

Shwachman-Diamond Syndrome

...and the patient perspective on
biobanking and registries

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What is SDS?

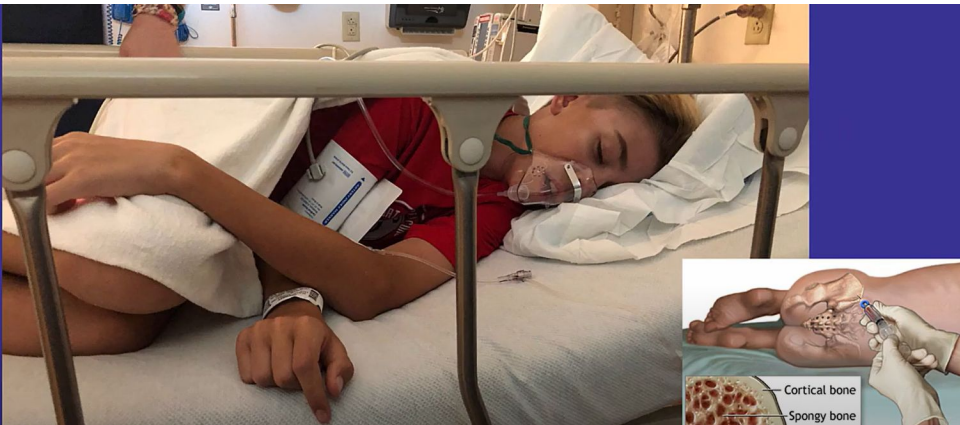
A Congenital Neutropenia

Rare **genetic disorder** affecting protein production in every cell of the body.

E.g. problems digesting food, compromised immune system...

SDS causes a **very high risk** of developing **leukemia** (blood cancer):

- Nearly 100% fatal in SDS patients
- Frequent blood draws and bone marrow biopsies
- Constant fear of leukemia



Why are we here?

We want to give our kids the opportunity to live a full life — birthdays, graduation, weddings, and even starting a family — without fear of leukemia.



Your work gives us hope.

SDS Alliance: Driving SDS Research

- Founded in 2020 by biotech professionals who are also parents of a child with SDS.
- **Mission: Finding a cure for SDS**
- First goal: Develop therapies that prevent leukemia.
- Give patients and families the opportunity to live without the fear of leukemia.

Patients want their data and samples to have maximal impact and benefit.

What data and resources are missing? ⇨ Create it!

How can we maximize the impact of the existing resources?

1. Create Patient Reported Outcomes / Survey Platform

Coming soon.

- Platform that meets **regulatory requirements / privacy** (FDA, EMEA, GDPR, etc.)
- **Patient reported** data, but **structured**, using established data dictionaries, etc.
Based on ClinGen and the [Common Data Elements developed by the European Commission](#).
- **Validated surveys**...to inform the development of outcome measures, quality of life, patient preference, impact, etc.
- Patient will be **consented to allow re-contact and sharing**.
- Let's discuss GUIDs and Pseudonymization tools.

Goal: Collaboration and sharing!

2. Create Biobank of Renewable Samples

We are building a collection of patient cell lines (LCLs, Fibroblasts, and iPSCs) that can be

- easily ordered from a catalogue
- cost effective
- high quality
- comes with rich data
- anonymous



The Coriell Institute and the NIGMS Human Genetic Cell Repository



- **Non-profit** organization dedicated to advance biomedical research, facilitate scientific collaboration, and work towards the diagnosis, treatment, and prevention of human genetic diseases
- Renowned institution generating, storing, and distributing world-class biomaterials: well-characterized, high-quality cell lines and DNA since 1972
- Centralized, accessible scientific resource for researchers to both submit samples to make available to the scientific community and utilize specimens to conduct research
- NIGMS Human Genetic Cell Repository
 - Houses libraries of catalogued biospecimens, with associated clinical data
 - Diverse collections of disease-specific and human variation samples
 - Support biomedical research into: genetic investigation and genomic exploration, assay development and validation, iPSC generation, proficiency testing, therapy development
 - Partners with the rare disease community



Get Involved with the NIGMS Repository



- Submit samples (blood draw, skin biopsy, cell culture)*
 - For individuals affected with rare disease
 - For clinicians working with rare disease-affected patients
 - *includes “back-to-submitter” sample free-of-charge upon successful submission
- Perform research on biospecimens on catalog-available materials (cell lines, DNA products)
 - For scientists affiliated with research institution (academic/non-profit, commercial)
- Collaborate to characterize reference materials
 - For researchers able to provide molecular and biochemical characterization of presently uncharacterized materials (provided free-of-charge)
- Serve on Scientific Advisory Committee overseeing Repository operations
 - For experts in a diverse range of relevant biomedical fields
 - E.g. molecular biology, clinical/population genetics, cytogenetics, pharmacogenetics, stem cell, bioethics, and medicine

Submitting to the NIGMS Repository

- Affected individual must be **diagnosed with a heritable genetic disease or chromosomal aberration**
 - Unaffected family members (e.g. parents / siblings) are also eligible
 - Must have clinical documentation of the disorder (e.g. medical records, genetic test results, physician summaries)
- Sample types accepted
 - Whole blood - for **lymphoblastoid cell lines**, iPSCs, and DNA products
 - Skin biopsy - for **fibroblasts**, iPSCs, and DNA products
 - Cell cultures (early passages of previously established cell lines)
- Submission process
 - Email NIGMS@coriell.org and a team member will assist to provide a donation kit free-of-charge
 - Complete submission forms (informed consent, clinical data summary, submission form)
 - Draw blood/collect biopsy, and return sample with paperwork to Coriell
- Back-to-submitter rights
 - Submitters are eligible to receive one cell line (excluding iPSCs) or DNA product back-in-kind for each sample donated that is successfully included in the NIGMS Repository catalog



Easy to search and order!

www.coriell.org

1. Type: **Shwachman**, for example, then click Enter.

The screenshot shows the Coriell Institute website with a search bar at the top. Below the search bar, the results for 'shwachman' are displayed. A purple arrow points to the search bar. At the bottom of the results, a table lists catalog items. A purple arrow points to the first item, GM28311.

