

Resource Summary Report

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Barth Syndrome Biorepository

RRID:SCR_004775

Type: Tool

Proper Citation

Barth Syndrome Biorepository (RRID:SCR_004775)

Resource Information

URL: <http://www.barthsyndrome.org/english/View.asp?x=1513>

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Description: The Barth Syndrome Registry and Repository (BRR) at the University of Florida and Children's Hospital Boston was created to provide additional information about this rare disorder. By collecting information directly from families and abstracting medical information from patient records, we hope to build a large anonymized database that is useful for answering scientific research questions and that is a source of medical information. Additionally, we are collecting blood samples for DNA isolation and storage and for the development of special cell lines. These tools will allow researchers to better understand the cellular and genetic nature of this disease and how these abnormalities may impact the clinical variability of Barth syndrome patients. Currently available data from the Barth Syndrome Registry and Repository includes self-reported data. This includes basic demographics, age at diagnosis of Barth Syndrome, reason for diagnosis, family history; symptoms, presence or absence of a history of cardiomyopathy, history regarding hospitalizations, cardiac transplant or listing for transplant, presence or absence of a history of neutropenia; history of infections, medical therapy and basic developmental history. We are in the process of abstracting medical data to build a more complete medical database and may be able to provide additional data with a specific request. Currently the DNA bank has data from 65 patients, genomic DNA from 50 patients, lymphoblast lines from 41 patients, fibroblast lines from 3 patients, and myocardium/autopsy from 3 patients. However, this is increasing as new patients enroll in the Registry. Please contact us with questions and specific requests about data or specimens.

Abbreviations: BRR

Synonyms: Barth Syndrome Registry and Repository

Resource Type: biomaterial supply resource, tissue bank, material resource

Related Condition: Barth syndrome

Resource Name: Barth Syndrome Biorepository

Resource ID: SCR_004775

Alternate IDs: nlx_77178

Record Creation Time: 20220129T080226+0000

Record Last Update: 20260801T042557+0000

Ratings and Alerts

No rating or validation information has been found for Barth Syndrome Biorepository.

No alerts have been found for Barth Syndrome Biorepository.

Data and Source Information

Source: [SciCrunch Registry](#)

Usage and Citation Metrics

We have not found any literature mentions for this resource.