Resource Summary Report

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Telethon Network of Genetic Biobanks

RRID:SCR_004658
Type: Tool

Proper Citation

Telethon Network of Genetic Biobanks (RRID:SCR_004658)

Resource Information

URL: http://biobanknetwork.telethon.it/

Proper Citation: Telethon Network of Genetic Biobanks (RRID:SCR_004658)

Description: Network of non profit association of Italian repositories to form catalogue of biospecimens and associated data. Used to collect, process, preserve and distribute biological samples and related clinical data from individuals affected by rare diseases, their relatives or from healthy control individuals, with standards complying with Italian laws and international recommendations. You may browse sample catalogue by diagnosis or use advanced search option. Request for samples is granted only if project is in agreement with TNGB mission and after receiving signed material transfer agreement form.

Abbreviations: TNGB

Synonyms: Telethon Network of Genetic Biobanks

Resource Type: biomaterial supply resource, material resource

Defining Citation: PMID:24004821

Keywords: Italian, repository, catalogue, biospeciment, data, collect, process, preserve, distribute, rare, disease

Funding: Telethon Foundation ;
Italian Health Department

Availability: Restricted

Resource Name: Telethon Network of Genetic Biobanks

Resource ID: SCR_004658

Alternate IDs: nlx_65989

Record Creation Time: 20220129T080225+0000

Record Last Update: 20250422T055159+0000

Ratings and Alerts

No rating or validation information has been found for Telethon Network of Genetic Biobanks.

No alerts have been found for Telethon Network of Genetic Biobanks.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 28 mentions in open access literature.

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

Hop PJ, et al. (2024) Systematic rare variant analyses identify RAB32 as a susceptibility gene for familial Parkinson's disease. Nature genetics, 56(7), 1371.

Cavestro C, et al. (2024) Emerging variants, unique phenotypes, and transcriptomic signatures: an integrated study of COASY-associated diseases. Annals of clinical and translational neurology, 11(6), 1615.

Mao K, et al. (2023) FOXI3 pathogenic variants cause one form of craniofacial microsomia. Nature communications, 14(1), 2026.

Croci S, et al. (2022) The polymorphism L412F in TLR3 inhibits autophagy and is a marker of severe COVID-19 in males. Autophagy, 18(7), 1662.

Azevedo C, et al. (2022) Parkinson's disease and multiple system atrophy patient iPSC-derived oligodendrocytes exhibit alpha-synuclein-induced changes in maturation and immune reactive properties. Proceedings of the National Academy of Sciences of the United States of America, 119(12), e2111405119.

Straniero L, et al. (2022) Role of Lysosomal Gene Variants in Modulating GBA-Associated Parkinson's Disease Risk. Movement disorders : official journal of the Movement Disorder Society, 37(6), 1202.

Gialluisi A, et al. (2021) Identification of sixteen novel candidate genes for late onset Parkinson's disease. Molecular neurodegeneration, 16(1), 35.

Palombo F, et al. (2021) The relevance of mitochondrial DNA variants fluctuation during reprogramming and neuronal differentiation of human iPSCs. Stem cell reports, 16(8), 1953.

Russ K, et al. (2021) TNF-? and ?-synuclein fibrils differently regulate human astrocyte immune reactivity and impair mitochondrial respiration. Cell reports, 34(12), 108895.

Maini I, et al. (2021) Clinical Manifestations in a Girl with NAA10-Related Syndrome and Genotype-Phenotype Correlation in Females. Genes, 12(6).

Cavestro C, et al. (2021) Novel deep intronic mutation in PLA2G6 causing early-onset Parkinson's disease with brain iron accumulation through pseudo-exon activation. Neurogenetics, 22(4), 347.

Yau WY, et al. (2021) Low Prevalence of NOTCH2NLC GGC Repeat Expansion in White Patients with Movement Disorders. Movement disorders : official journal of the Movement Disorder Society, 36(1), 251.

Mazzetti S, et al. (2020) ?-Synuclein oligomers in skin biopsy of idiopathic and monozygotic twin patients with Parkinson's disease. Brain : a journal of neurology, 143(3), 920.

Fiacco E, et al. (2020) Derivation of two naturally isogenic iPSC lines (KAUSTi006-A and KAUSTi006-B) from a mosaic Klinefelter Syndrome patient (47-XXY/46-XY). Stem cell research, 49, 102049.

Simmnacher K, et al. (2020) Unique signatures of stress-induced senescent human astrocytes. Experimental neurology, 334, 113466.

Brazdis RM, et al. (2020) Demonstration of brain region-specific neuronal vulnerability in human iPSC-based model of familial Parkinson's disease. Human molecular genetics, 29(7), 1180.

Straniero L, et al. (2020) The SPID-GBA study: Sex distribution, Penetrance, Incidence, and Dementia in GBA-PD. Neurology. Genetics, 6(6), e523.

Frasca A, et al. (2020) MECP2 mutations affect ciliogenesis: a novel perspective for Rett syndrome and related disorders. EMBO molecular medicine, 12(6), e10270.

Gialluisi A, et al. (2019) Whole Exome Sequencing Study of Parkinson Disease and Related Endophenotypes in the Italian Population. Frontiers in neurology, 10, 1362.

Malerba N, et al. (2019) Generation of the induced human pluripotent stem cell lines CSSi009-A from a patient with a GNB5 pathogenic variant, and CSSi010-A from a

CRISPR/Cas9 engineered GNB5 knock-out human cell line. Stem cell research, 40, 101547.