative clinical trials or clinical trials to validate diagnostic tools, natural history of disease.
  Score=4: Preclinical Studies; Disease Gene Identification. Pre-clinical studies testing efficacy/safety of therapeutic strategies (in vitro and/or in animal models); discovery of new disease genes.
  Score=3: Mechanisms, Structure, Function, Targets, Drug Discovery. Basic (laboratory) studies on mechanisms; functional/structural omics studies; search for new therapeutic targets.

The **overall score** will be automatically calculated by combining the two scores according to their relative weight.

Written Comments

Written comments are an essential part of the review and are critical in developing summary statements for the Applicants.

The individual written comment will be anonymously incorporated into a complete review report that will be fed back to the Applicant. It is therefore important that the written material is accurate, clearly written, and does not include derogatory language.

Please note: External Reviewers' written comments will also be included as such in the review report.

Description (max 2,000 characters including spaces)
Primary reviewers only are requested to fill in the description field. This summarises the objectives of the study and the hypothesis to be tested, it also concisely describes the specific aims and procedures of the proposed research.

Scientific Merit (max 12,000 characters including spaces)
This section should present a comprehensive evaluation of the application.

For revised applications only, the Reviewers will provide their evaluation of the changes and responses to the critiques from the previous review, with the indication of whether the Application has been improved comparing to the previous submission. The Telethon Review Report of the previously submitted application together with the Applicant's rebuttal are available within the Application (*Cover Letter* section).
The Reviewers evaluate the overall scientific merit of the proposal by providing an analysis of the strengths and weaknesses on the basis of the following parameters:

- *Link to genetic diseases*: is the proposal addressing a genetic disease? Does the proposal bear the potential to advance knowledge on the disease(s) of interest?

- *Significance*: Does this study address an important problem? If the aims of the application are achieved, how will scientific knowledge be advanced? What will the effect of these studies be on the concepts or methods that drive this field?
- *Originality of science*: Does the project employ novel concepts, approaches or methods? Are the aims original and innovative? Does the project challenge existing paradigms or develop new methodologies or technologies?
- *Appropriateness of design* and methods: Are the conceptual framework, design, methods, and analyses adequately developed, well-integrated, and appropriate to the aims of the project?
- *Preliminary results*: Are proof-of-principle experiments provided, adequately supporting new principles to be tested in the grant? Are novel tools or reagents well-characterized?
- *Feasibility*: Does the applicant acknowledge potential problem areas and consider alternative tactics?
- *Safety*: Please evaluate the adequacy of the proposed protection for humans, animals or the environment, to the extent they may be adversely affected by the project proposed in the application (if any).
- *Previous achievements* (dedicated section for former grantees only): did the previous Telethon grant produce relevant results relative to the stated aims?

Note: thanks to the agreement between Fondazione Telethon and CINECA (a non-profit Consortium made up of Italian universities, research institutions and the Ministry of Education, University and Research; http://www.cineca.it/en), successful applications entailing next generation sequencing experiments or high performance computing will gain access to CINECA's bioinformatics services. For this reason, applications include a dedicated section that will allow CINECA's technical team to assess and validate the requests, where applicable.

Impact on patients (max 2,000 characters including spaces)
What is the potential of the proposed project to make progress towards therapy or to provide any other impact on patients' clinical management and/or quality of life? How close in time is such a development envisaged?

Comments on Applicant (max 2,000 characters including spaces)
Is the investigator appropriately trained and well-suited to carry out this work? Is the work proposed proportionate to the level of experience of the **principal investigator and key personnel**? Is the Applicant a significant player in the field of the submitted research project?

Please note that Fondazione Telethon does not apply assessment of Candidate's CV based on journal-based metrics, such as Journal Impact Factor. The Fondazione signed and endorses the San Francisco Declaration on Research Assessment (DORA, http://www.ascb.org/dora/).

Key Personnel - Please note that Italian public bodies, such as universities, are obliged, in compliance with the national legislation in force, to recruit personnel through public competition announcements containing all the selection and appointment criteria. Consequently, in the proposal only the skills/profile needed can be outlined and the names of the personnel to be enrolled will become known only after the public selections procedures have been carried out.

Comments on Budget Allocation (max 2,000 characters)
For single-center Applications a maximum of 100,000 €/year is allowed.
For two-center Applications a total budget of 160,000 €/year is allowed; the Coordinator may ask for a maximum of 100,000 €/year.
Reviewers evaluate the appropriateness of the budget in relation to the proposed research and whether all items are considered realistic and justified. Reasons for each recommended modification in amount

or duration of support must be presented. Reviewers are asked to identify any apparent scientific or budgetary overlap with active or pending support.

Overall evaluation (max 2,000 characters)
This section is for the Reviewers to summarize the **key reasons** for their overall rating, indicating the relative **strengths**, **weaknesses** and overall **final considerations**.