



Paper Application Process

Please return your completed application and study recommendation letter by mail or fax to:

Undiagnosed Diseases Network
Attn: Paul Mazur
Department of Biomedical Informatics
Harvard Medical School
10 Shattuck Street
Boston, MA 02115
Fax: (617) 432-5105

Alternatively to apply online go to: <http://undiagnosed.hms.harvard.edu/apply>

If you have any questions, please contact:

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

Email: UDN@hms.harvard.edu

Are you the applicant or legal guardian of the applicant?☐ Yes

☐ No- Please review the information on this page with the applicant or their legal guardian. By agreeing to the fields on this page, you are confirming that the applicant or their legal guardian agrees to the information.

Name: _____

Relation to applicant: _____

Directions

Please read the following consent form carefully. The consent form refers to the person applying to the Undiagnosed Diseases Network (UDN) and “you” refers to the applicant.

Introduction

We invite you to take part in a research study supported by the National Institutes of Health (NIH).

First, we want you to know that:

- Taking part in this research is entirely voluntary.
- You may choose to not take part, or you may withdraw from the study at any time. In either case, you will not lose any benefits to which you are otherwise entitled.
- You may receive no benefit from taking part. The research may give us knowledge that may help in the future.

Second, some people have personal, religious, or ethical beliefs that may limit the kinds of medical or research treatments they would want to receive (such as blood transfusions). If you have such beliefs, please discuss them with your health care providers or research team before you agree to the study.

Now we will describe this research study. Before you decide to take part, please take as much time as you need to ask any questions and discuss this study with anyone at the Undiagnosed Diseases Network (UDN), or with your family, friends, or your personal doctor or other health professional.

☐ I have read and understand this section.

About the Study

Why is this study being done?

The Undiagnosed Diseases Network (UDN) is a group of medical and research centers across the United States. The purpose of the UDN research study is to diagnose people with unnamed conditions and improve our ability to care for people with rare and undiagnosed diseases.

What is involved in the study?

To apply to the UDN, you will likely spend an hour or more completing the information on this web site and speaking with your health care provider. Once you complete your application, it will be given to one of the UDN medical centers. The medical center will contact you and ask you to send all of your medical records. This may take you a couple of hours or many hours to collect and send your records to the medical center. Medical information, including photographs, about you will be shared with health care providers in the UDN.

We cannot accept everyone into the UDN. In order to determine which people to accept, we ask applicants to send us a letter from a health care provider, medical records that describe their disease, and medical test results. We also ask for the applicant's name, contact information, and some additional information, like race and gender. If you apply and provide this information but are not accepted into the UDN, we will keep this information. Keeping your information will allow us to evaluate our admission process. We may publish some of this information in papers about the UDN. All of these papers would discuss the UDN applicants as a whole and would not include any of the information that could identify you, like your name or birth date. If you are accepted into the UDN you will be asked to sign a consent form for the entire study.

If you are not accepted into the UDN, there is no process to request for someone else to review your application. If you have new medical information, you can ask the UDN medical center to review your application again. However, there is no guarantee that the decision will change.

If you are accepted into the UDN, you may need to spend several more hours obtaining additional medical records and speaking with the medical center staff to plan the visit. You would travel to one of the UDN medical centers to be seen by a team of doctors, nurses, and other people who work at a hospital. This visit would last about a week. During the visit, you would be looked at by multiple doctors. We would ask you questions about your health and the health of your family. After the visit, you would likely talk to people at the medical center regularly about updates of the testing.

☐ I have read and understand this section.

Risk and Benefits

What are the risks of applying to the UDN?

The risks of applying to the UDN and sending information about your condition are:

1. *Use of information:* Some people are concerned that information about them from their medical records could be released. Possible problems related to the release of information include trouble getting insurance or getting a job. We will try to make sure that this does not happen by taking measures to protect your information.
2. *Unanticipated medical information:* It is possible (although not likely) that we will find information about your health that you did not expect. If this information is important to your health care, we will give you and your doctor the information.

If you are accepted into the UDN you will be asked to sign another consent form that goes over the risks of the entire study.

Your decision to participate in this study will not affect your current health care.

What are the benefits of applying to the UDN?

Applying to the UDN may not benefit you directly. You may receive a diagnosis if you are accepted into the UDN.

What are my other options?

You do not have to participate in this study if you do not want to.

What if I change my mind?

You may stop participating at any time. If you choose, you may request to have your information destroyed.

☐ I have read and understand this section.

Confidentiality

Who else will know that I am in this study?

Only people at the UDN medical and research centers will know that you applied to the UDN and will have your contact information. All of the information that you share with the UDN during the application process will be kept confidential and private.

☐ I have read and understand this section.

Payment

Will I receive payment for being in this study?

You will not receive payment for taking part in this study. We will cover travel expenses and meals during the visit if you are accepted into the UDN.

