Resource Summary Report

Generated by NIF on May 8, 2025

Telethon Foundation

RRID:SCR_003803
Type: Tool

Proper Citation

Telethon Foundation (RRID:SCR_003803)

Resource Information

URL: http://www.telethon.it/en

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Description: Since 1990 Telethon, along with millions of Italians, has stepped up to the challenge of beating muscular dystrophy and the other genetic diseases. It is a marathon against time, because there are many people who live with these rare disorders, and the resources to deal with them have to be carefully measured out because there is not much public or private funding invested in this field of research, and the path to finding cures is often long and tortuous. The foundation In order to guarantee as much research funding as possible into muscular dystrophy and other genetic diseases, the Telethon team works throughout the year and has adopted a management system for the donated funds that is strict and efficient. For every euro raised by Telethon, about eighty euro cents reach the cutting edge laboratories and excellent research centers. Scientific area The selection of the best research projects, the funding of dedicated researchers and the foundation and maintenance of its research institutes make Telethon a point of Italian excellence in the world. Along with recognition from the international scientific community, Telethon's world of research is the biggest ally of all the people who live with muscular dystrophy or other genetic disorders every day. The online database provides complete information about the projects funded by Telethon from 1991 to the present. The archive contains information about all the Foundation's efforts in the field of biomedical research. In addition to a search by disease, it is possible, using the advanced search function, to interrogate the database by groups of disorders, by the name of a researcher or research institute, or by the town, province, or region where projects are based. The use of another search filter makes it possible to check which research projects are ongoing and which have come to an end.

Abbreviations: Telethon

Synonyms: TeleThon.it

Resource Type: portal, database, funding resource, data or information resource, disease-related portal, topical portal

Funding:

Resource Name: Telethon Foundation

Resource ID: SCR_003803

Alternate IDs: nlx_143533

Record Creation Time: 20220129T080221+0000

Record Last Update: 20250508T064847+0000

Ratings and Alerts

No rating or validation information has been found for Telethon Foundation.

No alerts have been found for Telethon Foundation.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 25 mentions in open access literature.

Listed below are recent publications. The full list is available at NIF.

Vavassori S, et al. (2024) Unlocking the full potential of rare disease drug development: exploring the not-for-profit sector's contributions to drug development and access. Frontiers in pharmacology, 15, 1441807.

Boycott KM, et al. (2019) International collaborative actions and transparency to understand, diagnose, and develop therapies for rare diseases. EMBO molecular medicine, 11(5).

Takahashi H, et al. (2018) Identification of functional features of synthetic SINEUPs, antisense lncRNAs that specifically enhance protein translation. PloS one, 13(2), e0183229.

Salinas PC, et al. (2017) Gender Equality from a European Perspective: Myth and Reality. Neuron, 96(4), 721.

Sicca F, et al. (2016) Gain-of-function defects of astrocytic Kir4.1 channels in children with autism spectrum disorders and epilepsy. Scientific reports, 6, 34325.

Wilson IJ, et al. (2016) Mitochondrial DNA sequence characteristics modulate the size of the genetic bottleneck. Human molecular genetics, 25(5), 1031.

Tosco A, et al. (2016) A novel treatment of cystic fibrosis acting on-target: cysteamine plus epigallocatechin gallate for the autophagy-dependent rescue of class II-mutated CFTR. Cell death and differentiation, 23(8), 1380.

Valvo G, et al. (2016) Temporal lobe connects regression and macrocephaly to autism spectrum disorders. European child & adolescent psychiatry, 25(4), 421.

Grone BP, et al. (2016) Epilepsy, Behavioral Abnormalities, and Physiological Comorbidities in Syntaxin-Binding Protein 1 (STXBP1) Mutant Zebrafish. PloS one, 11(3), e0151148.

Pellacani S, et al. (2016) The Revolution in Migraine Genetics: From Aching Channels Disorders to a Next-Generation Medicine. Frontiers in cellular neuroscience, 10, 156.

Decostre V, et al. (2015) Wrist flexion and extension torques measured by highly sensitive dynamometer in healthy subjects from 5 to 80 years. BMC musculoskeletal disorders, 16(1), 4.

Finotti A, et al. (2015) Recent trends in the gene therapy of ?-thalassemia. Journal of blood medicine, 6, 69.

Monaco L, et al. (2014) The challenge for a European network of biobanks for rare diseases taken up by RD-Connect. Pathobiology : journal of immunopathology, molecular and cellular biology, 81(5-6), 231.

Marchese M, et al. (2014) Autism-epilepsy phenotype with macrocephaly suggests PTEN, but not GLIALCAM, genetic screening. BMC medical genetics, 15, 26.

Buniello A, et al. (2013) Headbobber: a combined morphogenetic and cochleosaccular mouse model to study 10qter deletions in human deafness. PloS one, 8(2), e56274.

Valvo G, et al. (2013) Somatic overgrowth predisposes to seizures in autism spectrum disorders. PloS one, 8(9), e75015.

Pistoni M, et al. (2013) Rbfox1 downregulation and altered calpain 3 splicing by FRG1 in a mouse model of Facioscapulohumeral muscular dystrophy (FSHD). PLoS genetics, 9(1), e1003186.

D'Angelo A, et al. (2012) Ofd1 controls dorso-ventral patterning and axoneme elongation during embryonic brain development. PloS one, 7(12), e52937.

Cabianca DS, et al. (2012) A long ncRNA links copy number variation to a polycomb/trithorax epigenetic switch in FSHD muscular dystrophy. Cell, 149(4), 819.

Lacoux C, et al. (2012) BC1-FMRP interaction is modulated by 2'-O-methylation: RNA-binding activity of the tudor domain and translational regulation at synapses. Nucleic acids research, 40(9), 4086.