

## About the Undiagnosed Diseases Network

### What is the Undiagnosed Diseases Network (UDN)?

The UDN is a research study funded by the National Institutes of Health Common Fund. The UDN is made up of clinical and research centers across the United States working to improve diagnosis and care of patients with undiagnosed diseases.

### What is the purpose of the UDN?

The UDN has two main goals:

1. To provide answers for patients and families affected by mysterious conditions, and
2. To learn more about rare and common diseases.

### Where are the UDN clinical sites?

- Bethesda, MD ([NIH Undiagnosed Diseases Program](#))
- Boston, MA ([UDN Clinical Site at Harvard Medical School](#))
- Durham, NC ([Duke University and Columbia University](#))
- Houston, TX (Baylor College of Medicine, Texas Children's Hospital, and Baylor CHI St. Luke's Medical Center)
- Los Angeles, CA ([UCLA Undiagnosed Diseases Clinic](#))
- Miami, FL (University of Miami School of Medicine)
  
- Nashville, TN ([Vanderbilt Center for Undiagnosed Diseases](#))
  
- Philadelphia, PA ([Children's Hospital of Philadelphia](#) and University of Pennsylvania)
- Salt Lake City, UT (University of Utah - Intermountain West)
- Seattle, WA ([Pacific Northwest Undiagnosed Diseases Clinical Site at University of Washington and Seattle Children's Hospital](#))
- Stanford, CA ([Center for Undiagnosed Diseases at Stanford](#))
- St. Louis, MO ([Washington University in St. Louis](#))

### What is unique about the UDN?

The UDN is trying to solve the most challenging medical mysteries by:

- Bridging the gap between clinical care and research.
- Using genetic data to try to find diagnoses.

- Working with researchers to figure out how diseases affect the body, which may lead to treatments.
- Training other doctors, nurses, genetic counselors, and scientists to use this new approach.

### **What is an undiagnosed disease?**

An undiagnosed disease is a medical condition without a known cause despite a lot of evaluation.

### **What is a rare disease?**

A rare disease is a disease that affects fewer than 200,000 people in the United States. There are so many separate rare diseases, that overall 1 in 10 people have a rare disease.

### **Who is eligible for the UDN study?**

Eligible participants:

- Have a condition that remains undiagnosed despite thorough evaluation by a health care provider
- Have at least one objective finding
- Agree to the storage and sharing of information and biomaterials in an identified fashion amongst the UDN centers, and in a de-identified fashion to research sites beyond the network

## **Applying to the UDN**

### **How do I apply to the UDN study?**

Before applying to the UDN, it is important to discuss the study with a health care provider. If you want to submit the application online, you will need an email address and access to the Internet. If you do not have an email address and access to the Internet, you can request a [paper application](#) by calling 1-844-RING-UDN (1-844-746-4836). You will also need to ask your health care provider to write a study recommendation letter. The study recommendation letter must include:

- A summary of the applicant's medical problems
- Date when symptoms were first noticed
- Previous diagnoses
- History of evaluations and tests

- History of treatments and medications
- Current medications
- Family history
- Health care provider's diagnostic impressions
- For pediatric patients: prenatal and birth history

Once you have the study recommendation letter, you can fill out an online application through our website, which we call the [UDN Gateway](#).

You will upload the study recommendation letter directly to this website. It should take less than 20 minutes to complete the online application. Please DO NOT email the application or study recommendation letter to the UDN.

When the UDN has received your application, you will receive a confirmation email. The UDN will let you know if more information is needed before your application can be reviewed.

**Why do I need a study recommendation letter from my health care provider?**

The UDN evaluation is only one part of your medical care. Your health care provider has been with you before the UDN evaluation and will likely continue to provide your medical care after the UDN evaluation. If you are accepted into the UDN study, the UDN will provide information from the evaluation to you and to your health care provider. Your health care provider will be responsible for your follow-up medical care.

**I have multiple health care providers. Who should I list on the application?**

Please provide contact information for the health care provider who should receive updates about your application.

**My relative is a health care provider. Can they write the study recommendation letter?**

No. Study recommendation letters must be written by health care providers who are not related to the applicant.

**I don't have a primary health care provider and I don't have health insurance. Can I still apply?**

Yes, but you will need a study recommendation letter from a licensed health care provider. If you receive care in a clinic or other health care setting, you can discuss your application with a member of the medical team.

Having insurance is not a requirement to participate in this study. We aim to minimize your out of pocket costs as much as possible. If you are accepted into the UDN, talk with your UDN clinical site about any possible charges you may incur.

**More than one member of my family is affected by an undiagnosed condition. Do I need to submit an application for each person?**

No, one application should be submitted even if there are multiple people affected in a family. This application should be for one individual, not the whole family (ex. “John Smith”, not “Smith Family”). You may upload multiple study recommendation letters to a single application.

**I need help submitting my application. Who should I contact?**

The UDN Helpdesk at the UDN Coordinating Center can answer general questions about the UDN research study and assist applicants with submitting applications. The Helpdesk phone number is 1-844-RING-UDN (1-844-746-4836). The Helpdesk is open Monday–Friday, 9am – 5pm ET. The Helpdesk is closed on national holidays.

**Is information available in languages other than English?**

The Helpdesk uses an on-demand translation service to communicate with applicants that do not speak English. The paper UDN application is available in Spanish. Paper applications completed in Spanish are back-translated into English at the Helpdesk.

**What happens with my application?**

After your application is submitted, it will be assigned to one of the UDN clinical sites for review. The assigned clinical site will contact you (typically within one month of application submission) to request additional information, such as:

- Medical records
- Laboratory studies
- MRI, X-ray, or CT images (can be sent on CD)
- Photographs or videos
- Pathology slides and reports
- For pediatric patients: growth curves

In order to help with the application review process, you can begin to collect these records

while preparing your application. However, you should not send any medical records until you receive specific instructions to do so from your assigned UDN clinical site. Your assigned clinical site will provide instructions for submitting medical records. Do not send any medical records to the UDN Coordinating Center as this may delay processing of your application.