☐ I have read and understand this section.

Other Pertinent Information

1. **Confidentiality.** When results of a research study are reported in medical journals or at scientific meetings, the people who take part are not named and identified. In most cases, the UDN will not release any information about your research involvement without your written permission. However, if you sign a release of information form, for example, for an insurance company, if asked the UDN will give the insurance company information from your medical record. This information might affect (either favorably or unfavorably) the willingness of the insurance company to sell you insurance. The Federal Privacy Act protects the confidentiality of your UDN medical records. However, you should know that the Act allows release of some information from your medical record without your permission, for example, if it is required by the Food and Drug Administration (FDA), members of Congress, law enforcement officials, or authorized hospital accreditation organizations.
2. **Policy Regarding Research-Related Injuries.** In general, no long-term medical care or financial compensation for research-related injuries will be provided by the UDN medical and research centers or the Federal Government. However, you have the right to pursue legal remedy if you believe that your injury justifies such action.
3. **Payments.** In general, patients are not paid for taking part in research studies at the UDN medical and research centers. Reimbursement of travel and meals will be offered consistent with center guidelines.
4. **Problems or Questions.** If you have any problems or questions about this study, or about your rights as a research participant, or about any research-related injury, contact the UDN Coordinating Center through the website, by email, or by phone.
Email address: UDN@hms.harvard.edu
Phone: 1-844-RINGUDN (1-844-746-4836)
5. **Future Contact.** In the future, we may re-contact UDN applicants to ask if they are interested in participating in activities like surveys and interviews.
6. **Consent Document.** Please keep a copy of this document in case you want to read it again.

☐ I have read and understand this section.

☐ I would like to be contacted in the future even if I am not accepted into the UDN.

Applicant Information

Applicant or parent/guardian email address: _____

Email address belongs to (e.g. applicant, mother, father): _____

Applicant first name: _____

Applicant last name: _____

Applicant street address: _____

Applicant apt/suite/floor (optional): _____

Applicant city, state, zip code: _____

Applicant country: _____

Applicant phone number: _____

Parent/guardian name (if applicable): _____

Applicant Information

Date of birth: _____

Sex: ☐ Male ☐ Female ☐ Other

If other, what sex was assigned at birth? _____

If other, what is the applicant's current gender identity? _____

Race (please select all that apply):

☐ American Indian or Alaska Native

☐ Native Hawaiian or Other Pacific Islander

☐ Asian

☐ White

☐ Black or African American

☐ Other

If other, please explain: _____

Ethnicity (please select one of the following):

☐ Not Hispanic or Latino ☐ Hispanic or Latino ☐ Unknown/Not Reported Ethnicity

Category of Applicant's Primary Symptoms

Many patients with undiagnosed conditions have symptoms that fall into multiple categories. However, it would be helpful for us to know the category that BEST describes the applicant's primary symptoms. If possible, please select the symptom category from the list shown below that BEST describes the applicant's primary symptoms. Not answering this question or not answering it correctly will not impact the status of the application.

- ☐ Allergies and disorders of the immune system
- ☐ Cardiology and vascular conditions (heart, artery, vein, and lymph disorders)
- ☐ Dentistry and craniofacial (bones of head and face)
- ☐ Dermatology (skin diseases and disorders)
- ☐ Endocrinology (disorder of the endocrine glands and hormones)
- ☐ Gastroenterology (disorder of the stomach and intestines)
- ☐ Gynecology and reproductive medicine (disorder of the reproductive system)
- ☐ Hematology (blood diseases and disorders)
- ☐ Infectious diseases
- ☐ Musculoskeletal and orthopedics (structural and functional disorders of muscles, bones, and joints)
- ☐ Nephrology (kidney diseases and disorders)
- ☐ Neurology (disorders of the nervous system, including brain and spinal cord)
- ☐ Oncology (tumors and cancer)
- ☐ Ophthalmology (eye disorders and diseases)
- ☐ Other (please explain: _____)
- ☐ Psychiatry
- ☐ Pulmonology (lung disorders and diseases)
- ☐ Rheumatology (immune disorders of the joints, muscles, and ligaments)
- ☐ Toxicology and environmental medicine (disorder related to toxin or environmental exposure)
- ☐ Urology

When approximately were the applicant's symptoms first noticed? If you are not sure about the month, take your best guess. _____

Does the applicant have any relatives with the same or similar symptoms?

- ☐ Yes ☐ No ☐ Unknown

If yes, please describe this family member's symptoms and how the family member is related to the applicant. If there is more than one relative with the same or similar symptoms, please indicate this.

Is there exposure to anything in the environment you think may have caused this undiagnosed condition? Examples could be hazards at work or around the home, an unusual diet, excessive drugs, poisoning, habits, hobbies, etc.

☐ Yes ☐ No

If yes, please describe.

