

# Help Us Find a Biomarker for Citrin Deficiency

Advance research for citrin deficiency by donating a bit of blood & answering simple questions

## What is citrin deficiency?

Citrin deficiency is a rare genetic condition with several age-dependent stages. There is currently no cure for the condition other than liver transplantation, and patients manage the condition with lifelong dietary management. After the first year of life, patients may develop symptoms such as fatigue, growth impediments, stomach aches, and fatty liver. If the condition is not well managed, patients may develop more serious complications such as increased levels of ammonia in the blood (hyperammonaemia) that can be fatal if untreated. There remains an urgent unmet need to develop effective therapies for citrin deficiency.

## What is a biomarker and why is it important?

A biomarker is a measurable substance in the body that helps track specific disease progression, or response to treatments. **Currently, there are no reliable biomarkers for citrin deficiency.** This makes it difficult to track how well patients are faring, and it also impedes the development process of effective treatments.

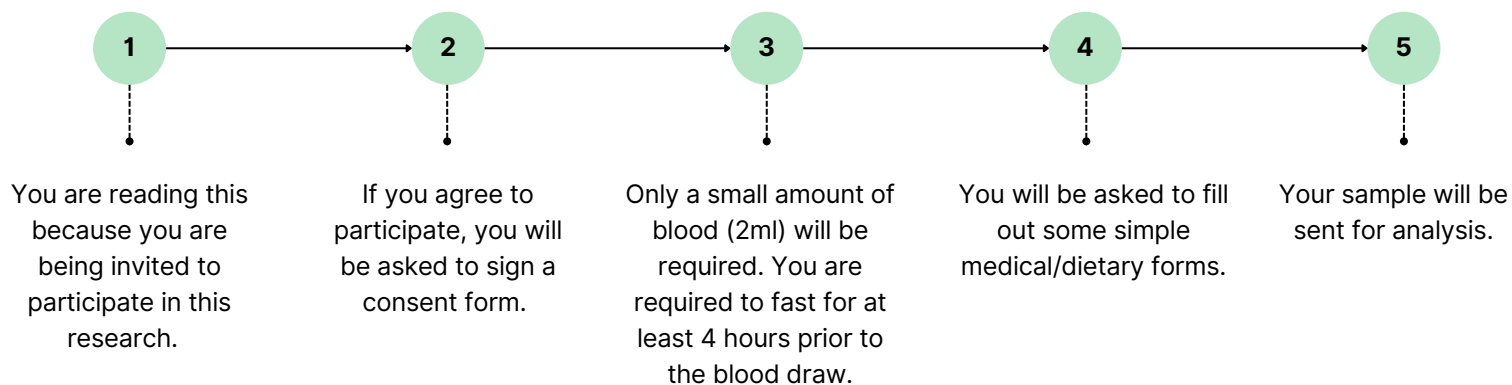
## The Citrin Foundation Global Omics Study

This landmark study aims to identify biomarkers in blood samples collected from at least 100 patients with citrin deficiency, as well as those from healthy volunteers to serve as controls. This is a multi-center project where participants can enroll from around the world through collaborating hospitals. The study is scheduled to be completed in 2025, followed by publication of the results in peer-reviewed scientific journals.

## Who can participate?

- Citrin deficiency patients of any age with a confirmed diagnosis.
- Volunteers of any age with no citrin deficiency or similar disorders as decided by your doctor.
- **Individuals who are carriers of citrin deficiency are not eligible to participate.**

## What does taking part entail?



Your participation, whether as a patient or a healthy volunteer, will contribute substantially to citrin deficiency research, leading to better patient care and the development of effective treatments. Join our study to advance research on citrin deficiency. Please inform your doctor if you're interested in participating in this landmark study!