

Resource Summary Report

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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>

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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 (IQ50) which increases to 1-1,5 when mild MR (IQ 50-70) is included. It is calculated that about 20-25 of mentally retarded males have a mutation in a gene on the X chromosome (X-linked mental retardation). X-linked mental retardation is a genetically heterogeneous condition. This is particularly true for the non-syndromic form (MRX), where MR is the only consistent clinical finding and no distinctive features between

patients exist. In this situation the only possibility to group patients from different families is represented by linkage analysis, which needs the availability of large families. However, families linked to the same region demonstrate different causative genes. In these conditions, the number of patients available for analysis is a discriminating factor since a large number of patients need to be tested in order to fully confirm or exclude the involvement of a gene in MRX. # Other. This section of the bank contains biological materials and clinical data of patients with other genetic disorders (different from Rett and X-linked mental retardation). Part of this section is dedicated to Alport syndrome. Services: * Isolation of leukocytes from human peripheral blood samples * Establishment of EBV transformed lymphoblastoid cell lines from human peripheral blood leukocytes. * DNA extraction. * Plasma isolation. * Storage: ** Cryo-preservation of transformed cell lines and primary leukocytes at 135??C ** Storage of DNA at 20 degrees C ** Storage of plasma at 20 degrees C * Distribution of the stored biological samples.

Abbreviations: Rett syndrome bank

Resource Type: biomaterial supply resource, material 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: THIS RESOURCE IS NO LONGER IN SERVICE

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: 20260801T191044+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 [ASWG](#) .

Patriarchi T, et al. (2016) Imbalance of excitatory/inhibitory synaptic protein expression in iPSC-derived neurons from FOXP1(+/-) patients and in foxg1(+/-) mice. European journal of human genetics : EJHG, 24(6), 871.