



| Reference Number | |
|---|--|
| Institution | |
| Lead Applicant | |
| Partners | |
| Total Requested | |
| General information | |
| Project Overview | |
| General Information | |
| deneral illioittiation | |
| roject title | |
| | |
| umber of Centers | |
| A maximum of 2 centers are allowed on this application. Indicate the num | ber of centers participating in the study, including the |

Project Duration (in months)

Coordinator Center (Lead applicant).

Cannot be more than 24 months. Only 12 or multiples of 12 are allowed

Total Budget Requested

Have you applied to a previous Fondazione Telethon Call?

Reference Number Page 1 of 36





| Has your project b | een funded? |
|---------------------|--|
| | |
| Have you applied t | to a previous Fondazione Cariplo Call? |
| | |
| Has your project b | een funded? |
| O | |
| | to a previous Fondazione Cariplo - Fondazione Telethon Joint Call? |
| o | |
| 0 | |
| Has your project b | een funded? |
| 0 | |
| • | |
| Type of Applicant | |
| | |
| Type of Applicatio | n |
| | |
| Previous Applicati | on Number |
| Type the previous | s Application Number in case of: Revised Application , Renewal Application |
| | |

Reference Number Page 2 of 36





| Previous role |
|--|
| Select your previous role in case of: Revised Application, Renewal Application |
| |
| |
| cover letter |
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Reference Number 3 Page 3 of 36





| Review Summary of the Previous Application | |
|---|----------------|
| | |
| | Total Files: 1 |
| Overview | |
| Abstract | |
| | |
| | |
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| | |
| | |
| Coordination and Management - Multicenter Studies | |
| | |
| | |
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Reference Number Page 4 of 36





| Role and Contribution of Partner(s) in the Project - Multicenter Studies | | | |
|--|--|--|--|
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| Relevance to the Call | | | |
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| | | | |
| Lay Summary - English | | | |
| Lay Summary Linguist | | | |
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| | | | |
| | | | |
| Project Title - Italian | | | |
| rioject ritie - italian | | | |
| | | | |
| Lay Summary - Italian | | | |
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| | | | |

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Type of Research **Disease Name Disease OMIM Number ICD-11 Code ORPHA Number Mesh Terms** Indicate Tdark(s) Indicate the rationale for which the Tdark(s) is/are related to the proposed disease **Research Type**

Reference Number Page 6 of 36





| Area of Research | | |
|---|--|--|
| Research Steps | | |
| | | |
| Does the project involve bioinformatic analyses? | | |
| | | |
| Please specify the type of analysis that will be performed and who will be responsible for such analysis. | | |
| | | |
| Overall Description of the Research Project (Triage Phase) | | |
| Please describe central hypothesis, objectives, specific aims, research design, methods, and potential outcomes | | |
| | | |
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Reference Number Page 7 of 36





| R | Research Proposal | | |
|---|--|--|--|
| Н | ypothesis, Background and Rationale for Proposed Research (Why?) | | |
| | ,, | | |
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| | | | |

Reference Number Page 8 of 38





| Hypothesis, Background and Rationale for Proposed Research Figures | | |
|--|----------------|--|
| | | |
| | Total Files: 1 | |
| Research Plan, Research Design and Methods (How?) | | |
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Reference Number Page 9 of 36





| Feas | easibility, possible Pitfalls and alternative approaches | | |
|------|--|--|--|
| | | | |
| | | | |
| | | | |
| | | | |
| | | | |
| Enga | gement and training of young researchers | | |
| | | | |
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| | | | |
| | | | |
| Gant | t Chart | | |
| | | | |
| | | | |

Total Files: 1

Reference Number Page 10 of 38





Impact

| Expected results on the scientific community and on patients with rare disease in the long-term | | | |
|---|--|--|--|
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| | | | |
| Literature | | | |
| | | | |
| Cited Literature: | | | |
| | | | |
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Reference Number Page 11 of 36





