

Shwachman Diamond Syndrome Registry and Study

Status: RECRUITING

Eligibility Criteria

Sex: ALL

Age:

Healthy Volunteers:

Inclusion Criteria: Shwachman Diamond syndrome, Shwachman-Diamond Syndrome-Like conditions, or a genetically undefined condition that shares clinical features with Shwachman Diamond Syndrome.

- Biallelic mutations in SBDS, or pathogenic mutations in DNAJC21, EFL1, or SRP54 OR
- Shwachman-Diamond Syndrome defined clinically OR
- Clinically suspected Shwachman-Diamond Syndrome OR
- Phenotypic features suggestive of SDS OR
- Parents, siblings, and other blood relatives of any age, living and deceased, of patients with SDS or SDS-Like conditions are eligible for this study

Exclusion Criteria:

- Patients with other diagnosed causes of bone marrow failure, exocrine pancreatic insufficiency and cancer predisposition will be excluded.

Conditions & Interventions

Conditions:

Shwachman-Diamond Syndrome, Shwachman-Diamond Syndrome-Like

Keywords:

Bone marrow failure, Inherited bone marrow failure syndromes, Bone marrow, Neutropenia, Leukemia, MDS, Pancreas, Cancer predisposition

More Information

Description:

Shwachman-Diamond syndrome (SDS) is a genetic condition characterized by bone marrow failure, medical co-morbidities, and leukemia predisposition. SDS-Like patients share clinical features with SDS but lack mutations in known SDS genes. Since SDS/SDS-Like syndromes are rare diseases, data are sparse regarding the clinical features, natural history, clinical outcomes with current management, and treatment. For this reason, the SDS Registry was formed to collect clinical data from medical records and to bank biological samples with the goal of understanding SDS/SDS-Like diseases to develop better treatments and improve the health of patients with these conditions.

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Principal Investigator:

Principal Investigator ID:

Phase:

IRB Number:

System ID: NCT06056908

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