

Natural History Study of GATA2 Deficiency and Related Disorders

NCT01905826

Status	RECRUITING
Sponsor	National Institute of Allergy and Infectious Diseases (NIAID)
Enrollment	600 participants

Plain Language Summary

This study is following people who have a rare genetic condition called GATA2 deficiency — which affects the immune system and blood — to learn more about how the disease progresses and what treatments work best over time.

****You may be eligible if...****

- You have been diagnosed with GATA2 deficiency (a rare genetic mutation affecting your immune and blood cells)
- You have a related condition such as a weakened immune system, bone marrow failure, lymphedema (swelling), or certain skin or hearing problems that may indicate GATA2 deficiency
- You are willing to come in for regular monitoring visits
- Family members of affected patients may also be eligible for genetic testing

****You may NOT be eligible if...****

- You are not able to travel to the study site for evaluation
- You are unwilling to consent to genetic testing and long-term follow-up
- You have another established diagnosis that fully explains all of your symptoms

Talk to your doctor to see if this trial is right for you.

Key Eligibility Criteria

Inclusion (14)

- Males and females greater than or equal to 2 years old must meet the following criteria to be eligible for participation in this study:
- Have a mutation in GATA2 proven by genetic testing (previous test results will be accepted) OR meet both of the following criteria:
- Clinical characteristics strongly consistent with GATA2 deficiency per the following criteria and at the discretion of the principal investigator (PI). Individuals without a GATA2 mutation must have a past or present history of 1 or more of the following to be considered for study enrollment:
- Disseminated NTM or invasive fungal infection.
- Severe or recurrent HPV or herpesvirus infection.

... and 9 more (see full listing online)

Exclusion (1)

- Individuals with any condition or who are taking any medications that, in the opinion of the investigator, contraindicates participation in the study will be excluded.

Locations (1 total)

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

<https://clinicaltrials.gov/study/NCT01905826>

DISCLAIMER: This document is for informational purposes only and does not constitute medical advice. Always consult your healthcare provider before enrolling in any clinical trial. Information may not be up to date — verify details at [ClinicalTrials.gov](https://clinicaltrials.gov). Generated by [ClinicalTrialsFinder.org](https://clinicaltrialsfinder.org).