After all of the necessary information is received by a UDN clinical site, it typically takes 6-8 weeks for the network to decide whether to accept an applicant.

**Can I choose which UDN clinical site I'm assigned to?**

You may ask to be assigned to a specific UDN clinical site on your application if you want to. However, there is no guarantee that you will be assigned to that site.

**If I apply, does that automatically mean that my application will be reviewed?**

Yes.

**If I apply, does that automatically mean I can go to a UDN clinical site?**

No. The UDN can only accept a small number of participants. The UDN team reviews each application very carefully to decide who to accept into the study. Even if you are not accepted, you might receive some useful feedback about your medical condition.

**How will I find out the UDN's decision?**

We will contact you and the health care provider you list on the application.

**If my application is not accepted, can I ask someone to review the application again?**

If you have new medical information, you can ask the assigned UDN clinical site to review your application again. However, there is no guarantee that the decision will change.

The UDN makes all decisions as a network. There is no process to request for someone else to review an application after it is not accepted.

**UDN Evaluation Process**

**If I am accepted into the UDN study, what happens next?**

You will work with your assigned UDN clinical site to determine the dates for a 2-5 day visit to the clinical site. You will travel to your assigned UDN clinical site for a medical and research evaluation. You may also be asked to provide copies of additional medical records.

**What will happen at the clinical site?**

You will be asked to enroll in the study and sign a consent form. You will also be asked to provide information about the health of your family members and to give permission for photographs, blood samples, and other samples (such as skin) to be taken.

You may be asked to send a blood sample for genetic testing to the clinical site several weeks before your evaluation. Based on the types of symptoms you have, various medical specialists will evaluate you during your clinical site visit. You will also be asked to fill out some forms and surveys regarding your health and your experience with the UDN before, during, and after the clinical site visit.

The UDN works to provide a similar experience for all UDN participants, however, practices may vary by clinical site.

**Will participants also have DNA tested as part of this study?**

Most, but not all of the time, blood will be taken for DNA testing. The hope is that DNA testing will help the UDN team diagnose conditions. You will be informed about how your DNA will be used. If available and willing to participate, your biological family members may also provide DNA samples for testing. Your UDN clinical site will determine which family members should be asked to participate. More information about genetic testing through the UDN is available in the [Genetic Testing Handout](#).

There may be people who have a history that suggests that the undiagnosed disease is not genetic. If you suspect an environmental cause of your condition, be sure to say so on the application.

**Why might my family members need to be tested?**

If your DNA is being analyzed, it is helpful to also get blood from your family members to compare their DNA with your DNA. This helps the UDN team to assess the meaning of your results.

**What if my family members are unable or unwilling to participate?**

You may still qualify to participate. However, this situation may make it harder for the UDN team to find a diagnosis.

**Do my family members or I have to pay for the genetic testing?**

No. The UDN will cover the cost of genetic testing if you are accepted into the study.

**What laboratory will do the genetic testing?**

Baylor College of Medicine in Houston, Texas.

**Will the UDN evaluation give me a diagnosis?**

The goal is to give you a diagnosis, but this may not happen.

**Will I receive treatment through the UDN?**

No, treatment will usually not be provided as part of the UDN. Treatment will remain your and your health care provider's responsibility. But you and your provider may receive treatment recommendations from the UDN.

**What is the cost for participating if accepted?**

We aim to minimize your out of pocket costs as much as possible. If you are accepted into the UDN, talk with your clinical site representative about any possible charges you may incur.

**What happens after the UDN evaluation?**

After the UDN evaluation, you and your health care provider will receive the information from the evaluation. Long term care will not be provided by the UDN. Some UDN participants may be eligible for other research studies at the UDN site or other academic centers.

**Health Care Providers**

**My patient asked me to write a recommendation letter. What should this letter include?**

The study recommendation letter must include:

- A summary of the applicant's medical problems
- Date when symptoms were first noticed
- Previous diagnoses
- History of evaluations and testing
- History of treatments and medications
- Current medications
- Family history
- Health care provider's diagnostic impressions
- For pediatric patients: prenatal and birth history

Example letters are available [here](#).

**How do I send the recommendation letter to the UDN?**

If your patient has started an application, the letter can be given to your patient to submit with the application.

If your patient has not started an application, you can fill out an application on their behalf through the [UDN Gateway](#). It should take less than 20 minutes to complete the online application. The recommendation letter can be uploaded to this application.

If you are unsure if your patient has started an application, please contact the UDN Helpdesk (email: [UDN@hms.harvard.edu](mailto:UDN@hms.harvard.edu), phone: 1-844-746-4836).

**I want to receive updates about my patient's application. How do I get updates?**

If you would like to receive updates about your patient's application, please ask your patient to list your contact information on their application.

**How will I find out the UDN's decision?**

We will contact you and your patient with the decision after the application review is complete.

**If my patient is accepted, will the UDN take over my patient's care?**

No. After the UDN evaluation, you will receive the information from the evaluation. Long



term care will not be provided by the UDN.

## **Contact Information**

You can call or email the Helpdesk at the UDN Coordinating Center to find out more about:

- Participating in the UDN research project
- Eligibility
- What to include in the study recommendation letter
- The status of your application
- Requesting a paper application

Hours: Monday-Friday, 9am - 5pm ET

Email: [UDN@hms.harvard.edu](mailto:UDN@hms.harvard.edu)

Phone: 1-844-746-4836 (1 844 Ring UDN)

Note: please do not contact the main hospital of the UDN clinical sites.