Ethical Documents Human Subjects If the grant is approved for funding, funds WILL NOT BE PROVIDED until the pertinent Ethical Documentation has been obtained. Please activate in due time all necessary procedures to obtain this approval in accordance with the relevant Italian laws. For clinical studies, please refer to https://www.aifa.gov.it/modulistica-sperimentazione-clinica. Fondazione Telethon ETS (and/or its Alliance partners, in case of Joint Calls, Initiatives) reserves the right to ask for a copy of all the relevant approval documentation Indicate whether the study involves (consider both Applicant and collaborator(s) activities): Please indicate if the Ethical Authorization is already in place. **(** Please, specify the origin of the samples. If the study involves patient-derived samples, please include a summary of patients' details (e.g., age, sex, origin, genetics, ...) **Vertebrate Animals** If the grant is approved for funding, funds WILL NOT BE PROVIDED until the pertinent Ethical Documentation has been obtained. Please activate in due time all necessary procedures to obtain this approval in accordance with the relevant Italian laws. Fondazione Telethon ETS (and/or its Alliance partners, in case of Joint Calls, Initiatives) reserves the right to ask for a copy of all the relevant approval documentation. Indicate if the study involves the use of vertebrates or not. **(**

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| Please indicate if the Authorization is already in place. | | |
|---|--|--|
| ⊙ | | |
| | | |
| | | |
| | | |
| Host Institution | | |
| | | |
| Applicant Details | | |
| | | |
| Permanent position | | |
| | | |
| Position Title | | |
| | | |
| | | |
| Main Research Fields | | |
| | | |
| | | |
| Name of Laboratory/Clinical Unit | | |
| | | |
| | | |
| Number of staff members | | |
| | | |
| Are you the Chief of the Laboratory/Clinical Unit? | | |
| | | |
| Chief of the Laboratory | | |
| Onlier of the Laboratory | | |

Reference Number Page 13 of 36





Independence Statement

| An Independence statement must be uploaded before final submission (PDF only). | |
|--|----------------|
| | |
| | Total Files: 1 |
| Any other appointment (including foreign)? | |
| Other Appointment | |
| Financial Interests Disclosure | |
| | |
| Facilities And Resources | |
| Laboratory space | |
| Clinical Resources | |
| Office space | |

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| Majo | Major Equipment | | | | |
|------|-------------------------|-------------|-------|------|--|
| | | | | | |
| | | | | | |
| | | | | | |
| Core | Facilities and Services | | | | |
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| Con | ntacts | | | | |
| | | | | | |
| An | Approver | | | | |
| ٦,١٦ | F. 3. 0. | | | | |
| # | Full Name | Institution | Email | Role | |
| | | | | | |

Reference Number Page 15 of 36





Collaborators

Use icon to invite collaborators. Once they have accepted the invite they will be able to see this application.

To remove invited Collaborators, click on and click on icon displayed next to their names in the list.

| Prefix | First Name | Last Name | Email | Role | Status |
|--------|------------|-----------|-------|------|--------|
| | | | | | |

Active Collaborators

| Table below displays all | Table below displays all Active collaborators added on the application. | | |
|--|---|--|--|
| # Full Name: Institution: Email: Role: Laboratory: | | | |
| Contribution: | | | |

Partners

Reference Number Page 16 of 36



Prefix First Name Last Name



Role

Status

Use icon below to invite Multicenter application partners on your application. Once they have accepted, they will be able to view this application, and complete their part of the application package.

To remove invited Multicenter application partners, click on and click on icon displayed next to their names in the list.

Email

You may add up to 1 partners on this application. You will not be able to submit your application if number of partners exceed this limit.

| dget | | | |
|-----------------------|-------------|---------------------|------|
| | | | |
| Principal Investigato | | | |
| Application Number: | | | |
| Institution Name: | | | |
| Project Title: | | | |
| Year 1 | | | |
| Equipment | | | |
| Item | Description | Managed by | Cost |
| | | Managed Directly | |
| | 1 | | |
| IT Equipment | | | |
| Item | Description | Managed by | Cost |
| | | Managed Directly | |
| | | | |
| Software | | | |
| Item | Description | Managed by | Cost |
| | | Managed Directly | |
| | | | |
| Materials, Supplies a | nd Services | | |
| Item | Description | Managed by | Cost |

