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.

The FT GENERAL GRANT project duration can range from 1 to 3 years.

The call also includes the following funding opportunities:

- The TELETHON CAREER AWARD in support of young researchers proposing – as Lead Applicant - innovative projects in the field of rare genetic diseases and with the need of a salary coverage. This award is open to researchers with a minimum of 2 and a maximum of 7 years’ experience since completion of the PhD (or specialization for medical degree). FT will provide salary coverage for the entire duration of the project, which is fixed for 3 years.

- The FONDAZIONE PISANA PER LA SCIENZA/FONDAZIONE TELETHON (FPS/FT) award aimed at attracting researchers willing to join the scientific community of the Fondazione Pisana and proposing innovative projects exclusively in the field of neurological and/or cardiovascular rare genetic diseases (http://www.fpscience.it/en/research-activities/5/). This award is open to researchers of all levels of seniority demonstrating the ability to formulate and conduct an independent research line in synergy with the scientific activities of FPS. FPS will provide salary coverage for the principal investigator for the entire duration of the project, which is fixed for 3 years.

APPLICATION’S SECTIONS

The Full Application includes information directly embedded from the initial Letter of Intent (LOI); further information of the Letter of Intent can be found in the LOI section at end of the Application.

EXTERNAL REVIEWER’S ROLE

External reviews will be made available to the Telethon Scientific Committee members, to support them in the evaluation process.
FULL EVALUATION INSTRUCTIONS

External Reviewers are requested to fill in the “External Reviewers Evaluation Form” available in TETRA - Telethon Projects Managements system portal at https://projects.telethon.it accessible through personal login and password.

Written Comments

Written comments are an essential part of the review and are critical in developing review reports 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.

_Description_ (max 2,000 characters including spaces)
This should summarise the objectives of the study and the hypothesis to be tested, it also concisely describes the specific aims and procedures of the proposed research.

SCIENCTIFIC COMMENTS

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).

- **Scientific Merit** (max 12,000 characters including spaces): the Reviewers are asked to 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?
  - _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?
  - _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?
  - _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?
  - _Feasibility, Pitfalls and Alternative Approaches_: Does the applicant acknowledge potential problem areas and consider alternative tactics?
  - _Significance and Innovation_: Does this study address an important problem? If the aims of the application are achieved, how will scientific knowledge be advanced? What will be the effect of these studies be on the concepts or methods that drive this field?
  - _Safety_: 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?
• **Impact on patients and relevance to Telethon's mission** (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)

A maximum of 80,000 €/year for research expenses is allowed for all the different funding opportunities.
The Lead Applicant salary coverage request is not allowed except for:
**Career Award**: FT will provide salary coverage for the 3 years project, for an amount of maximum 40,000 €/year.
**Fondazione Pisana per la Scienza/FT award**: FPS will provide salary coverage for the Lead Applicant for the 3 years project. Salary will be established based on the seniority of the researcher, in due time, once the project will be selected for funding.

Reviewers are asked to 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.

**Overall recommendation**
Based on the specific points raised in the written critique, the External Reviewers are asked to choose their recommendation as follows:

A - Strongly recommended; no substantial points need discussion.
B - Recommended; minor points need discussion.
C - Revision recommended; a small number of specific major points need to be addressed.
D - Major revision necessary; multiple or fundamental problems can be addressed in a major revision.
E - Revision not recommended; multiple or fundamental problems that will be difficult to address in a revision.