



Frequently Asked Questions

What is the program's purpose?

This program has two main goals:

- To provide answers to patients with mysterious conditions that have long eluded diagnosis.
- To advance medical knowledge about rare and common diseases.

How will the program define an undiagnosed disease?

Any medical condition that eludes diagnosis by a referring physician can be considered undiagnosed and may be of interest to this pilot study. Some patients wait years for a definitive diagnosis. For this study, only a fraction of cases referred will be invited to proceed in the study at the discretion of the programs medical team.

What is a rare disease?

A rare disease is generally considered to have a prevalence of fewer than 200,000 affected individuals in the United States.

How will the program work?

If you are interested in participating in this clinical research program, discuss the option with your primary physician or health-care provider (nurse practitioner or physicians assistant). Information specialists at the Clinical Center's Patient Recruitment Call Center (1-866-444-8806) can provide more information about eligibility and what kinds of medical information referring physicians must submit for review by the programs medical team. You or your health-care provider can call.

Patients must be referred by a physician or health-care provider. Information your physician must provide directly to NIH includes:

- A summary letter describing your condition, when it was first noted and your current health status.
- A list of treatments and medications that have already been tried and their effects.
- Copies of reports and results of pertinent diagnostic tests, along with X-rays, MRI results, and other imaging records/studies. Copies of the actual imaging studies should be sent on CD.
- Referring physicians of pediatric patients should also send prenatal and birth history, growth curves, and photos or videos of the patient. Family history, at least of the nuclear family, should also be provided.

Ask for and keep a copy of these materials for yourself. Because of patient confidentiality considerations, no e-mail submissions will be accepted. Your referring physician must mail the summary letter and related materials to:

**National Human Genome Research Institute
National Institutes of Health
Undiagnosed Diseases Program
10 Center Drive - MSC 1851, Building 10, Room 10C103
Bethesda, MD 20892-1851**

The Undiagnosed Diseases Program staff will notify you by mail when the information from your physician has been received and if additional information is required before the chart can be reviewed.

The program's medical team will review the information submitted for each case. Cases meeting the criteria will be presented to the Undiagnosed Diseases Program's board of specialists for further consideration. Plans call for inviting 50 to 100 patient participants to the NIH annually for a thorough evaluation and consultation.

If your case is accepted for NIH evaluation, NIH will provide information from the evaluation to you and to your health-care provider, who will be responsible for your medical care after you leave the NIH. In this manner, follow-up care will be assured.

Will all cases submitted be reviewed by the Undiagnosed Disease Program?

Yes, the programs medical team will review the cases submitted for consideration. Cases meeting Program criteria will be presented to the board of specialists. Fifty to 100 cases will be invited to NIH for thorough evaluation and consultation each year.

What's involved for patients accepted by NIH for participation?

Patients in the program will be evaluated using the unique combination of scientific and medical expertise and resources at the NIH Clinical Center in Bethesda, Md. Dozens of NIH senior attending physicians may be asked to consult on the cases and evaluations. Their specialties include rheumatology, immunology, oncology, mental health, nephrology, hematology, ophthalmology, neurology, laboratory medicine, pain and palliative care, bone disorders, endocrinology, oncology, immunology, dermatology, primary immunodeficiency, dentistry, genetics, pathology, pulmonology, cardiology, primary immunodeficiency, internal medicine, pediatrics and hepatology.

The referring provider and patient will receive the information resulting from NIH evaluation as a part of this program. Long-term care will not be provided by the NIH, but selected patients may be eligible for an ongoing research study.

Will all evaluations result in a diagnosis?

No, but the evaluations will yield valuable information medical researchers will use to:

- Help identify previously unrecognized rare diseases.
- Suggest new ways to treat and prevent common illnesses.
- Determine promising options for continued medical research.

How can I find out more about participating?

Call 1-866-444-8806

Information specialists with the Clinical Center Patient Recruitment Call Center will ask you a few questions to begin to determine your eligibility to participate. A physician's referral is required.

Who is eligible?

Generally, physicians may refer individuals who:

- Are at least six months old.
- Have a disease or condition that remains undiagnosed despite ongoing care and follow-up by a physician or other healthcare provider.
- Are able to travel.

Will participants also have DNA taken and studied as part of this program?

Yes, the hope is that DNA will be one way that helps the NIH medical team determine a patient's illness. Patients will be informed of how the DNA will be used.

What is the cost for participating if accepted?

There is no charge for participating in this research program at the Clinical Center. Travel, meals and lodging expenses may be covered for research participants according to NIH policies.

I don't have a physician and I don't have health insurance. What are my options for participating in this research program?

A referral from a primary healthcare provider is required. If you don't have a personal physician, but receive care in a clinic or other healthcare setting, discuss referral with a member of the medical team currently providing your care. This could be a nurse practitioner or a physician's assistant.

Why is a physician's referral required?

Consultation with the patient's primary health-care provider will be important throughout the process. Follow-up care will be addressed as part of the evaluation at NIH. If your case is accepted for NIH evaluation, NIH will provide information from the evaluation to you and to your healthcare provider, who will be responsible for your follow-up medical care.

Why isn't NIH providing the follow-up care?

Long-term follow up care at the NIH is not part of this program. The NIH Clinical Center is a research hospital and its mission is to perform clinical research to advance the overall health of the nation.

How will I find out NIH's decision on accepting my case for evaluation as part of this research program?

The decision will be communicated in writing to you and to the primary healthcare provider who sent your summary letter and other medical records. Once all materials have been received, NIH review is expected to take about 8 to 12 weeks.

If my case is accepted for evaluation at NIH, what happens next?

You may be asked to provide copies of additional medical records and to travel to the NIH Clinical Center in Bethesda, Md, for comprehensive medical evaluations. This will be a 2 to 5 day visit for inpatient and/or outpatient care. Travel, meals and lodging expenses may be covered for research participants according to NIH policies.

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