Shwachman-Diamond Syndrome

…and the patient perspective on biobanking and registries

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SHWACHMAN-DIAMOND SYNDROME ALLIANCE

What is SDS?

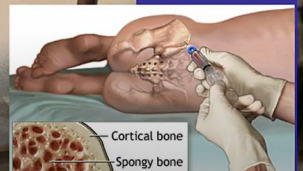
A Congenital Neutropenia

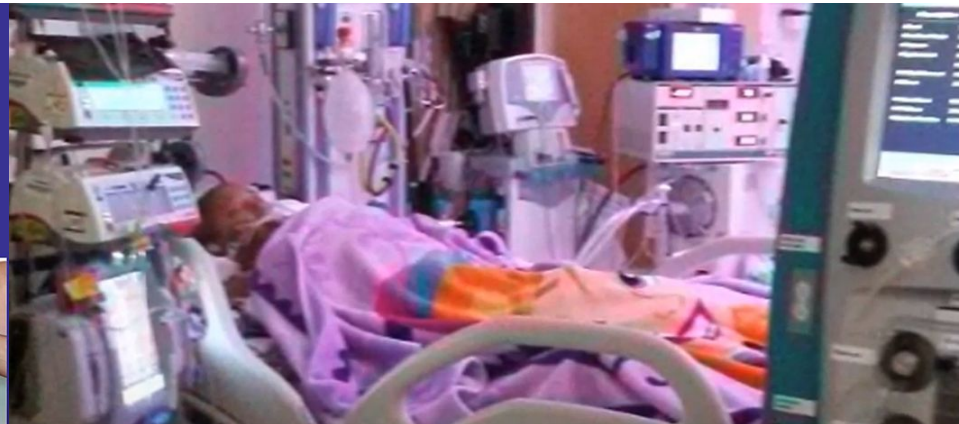
Rare **genetic disorder** affecting protein production in every cell of the body.

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.

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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.

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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?

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1. Create Patient Reported Outcomes / Survey Platform

Coming soon.

- Platform the 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!

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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

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The Coriell Institute and the NIGMS Human Genetic Cell Repository

CORIELL INSTITUTE
FOR MEDICAL RESEARCH

- **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

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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

NIH NIGMS

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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

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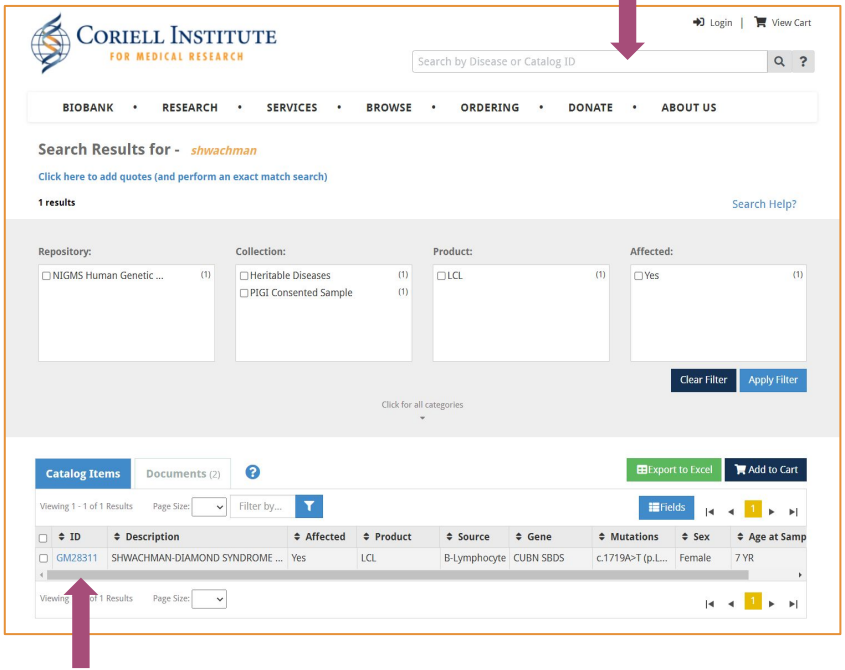
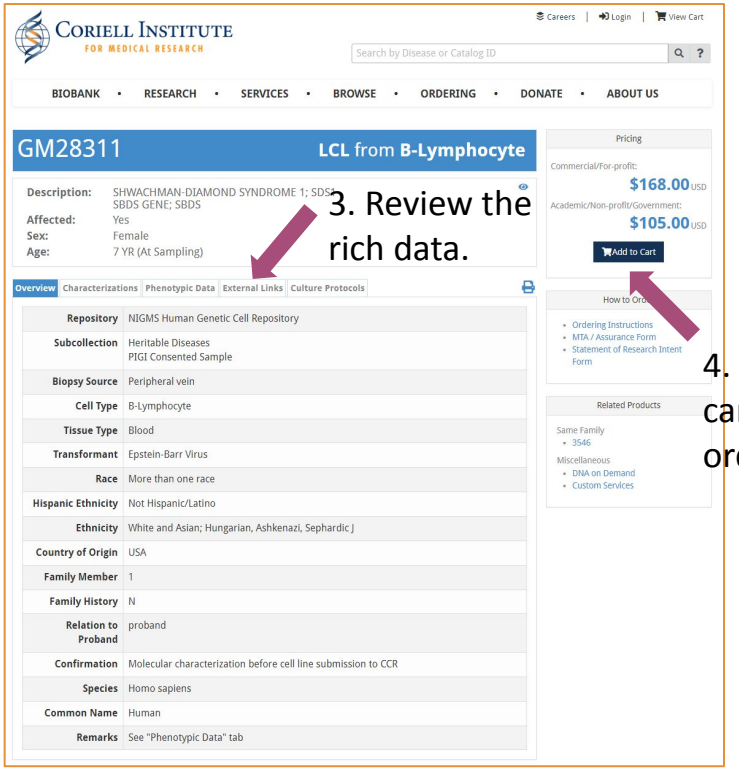
Easy to search and order!

