

TRAINING AND EMPOWERING PATIENTS ON SCIENTIFIC RESEARCH: THE EXAMPLE OF FONDAZIONE TELETHON

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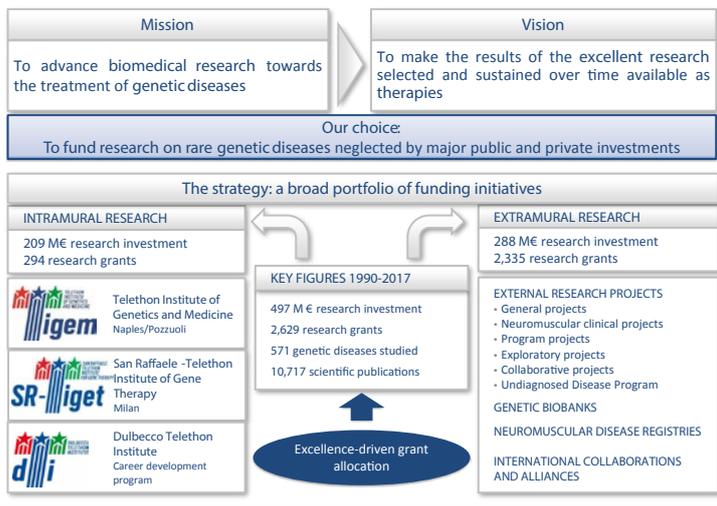
ABSTRACT

Fondazione Telethon (FT) is a main Italian charity funding biomedical research towards the cure of rare genetic diseases (www.telethon.it). It relies on donations from general public through major fundraising events (such as the annual television marathon) and various fundraising initiatives held throughout the year. Accountable fund management and careful expense monitoring are performed to fulfill FT promise to patients and donors who continue to support its mission and all its funding decisions are made by an international scientific committee through a peer review-based selection system.

Telethon-funded research, tackling the challenge of genetic diseases from diagnosis to basic and clinical science, is constantly moving forward towards the development of therapies for an increasing number of pathologies including metabolic, hematological and neuromuscular disorders. After 28 years of activity, its results are globally acknowledged and FT has placed itself among the main players in the world in the field of biomedical research for rare genetic diseases (EMBO Mol Med: 2017, 9 (3), 289-292). The Telethon research portfolio includes intramural research led by the three Telethon Institutes and extramural research, funded through several funding programs. FT participates in several national and international collaborations and alliances, such as the European RD-Connect project, the International Rare Diseases Research Consortium (IRDIRC), the European NeuroMuscular Centre (ENMC) Consortium and coordinates the EuroBioBank Network. Moreover, FT joined Europe PubMed Central as a funder to offer open access to the results of the Telethon-funded research.

The close relationship with Patient Organizations (POs) is at the basis of the Telethon strategic plan and has led to the implementation of several initiatives: the Telethon-Uildm Call for clinical neuromuscular research; the Telethon Network of Genetic Biobanks (TNGB, <http://biobanknetwork.telethon.it/>); the Call for Exploratory Projects and the Undiagnosed Disease Program. FT has set up activities dedicated to people living with a rare disease (RD), such as the Helpline Info_Rare to support patients and provide information on biomedical research and the network of the POs Friends of Telethon. FT has also promoted patient empowerment on research topics through the organization of several training meetings, which are described more in detail below.

1. FONDAZIONE TELETHON PORTFOLIO



2. FONDAZIONE TELETHON AND THE PATIENT ORGANIZATIONS

Born thanks to the will of patients and their families, FT has been working since its foundation with the leading POs of people with RD. Through the years, FT has set an informal network of 200 POs called Friends of Telethon, with the aim of collaborating to speed up research towards the cure of genetic diseases, while simultaneously raising attention to the needs of people living with a RD. The network is not a federation, but it allows POs to have a direct and continuous communication channel with the Foundation, to be included in its training events, to request its patronage for their scientific events, to receive advice on scientific research policies, to give visibility to their associative meetings on the FT on-line Calendar. Furthermore, FT has a direct collaboration with Uniamo, the Italian National Alliance of Italian Patient Organizations.