ID	Description	Affected	Product	Source	Gene	Mutations	Sex	Age at Samp
GM28311	SHWACHMAN-DIAMOND SYNDROME ...	Yes	LCL	B-Lymphocyte	CUBN	SBDS	c.1719A>T (p.L...	Female 7 YR

2. Select the result of interest.

The screenshot shows the product page for GM28311, LCL from B-Lymphocyte. A purple arrow points to the 'Add to Cart' button. The page contains detailed information about the cell line, including its description, affected status, sex, age, and various characterization data.

Repository	NIGMS Human Genetic Cell Repository
Subcollection	Heritable Diseases PIGI Consented Sample
Biopsy Source	Peripheral vein
Cell Type	B-Lymphocyte
Tissue Type	Blood
Transformant	Epstein-Barr Virus
Race	More than one race
Hispanic Ethnicity	Not Hispanic/Latino
Ethnicity	White and Asian; Hungarian, Ashkenazi, Sephardic
Country of Origin	USA
Family Member	1
Family History	N
Relation to Proband	proband
Confirmation	Molecular characterization before cell line submission to CCR
Species	Homo sapiens
Common Name	Human
Remarks	See "Phenotypic Data" tab

3. Review the rich data.

4. Add to cart to order.

3. Maximize the Impact of Existing Resources

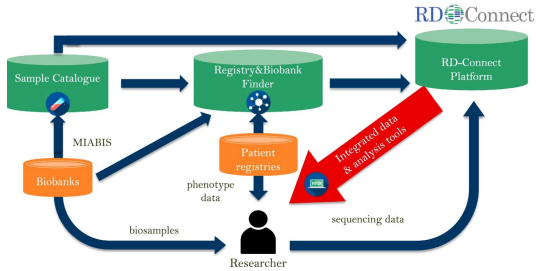
- 1 North American SDS Registry (160/240)
- 2 NIH IBMFS Cohort Study (5)
- 3 Severe Chronic Neutropenia International Registry (SCNIR)
- 4 Canadian Inherited Marrow Failure Registry (CIMFR)
- 5 Italian SDS registry (121 SBDS)
- 6 French congenital neutropenia registry
- 7 Severe Chronic Neutropenia International Registry (SCNIR) - Europe
- 8 UK SDS registry (coming soon)
- 9 Greek biobank
- 10 BMF Registry Australia



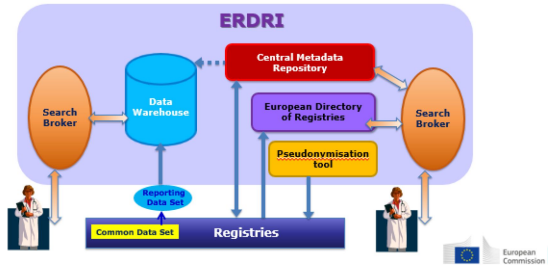
Studies vs. Resources

Tools to Address: Technical (and Regulatory) challenges

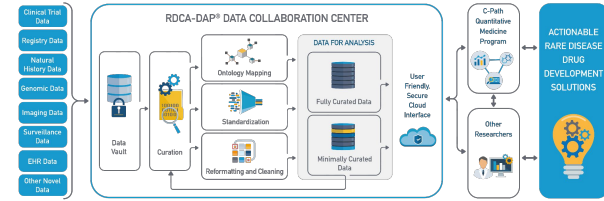
- RD-Connect



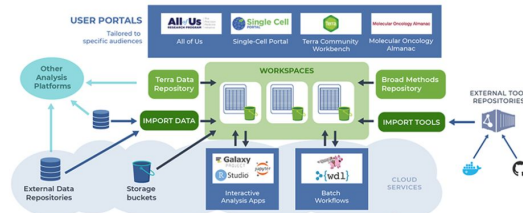
- European Platform for Rare Disease Registration



- RDCA-DAP (C-Path)



- Terra



Let's work together!

Let's discuss

- common data elements
- pseudonymization tools and GUIDs
- prioritizing (non-renewable) patient samples

The screenshot displays the ERDRI.dor website, which is part of the European Commission's EU Science Hub. The page is titled "ERDRI.dor - European Directory of Registries" and features a search bar at the top right. Below the search bar, there is a navigation menu with links to Home, Search, and Help. The main content area is divided into two sections: "Search" and "Search results".

Search Section:

- Name or Subject:** A text input field.
- Responsible:** A text input field.
- Rare disease:** A text input field.
- ICD-10 code:** A text input field.
- Country:** A dropdown menu.
- Year of the recruitment:** A text input field.
- Type:** A list of checkboxes for different types of registries: Epidemiology, Clinical, Basic Research, Pharmacological Research, Patient, Healthcare planning, Economic evaluation, and Has a biobank.
- Buttons:** "Search" and "Reset" buttons.

Search results Section:

- Page 1 of 1, (3 entries found):** A pagination bar.
- Name:** A table with 3 columns: Name, ESID, and a button icon.
- Search results:** A table with 3 rows of search results.
- Page 1 of 1, (3 entries found):** A pagination bar.

Name	ESID	
Severe Chronic Neutropenia International Registry		
Spanish Rare Diseases Research Patient Registry		

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Thank you!