# Severe Combined Immunodeficiency (SCID) Patient Clinical Journey

## DELAYED DIAGNOSIS – CLINICAL OR SYMPTOM-PRESENTING

*Items in red may be prevented with a Timely Diagnosis.*

### PRE-DIAGNOSIS (before age 1)

**Typical Accompanying Diagnoses**

*The following present between the first month and one year*

- Anemia
- Chronic diarrhea
- Enlarged spleen
- Failure to thrive
- Frequent infections
- Other less common or preventable childhood infections (e.g., chicken pox, CMV, meningitis)
- Pneumonia
- Recurrent infections
- Severe rashes
- Hypogammaglobulinemia
- Lymphocytopenia
- Neutropenia

**Events**

- Hospital stay – *May include reverse isolation protocol*
- PICU stay

**Specialists**

- Dermatologist
- Ear, Nose, and Throat (ENT) Doctor
- Immunologist
- Infectious Disease Specialist
- Pediatrician – *For infectious disease reoccurrences*
- Pulmonologist

**Testing & Procedures**

- Manual blood count – *If a patient is presenting with recurring infections, this test checks to see if there are white blood cells*
- Flow cytometry – *If there are no white blood cell flows, this test checks to see how many T, B, NK cells the patient has*
- Whole genome/whole exome sequencing
- Blood chemistry
- Radiologist/chest x-ray
- Skin exam/skin biopsy
- Bone marrow biopsy

***Without an early diagnosis and treatment, there is a high probability of mortality within first year of life***

YEAR 1

***With treatment, life expectancy varies depending on success of HSCT***

### TIME OF DIAGNOSIS (between 3 months and 1 year)

**Specialists**

- Immunologist
- Hematologist – *Only for transplant patients*

**Treatments & Supportive Therapies**

- Hemopoietic stem cell transplant (HSCT) – *First choice treatment option if there is a match donor. Ideally occurs within 3 months of diagnosis but can vary by patient. Often referred to as a bone marrow transplant (BMT).*
- Immunoglobulin infusions
- Enzyme replacement therapy – *Only for SCID ADA*
- Gene Therapy – *Currently, there are not FDA-approved therapies for all forms of SCID*

### POST DIAGNOSIS (up to age 15)

**Treatments & Supportive Therapies**

- Hemopoietic stem cell transplant (HSCT) – *First choice treatment option if there is a match donor. Ideally occurs within 3 months of diagnosis but can vary by patient.*
- Immunoglobulin infusion – *Supportive therapy that can be lifelong for a patient, in addition to stem cells or gene therapy.*
- Prophylaxis treatment
- Gene therapy – *Currently, not all forms of SCID have an FDA-approved therapy.*
- *The following treatment options are only as needed and depend on the type of SCID a patient has. They are usually not a direct result of SCID itself but rather damage done from not having an immune system.*
  - Auditory Therapy
  - Behavioral therapy
  - Feeding tube
  - Neurodevelopmental Delays

**Specialists**

- Counseling Support Services
- Immunologist
- Behavioral Therapist
- Genetic Counselor
- Mental Health Counselor
- Physical Therapist (PT)/Occupational Therapist (OT)/Speech Therapist (ST)

## TIMELY DIAGNOSIS

*Items in green are typically only seen with a Timely Diagnosis.*

*For a SCID patient, a timely diagnosis is considered a diagnosis via newborn screening or pre-natal testing.*

### TIME OF DIAGNOSIS

**Testing & Procedures**

- Newborn screening (dried blood spot) measuring T-cell receptor excision circles (TREC) with additional test to determine which cells involved (B, T, NK)
- Manual blood count – *If a patient is presenting with recurring infections, this test checks to see if there are white blood cells.*
- Flow cytometry – *If there are no white blood cell flows, this test checks to see how many T, B, NK cells the patient has.*
- Blood chemistry
- Bone Marrow biopsy

**Events**

- Hospital stay – *May include reverse isolation protocol*

**Specialists**

- Immunologist
- Hematologist – *Only for transplant patients*
- Pediatrician

**Typical Accompanying Diagnoses**

- Hypogammaglobulinemia
- Lymphocytopenia
- Neutropenia

**Treatments & Supportive Therapies**

- Hemopoietic stem cell transplant (HSCT) – *First choice treatment option if there is a match donor. Ideally occurs within 3 months of diagnosis but can vary by patient. Often referred to as a bone marrow transplant (BMT).*
- Immunoglobulin infusions
- Enzyme replacement therapy – *Only for SCID ADA*
- Prophylaxis treatment
- Gene therapy – *Currently, not all forms of SCID have an FDA-approved therapy.*

### POST DIAGNOSIS (after age 1)

**Specialists**

- Counseling Support Services
- Immunologist
- Behavioral Therapist
- Genetic Counselor
- Mental Health Counselor

**Treatments & Supportive Therapies**

*The following are possible secondary treatments for the loss of an immune system:*

- Gene therapy
- Hemopoietic stem cell transplant (HSCT)
- Immunoglobulin infusions
- Prophylaxis treatment

**This guide is not intended as a clinical decision-making tool. Please consult your specialists for any medical decisions.*