Main goals

- Listen to the needs of people living with a RD
- Create alliance with all the stakeholders in order to speed up the progress of research on genetic diseases
- Raise awareness on RD and on the need of financing research
- Empower people living with a RD in order to promote their participation in research
- Foster contacts and sharing of experience among people with similar conditions

Main activities

- INFORMATION on FT scientific research and progresses towards therapies
- TRAINING on research topics
- NETWORKING AND ALLIANCES
- RAISING AWARENESS
- SCIENTIFIC ADVISE



3. TRAINING FOR PATIENT ORGANIZATIONS

FT is promoting a participatory, innovative and responsible scientific research through the active participation of all stakeholders. This means also engaging people living with a RD in the process, as suggested by European Institutions that encourage to work together in "areas of unmet medical need, sharing input on clinical trial design, advising researchers when starting new programmes and participation in the safety assessment of new medicines" (www.eupati.eu). For this reason, since 2009 FT organizes regular annual meetings with POs representatives who are interested in a better understanding of policies and logics of scientific research. As "expert patients" they can gain the confidence and knowledge needed to bring their expertise and contribution to the working groups, including policy makers, clinicians, scientists, HTA bodies, industry and regulatory authorities.

Type of Audience at the training meetings



5. THE FEEDBACKS FROM POs

An extraordinary opportunity to meet other Patient Organizations and broaden the horizons of research AIBWS

You never take for granted what you can take home from a Telethon convention: every time you meet new friends, find a hope that doesn't let you down, a new determination, a goal to achieve once back home PKS Association

A great day for sharing knowledge and to meet again among friends... Asitai

We were so many: researchers, clinicians and patients, all together to learn, share and meet once again among friends Pten Italia

Registries, undiagnosed diseases, ELSI, informed consent: keywords that are useful tools for the patient organisations!

4. THE MAIN TRAINING EVENTS

The Telethon Friends Conference



A biennial event for POs is organized in conjunction with the main Telethon Scientific Convention, which conveys all the principal investigators with active Telethon projects and their staff. The event is attended by almost 1000 people (scientists, POs, volunteers, exhibitors and FT staff). The Telethon Friends Conference is dedicated to train patients on scientific research topics such as:

- Preclinical research and clinical trials
- Biobanks
- Registries and epidemiological data.

POs and researchers meet in a plenary session and also at the poster sessions where scientists present their projects and results. This is a unique opportunity for the patients' community to be in touch with researchers. Moreover it is a great possibility for young scientists to fully understand FT mission and the relevance to patients of their work.

The Spring meeting



Every other year, alternate with the Telethon Friends Conference, it takes place a Spring meeting where the POs share their best practices on RD. The meeting provides an inspiring and useful survival kit for the POs. Networking is another major goal of the Conference.

Science Dialogues



A single event in collaboration with the European Society of Gene and Cell Therapy (ESGCT) took place in 2016. "A one-day interactive event that provided an opportunity to discuss and debate cutting edge research on stem cells and gene therapy with scientists, patients, researchers, clinicians and to think about the impact that this research has on patients and on society" (www.esgct.eu)

CONCLUSIONS

POs acknowledge these training meetings as a great opportunity to meet with professionals of research. In addition they have the chance to interact among POs and share their best practices and success experience (but also failure) in order to learn from each other with a peer to peer approach. All this knowledge and contribution means an extraordinary value for all the RD community who wants to be involved in all the process that concern them. By receiving training, patients who are already experts on their diseases become advocates providing a valuable contribution to shape the RD health and social policy, foster research on RD, participate in clinical trial design and orphan drugs assessment. The active engagement of POs in research is not always reality yet, but it's an ongoing process that should be encouraged by all the stakeholders. This implies a cultural change, made of respectful communication, independency of all partners, society acceptance, support and pro-active role of all stakeholders, especially patients, care-givers and parents.