Reference Number Page 17 of 36





| Travel | | | | |
|----------------------|--------------------------------------|--------------|------|--|
| Item | Description | Managed by | Cost | |
| | | | | |
| | | | | |
| Other Evpenses | | | | |
| Other Expenses | D | Adam and the | 0 | |
| Item | Description | Managed by | Cost | |
| | | | | |
| | | | | |
| | | | | |
| Overheads | | | | |
| Item | Description | Managed by | Cost | |
| | | | | |
| | | | | |
| | | | | |
| Personnel & Sala | aries | | | |
| Role on Project | | | | |
| Annual effort % | | | | |
| Name | | | | |
| Surname | | | | |
| Birthdate | | | | |
| Degree | and the file and to add the discount | | | |
| | at the Host Institution | | | |
| Is this contract al | | | | |
| Managed by | equesteu: | | | |
| Salary | | | | |
| | | | | |
| Role on Project | | | | |
| Annual effort % | | | | |
| Name | | | | |
| Surname Birthdate | | | | |
| Degree | | | | |
| Degree | | | | |

Reference Number Page 18 of 36





| Type of Contract at t | | | |
|-------------------------|--------------|--------------------------|------|
| Is this contract alread | | | |
| Is a salary being requ | uested? | | |
| Managed by | | | |
| Salary | | | |
| | | | |
| | | Total Direct Costs: | |
| | | Indirect Costs: | |
| | | Total Estimated Budget: | |
| Year 2 | | Total Zollinatou Zuugoti | |
| Materials, Supplies | and Services | | |
| Item | Description | Managed by | Cost |
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| | | | |
| | | | |
| | | L | |
| Travel | | | |
| Item | Description | Managed by | Cost |
| | | | |
| | | | |
| Other Expenses | | L | |
| Item | Description | Managed by | Cost |
| item | Description | Wianageu by | COST |
| | | | |
| | | | |
| | | | |
| | | | |
| | | | |
| Overheads | | | |
| Item | Description | Managed by | Cost |
| | | | |
| | | | |
| | | L | |
| Personnel & Salaries | S | I I | |
| Role on Project | | | |
| Annual effort % | | | |
| Name | | | |

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| Surname | | |
|--|---------------------|--|
| Birthdate | | |
| Degree | | |
| Type of Contract at the Host Institution | | |
| Is this contract already active? | | |
| Is a salary being requested? | | |
| Managed by | | |
| Salary | | |
| Role on Project | | |
| Annual effort % | | |
| Name | | |
| Surname | | |
| Birthdate | | |
| Degree | | |
| Type of Contract at the Host Institution | | |
| Is this contract already active? | | |
| Is a salary being requested? | | |
| Managed by | | |
| Salary | | |
| | | |
| - | Total Direct Costs: | |
| | Indirect Costs: | |
| Total | Estimated Budget: | |
| Total I | Lotimated Budget. | |
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Reference Number Page 20 of 36





| Summary | | | |
|----------------------------------|--------|--------|-------|
| Category | Year 1 | Year 2 | Total |
| Equipment | € | € | € |
| IT Equipment | € | € | € |
| Materials, Supplies and Services | € | € | € |
| Personnel & Salaries | € | € | € |
| Travel | € | € | € |
| Other Expenses | € | € | € |
| Overheads | € | € | € |
| Total Direct Costs | € | € | € |
| Indirect Costs | € | € | € |
| Total Estimated Budget: | € | € | € |

Other Financial Support

| Granting Agency: Project Title: Status: Period From: Period To: Gross Amount: Currency: | |
|---|--|
| Brief Description: | |
| Specify overlaps with this application, if any: | |
| | |
| BIOSKETCH | |
| | |
| Name: | |
| Position Title: | |
| ID Research Platform: | |
| Personal Author ID: | |

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| E | mail: | | | |
|-----------------|--------------|------------------------|------------------|----------------|
| M | lobile: | | | |
| Education/Tra | ining | | | |
| Institution | Loca tion | Degree (if applicable) | Completi on date | Field of study |
| | | | | |
| | | | | |
| | | | | |
| | | | | |
| Personal State | ement | | | |
| | | | | |
| Positions, Scie | entific Ap | pointments, and | d Honors | |
| | | | | |
| Contributions | to Scienc | ee | | |
| | | | | |
| Relevant Publi | ications | | | |
| | | | | |
| | | | | |

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| Patents (whether relevant or not for the project) | |
|--|----------------|
| Companies (founder of or appointed by any start-up? Involved in industrial partnership?) | |
| Biosketch File | |
| Upload your Biosketch based on the template in Reference Documents on top of the application | |
| | |
| | Total Files: 1 |
| | |
| Reviewers | |