For undiagnosed children who are applying: were there any exposures during the mother or father's lives that might have resulted in the child's current condition? Examples could be hazards at work or around the home, an unusual diet, excessive drugs, poisoning, habits, hobbies, etc. Were there any exposures the mother had during the pregnancy that might have resulted in the child's current condition? Were there any accidental or chronic exposures during infancy, childhood, or young adult life?

☐ Yes ☐ No

If yes, please describe.

Is the applicant currently involved in any other clinical trials?

☐ Yes ☐ No

Has the applicant previously been seen by health care providers at one of the following UDN clinical sites? Select all that apply.

- ☐ Baylor College of Medicine in Houston, TX
- ☐ Brigham and Women's Hospital, Boston Children's Hospital and the Massachusetts General Hospital in Boston, MA
- ☐ Children's Hospital of Philadelphia and University of Pennsylvania in Philadelphia, PA
- ☐ Duke Medical Center in Durham, NC

- ☐ Stanford Medical Center in Palo Alto, CA
- ☐ UCLA Medical Center in Los Angeles, CA
- ☐ Undiagnosed Diseases Program (UDP) at the National Institutes of Health (NIH) in Bethesda, MD
- ☐ University of Miami in Miami, FL
- ☐ University of Utah in Salt Lake City, UT
- ☐ University of Washington and Seattle Children's Hospital in Seattle, WA
- ☐ Vanderbilt Medical Center in Nashville, TN
- ☐ Washington University in St. Louis, MO

Please note: if the applicant participated in the NIH UDP prior to 9/15/2015, please email NIHUDN@nih.gov BEFORE submitting this application.

Applications are assigned to the UDN clinical sites based on site resources and location. If you would prefer to have your application reviewed by a specific site, please select that site from the list below. We will try to accommodate this preference but cannot guarantee that your application will be assigned to the selected site.

- ☐ Baylor College of Medicine in Houston, TX
- ☐ Brigham and Women's Hospital, Boston Children's Hospital and the Massachusetts General Hospital in Boston, MA
- ☐ Children's Hospital of Philadelphia and University of Pennsylvania in Philadelphia, PA
- ☐ Duke Medical Center in Durham, NC
- ☐ Stanford Medical Center in Palo Alto, CA
- ☐ UCLA Medical Center in Los Angeles, CA
- ☐ Undiagnosed Diseases Program (UDP) at the National Institutes of Health (NIH) in Bethesda, MD
- ☐ University of Miami in Miami, FL
- ☐ University of Utah in Salt Lake City, UT
- ☐ University of Washington and Seattle Children's Hospital in Seattle, WA
- ☐ Vanderbilt Medical Center in Nashville, TN
- ☐ Washington University in St. Louis, MO

Please explain why you want to be assigned to this site.

Does the applicant have any travel limitations we should know about? Select all that apply.

- | | |
|---|--|
| <input type="checkbox"/> None | <input type="checkbox"/> Wheelchair dependent |
| <input type="checkbox"/> Bed bound | <input type="checkbox"/> Partially ambulatory |
| <input type="checkbox"/> Permanently hospitalized | <input type="checkbox"/> Continuous intravenous infusion |
| <input type="checkbox"/> Ventilator dependent | <input type="checkbox"/> Other |

If other, please describe.

To your knowledge, have other family members of the applicant applied to the UDN or NIH UDP?

- ☐ Yes ☐ No ☐ Unknown

Please note: if the applicant or family members of the applicant applied to the NIH UDP prior to 9/15/2015, please email NIHUDN@nih.gov BEFORE submitting this application.

Does the applicant speak English?

- ☐ Yes ☐ No

If no, what is the applicant's preferred language? _____

How did you hear about the UDN?

Health Care Provider Contact Information

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

Health care provider email address: _____

Health care provider first name: _____

Health care provider last name: _____

Health care provider street address: _____

Health care provider apt/suite/floor (optional): _____

Health care provider city, state, zip code: _____

Health care provider country: _____

Health care provider phone number: _____

Health care provider fax number: _____

Attachments

Required attachment

Study recommendation letter: Please attach a study recommendation letter from one of the applicant's licensed health care providers. The letter should be on letterhead and signed by the health care provider. Please contact the UDN Coordinating Center (email: UDN@hms.harvard.edu, phone: 1-844-RINGUDN (1-844-746-4836)) to request an example study recommendation letter.

Optional attachments

Narrative summary: Applicants are welcome to provide a 1-page (500 words maximum) summary of the undiagnosed condition from their perspective. This narrative summary may be helpful for the UDN team to learn more about the applicant's experience with the undiagnosed condition. If you would like to provide a narrative summary, please attach it to this application.

Photo: Sometimes photos help us better understand the symptoms the applicant is experiencing. If you would like to provide a photo of the applicant, please attach it to this application. Up to 5 photos may be attached.