www.coriell.org

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



2. Select the result of interest.



3. Review the rich data.

4. Add to cart to order.

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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

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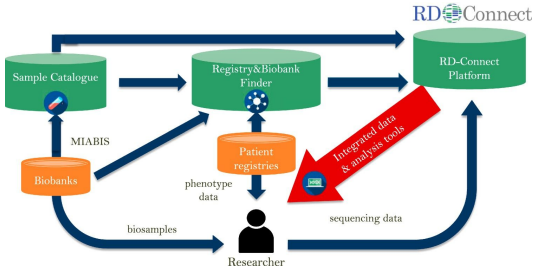
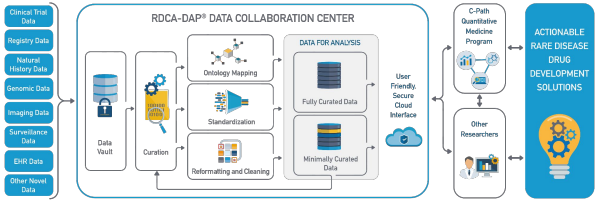
Tools to Address:
Technical (and Regulatory) challenges

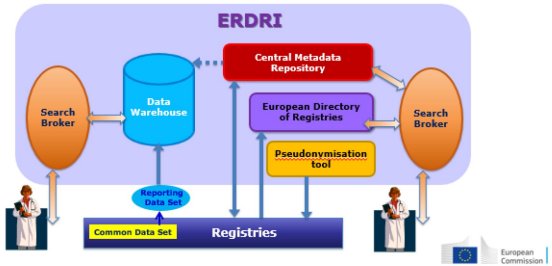
- RD-Connect



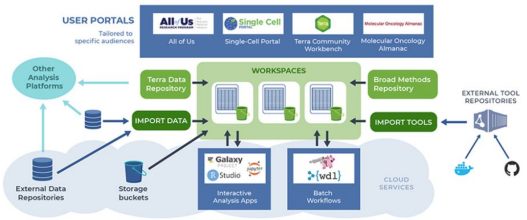
- RDCA-DAP (C-Path)



- European Platform for Rare Disease Registration



- Terra



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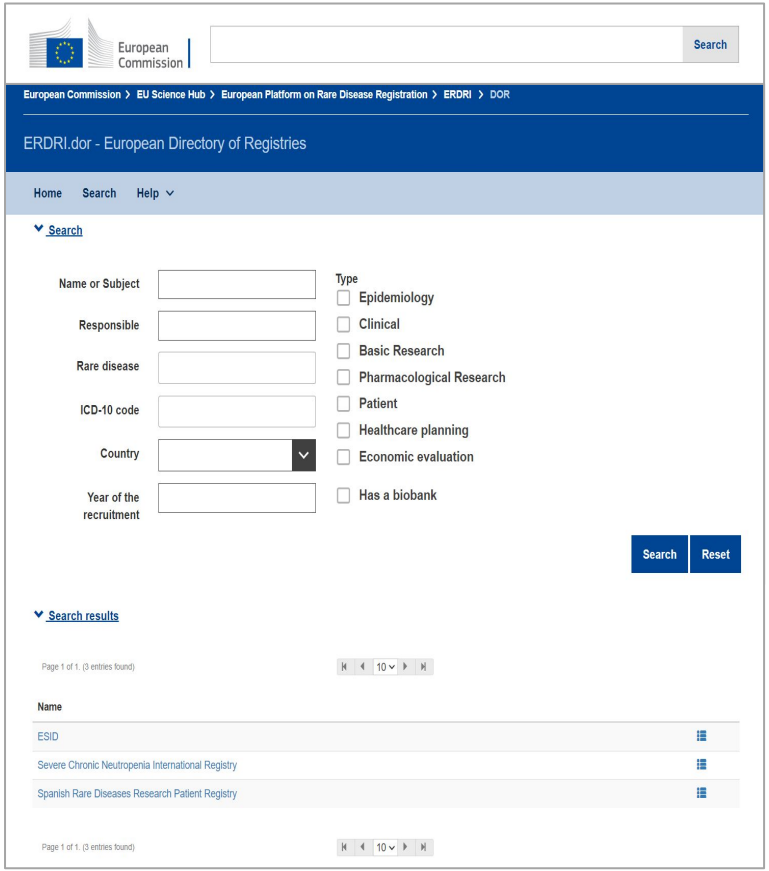
Let’s work together!

Let’s discuss

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



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