Resource Summary Report

Generated by NIF on May 25, 2025

Italian Rett Syndrome database

RRID:SCR_002000
Type: Tool

Proper Citation

Italian Rett Syndrome database (RRID:SCR_002000)

Resource Information

URL: http://www.biobank.unisi.it/Elencorett.asp

Proper Citation: Italian Rett Syndrome database (RRID:SCR_002000)

Description: Data and biospecimen from Rett Syndrome patients shared with the scientific community with the ability to visualize the list of available samples and select those with specific clinical and molecular features. It also contains information on biospecimen samples from x-linked retardation, microdeletion, duplication syndromes, autosomal MR, and retinoblastoma. The bank is active since 1998 and it is located in the Medical Genetics Unit, at the University Hospital of Siena. The bank is divided in three distinct sections: # Rett Syndrome. This section contains samples from patients affected by Rett syndrome, a neurodegenerative disease affecting almost exclusively girls with an estimated frequency of 1:10000-15000 live born. By accessing the section users can see a list of all patients available with their phenotype, the specific MECP2 or CDKL5 mutation if known and the kind of biological samples available for each patient. The availability of this large panel of patients is potentially important for the clarification of the molecular bases of Rett syndrome. In fact, a 20-30 of Rett cases do not have MECP2 or CDKL5 mutations. These patients might bear intronic/promoter MECP2 or CDKL5 mutations or they might have alterations in one or more genes different from MECP2 or CDKL5, as suggested by the identification of various chromosomal rearrangements. To confirm a causative role of these rearrangements, and to identify the relevant gene/s, it is important to collect a great number of patients in which to search for overlapping rearrangements or point mutations in candidate genes. # X-Linked Mental Retardation. This section contains samples collected by the centers belonging to the Italian network on X-linked mental retardation, which includes the laboratory of bank curators (for specific information on the network goals and organization, go to the section page). Mental retardation (MR) is the most frequent cause of serious handicap in humans with an estimated prevalence of 0,3-0,5 for moderate to severe MR (IQ

Abbreviations: Rett syndrome bank

Resource Type: material resource, biomaterial supply resource

Keywords: duplication syndrome, autosomal mr, microdeletion, retinoblastoma, mecp2, cdkl5, foxg1, clinical, mutation, phenotype, lymphoblastoid cell line, leukocyte, dna, plasma, blood, biomaterial manufacture

Related Condition: Rett Syndrome, Duplication syndrome, Autosomal MR, Microdeletion, Retinoblastoma, X-linked retardation

Funding: Telethon Foundation

Availability: Acknowledgement requested, Qualified investigators, Non-commercial

Resource Name: Italian Rett Syndrome database

Resource ID: SCR_002000

Alternate IDs: nif-0000-12492

Alternate URLs: http://www.biobank.unisi.it/ScegliArchivio.asp

Record Creation Time: 20220129T080210+0000

Record Last Update: 20250525T032434+0000

Ratings and Alerts

No rating or validation information has been found for Italian Rett Syndrome database.

No alerts have been found for Italian Rett Syndrome database.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 1 mentions in open access literature.

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

Patriarchi T, et al. (2016) Imbalance of excitatory/inhibitory synaptic protein expression in iPSC-derived neurons from FOXG1(+/-) patients and in foxg1(+/-) mice. European journal of human genetics : EJHG, 24(6), 871.