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Dissemination



| Suggested Reviewers | | | |
|--|--------|--|--|
| Title: First Name: Last Name: | | | |
| | | | |
| Expertise: | | | |
| Department: Institution: | | | |
| Email: Reason: | | | |
| Excluded Revi | iewers | | |
| Title: First Name: Last Name: | | | |
| | | | |
| Expertise: | | | |
| | | | |
| Department: Institution: Email: Reason: | | | |
| | | | |
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| | | | |
| Declarations | | | |
| | | | |
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| Actions, Activities and Results | |
|--|----------------------------------|
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| | |
| Notes | |
| | |
| Your Notes, if any | |
| | |
| | |
| Supporting Documents | |
| Please attach any supporting documentation (if any) | |
| r lease attach any supporting documentation (if any) | |
| | |
| | |
| | Total Files: 1 |
| | |
| Declarations | |
| Deciarations | |
| ✓ I hereby certify that all information submitted in the online application form is accurat | e and complete. |
| If I am awarded funding for this project, I will accept the conditions set by Fondazione partners, in case of Joint Calls Initiatives. | Telethon ETS and/or its Alliance |

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| Full Name: | |
|--|---|
| Place: | |
| Date: | |
| | |
| | |
| Attachments | |
| Partners deta | ils |
| | |
| Organization 2 - [| Department Aldo |
| | |
| Ethical Documents | |
| Human Subjects | |
| activate in due time all nece please refer to https://www | funding, funds WILL NOT BE PROVIDED until the pertinent Ethical Documentation has been obtained. Please essary procedures to obtain this approval in accordance with the relevant Italian laws. For clinical studies, v.aifa.gov.it/modulistica-sperimentazione-clinica. and/or its Alliance partners, in case of Joint Calls, Initiatives) reserves the right to ask for a copy of all the tation |
| Indicate whether the study | involves (consider both Applicant and collaborator(s) activities): |
| | |
| | |
| O | |

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| Please, specify the origin of the samples. If the study involves patient-derived samples, please include a summary of patients' details (e.g., age, sex, origin, genetics,) |
|--|
| |
| Vertebrate Animals |
| If the grant is approved for funding, funds WILL NOT BE PROVIDED until the pertinent Ethical Documentation has been obtained. Please activate in due time all necessary procedures to obtain this approval in accordance with the relevant Italian laws. Fondazione Telethon ETS (and/or its Alliance partners, in case of Joint Calls, Initiatives) reserves the right to ask for a copy of all the relevant approval documentation. |
| Indicate if the study involves the use of vertebrates or not. |
| OOOO |
| Host Institution |
| Applicant Details |
| Permanent position |
| Position Title |
| |
| Main Research Fields |
| |
| |
| |

Reference Number Page 27 of 36





| Name of Laboratory/Clinical Unit |
|--|
| |
| Number of staff members |
| Number of Staff Members |
| |
| Are you the Chief of the Laboratory/Clinical Unit? |
| |
| |
| Any other appointment (including foreign)? |
| |
| Oth are A managing to a supplemental and a suppleme |
| Other Appointment |
| |
| |
| Financial Interests Disclosure |
| |
| |
| |
| |
| |
| Facilities And Resources |
| |
| Laboratory space |
| |
| |
| Clinical Resources |
| |

Reference Number Page 28 of 36





| Office space | | | |
|------------------------------|-------------|-------|------|
| | | | |
| Major Equipment | | | |
| | | | |
| Core Facilities and Services | | | |
| | | | |
| | | | |
| Other | | | |
| | | | |
| | | | |
| | | | |
| Contacts | | | |
| | | | |
| Approver | | | |
| # Full Name | Institution | Email | Role |
| | | | |
| Collaborators | | | |

Reference Number Page 29 of 36





Use icon to invite collaborators. Once they have accepted the invite they will be able to see this application. To remove invited Collaborators, click on and click on icon displayed next to their names in the list. **Prefix First Name Last Name** Role **Email Status Active Collaborators** Table below displays all Active collaborators added on the application. 1 **Full Name:** Institution: Email: Role: Laboratory: Contribution: **Budget Principal Investigator:: Application Number: Institution Name: Project Title:** Year 1 **IT Equipment** Item Description Managed by Cost

