

Novel Genetic Disorders of the Immune System

NCT02257892

Status	RECRUITING
Sponsor	National Institute of Allergy and Infectious Diseases (NIAID)
Enrollment	500 participants

Key Eligibility Criteria

Inclusion (13)

- Patient and relatives aged 0-99 years old to include women who are pregnant or breastfeeding. Only patients ≥ 2 years of age, in stable clinical status and meeting the weight requirement of the NIH CC will be physically evaluated at the NIH CC.
 - Willingness to allow storage of blood, saliva, and other tissue specimens for future use in medical research.
 - Willingness to participate in genetic testing and allow sharing of genetic information in secure databases like dbGAP. These tests may include, but are not limited to, whole exome and whole genome sequencing.
 - Priority may be given to individuals with a family history (if readily available) suggestive of multiple affected members with a constellation of signs and symptoms suggestive of immune dysfunction among first- or second-degree relatives.
 - Eligibility of special populations
- ... and 8 more (see full listing online)

Exclusion (7)

- Patients will be excluded for any of the following:
 - Known genetic disorders that are already well characterized, such as severe combined immunodeficiency (SCID), chronic granulomatous disease (CGD), etc., and those in which we do not have an enduring research interest in the LCIM.
 - Patients with unknown immune disorders will be excluded if they have received chemotherapy within the last 6 months for a malignancy or have infections such as HIV or mycobacterial infections.
 - Severe clinical illness requiring highly specialized teams and institutions. The NIH may not be able to provide appropriate care for certain referred cases. The Principal Investigator (PI) may determine that the patient is eligible for enrollment but ineligible for admission to the Clinical Center. Patients and relatives with certain obstetric issues may pose a safety risk for travel and evaluation here. Eligibility for this group will be determined on a case by case basis by the PI.
 - Patients with well-defined autoimmune conditions such as systemic lupus erythematosus (SLE), Hashimoto's thyroiditis, Addison's disease, Graves disease, sarcoidosis and rheumatoid arthritis, among others.
- ... and 2 more (see full listing online)

Locations (1 total)

National Institutes of Health Clinical Center, Bethesda, Maryland, United States