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Materials, Supplies and Services



| Materials, Supplies and Services | | | |
|--|-------------|---------------------|------|
| Item | Description | Managed by | Cost |
| | | | |
| | | | |
| | | | |
| Travel | | | |
| Item | Description | Managed by | Cost |
| | | | |
| | | | |
| | | l | |
| Other Expenses | 1 | | |
| Item | Description | Managed by | Cost |
| | | | |
| | | | |
| Overheads | | | |
| Item | Description | Managad by | Cost |
| nem | Description | Managed by | Cost |
| | | | |
| | | , | |
| Personnel & Salaries | | | |
| Role on Project | | | |
| Annual effort % | | | |
| Name | | | |
| Surname | | | |
| Birthdate | | | |
| Degree | | | |
| Type of Contract at the Host Institution | 1 | | |
| Is this contract already active? | | | |
| Is a salary being requested? | | | |
| Managed by | | | |
| Salary | | | |
| | | | |
| | | Total Direct Costs: | |
| | | Indirect Costs: | |
| Total Estimated Budget: | | | |
| Year 2 | | 3 1 | |

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| Item | Description | Managed by | Cost |
|--|-------------|----------------------|------|
| | | | |
| | | | |
| Travel | | | |
| Item | Description | Managad by | Cost |
| nem | Description | Managed by | Cost |
| | | | |
| | | | |
| | | | |
| Other Expenses | | | |
| Item | Description | Managed by | Cost |
| , | | | |
| | | | |
| Overheads | | | |
| Item | Description | Managed by | Cost |
| | | | |
| | | | |
| Personnel & Salaries | | | |
| Role on Project | | | |
| Annual effort % | | | |
| Name | | | |
| Surname | | | |
| Birthdate | | | |
| Degree | | | |
| Type of Contract at the Host Institution | | | |
| Is this contract already active? | | | |
| Is a salary being requested? | | | |
| Managed by | | | |
| Salary | | | |
| | | | |
| Total Direct Costs: | | | |
| | | Indirect Costs: | |
| | Tot | al Estimated Budget: | |
| | | | |

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ID Research Platform:



| Summary | | | |
|----------------------------------|--------|--------|-------|
| Category | Year 1 | Year 2 | Total |
| Equipment | € | € | € |
| IT Equipment | € | € | € |
| Materials, Supplies and Services | € | € | € |
| Personnel & Salaries | € | € | € |
| Travel | € | € | € |
| Other Expenses | € | € | € |
| Overheads | € | € | € |
| Total Direct Costs | € | € | € |
| Indirect Costs | € | € | € |
| Total Estimated Budget: | € | € | € |
| her Financial Support | | | |
| OSKETCH | | | |
| Name: | | | |
| Position Title: | | | |

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| Personal A | uthor ID: | | | |
|---------------------|--------------------|--------------------------------------|-----------------------|----------------|
| Ema | il: | | | |
| Mobi | ile: | | | |
| Education/Training | g | | | |
| Institution | Location | Degree (if applicable) | Completion date | Field of study |
| | | | | |
| Personal Statemen | nt | | | |
| | | | | |
| Positions, Scientif | ic Appointments | s, and Honors | | |
| Contributions to S | cience | | | |
| Relevant Publicati | ons | | | |
| Patents (whether r | relevant or not fo | or the project) | | |
| Companies (found | ler of or appoint | ed by any start-up? Involved in indu | ıstrial partnership?) | |

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Biosketch File

| Upload your Biosketch based on the template in Reference Documents on top of the application | |
|--|----------------|
| | |
| | Total Files: 1 |
| | |
| Declarations | |
| Notes | |
| Your Notes, if any | |
| | |
| | |
| | |
| Supporting Documents | |
| Please attach any supporting documentation (if any) | |
| | |
| Declarations | |
| | |

Reference Number Page 35 of 36

✓ I hereby certify that all information submitted in the online application form is accurate and complete.





| V | If I am awarded funding for this project, I will accept the conditions set by Fondazione Telethon ETS and/or its Alliance partners, in case of Joint Calls Initiatives. | | | |
|----------|---|--|--|--|
| | Full Name: | | | |
| | Place: | | | |
| | Date: | | | |
| | _ | | | |

Attachments

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