Paper Case Submission Process

Please email your completed case submission form and study recommendation letter to UDN@hms.harvard.edu for processing. If you do not have the ability to submit your case submission form, please call the UDN Helpdesk at 1-844-746-4836.

To submit your case online, go to **http://undiagnosed.hms.harvard.edu/apply**

If you have any questions, please contact the UDN Helpdesk:

Phone: 1-844-746-4836

Email: UDN@hms.harvard.edu

Are you the participant or legal guardian of the participant?

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

Name: _____

Relation to participant: _____

Directions

Please read the following consent form carefully. The consent form refers to the person submitting their case to the Undiagnosed Diseases Network (UDN) and "you" refers to the participant.

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.

While the UDN has access to state-of-the-art research resources to help diagnose patients, it is important to remember that the UDN is a research study. Participation in the UDN does not replace the long-term, longitudinal care you receive at home. Your care team at home will remain responsible for treatment and management.

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 submit a case 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. It may take you a couple of hours or many hours to collect and send your records to the UDN. Once you submit your case, the UDN may contact you and ask you to send more of your medical records. Medical information, including photographs, about you will be shared with health care providers in the UDN. The information will also be summarized using computer tools, including artificial intelligence that has been trained by UDN health care providers to review UDN information. These tools help health care providers in the UDN review your case.

There are different types of evaluations in the UDN. All cases submitted will undergo a medical records evaluation, and some will be selected to undergo additional UDN evaluations. To complete the medical records evaluation, we ask participants to send us a letter from a health care provider and medical records that describe their disease and medical test results. We also ask for the participant's name, contact information, and some additional information, like race and gender. If you submit a case and provide this information, we will keep this information. Keeping your information will allow us to evaluate our review process. We may publish some of this information in papers about the UDN. All these papers would discuss the UDN cases as a whole and would not include any of the information that could identify you, like your name or birth date. If you are assigned to additional UDN evaluations, you will be asked to sign a consent form for the entire study.

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

If you are assigned to additional UDN evaluations, you may need to spend several more hours obtaining additional medical records and speaking with the medical center staff. You may need to have a virtual visit with a UDN medical center via a telephone call or video

meeting. You may need to 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. During the evaluation, you would be looked at by multiple doctors. We would ask you questions about your health and the health of your family. After the evaluation, 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 submitting your case to the UDN?

The risks of submitting your case 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, including taking specific measures to protect your information when we use computer tools to review your information. Only people who have permission to view UDN information will have access to 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 assigned for additional evaluation in 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 submitting your case to the UDN?

Submitting your case to the UDN may not benefit you directly. You may receive a diagnosis if you are evaluated by 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 submitted your case to the UDN and will have your contact information. All the information that you share with the UDN during the case submission 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.

☐ 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.
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 Data Management 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 participants 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 assigned for additional evaluation by the UDN.

Participant Contact Information

Participant or parent/guardian email address: _____

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

Participant first name: _____

Participant last name: _____

Participant street address: _____

Participant apt/suite/floor (optional): _____

Participant city, state, zip code: _____

Participant county: _____

Participant country: _____

Participant phone number: _____

Participant's preferred language:

Please select the language that the participant is most comfortable using to communicate with UDN study staff.

Written communications (emails, letters): _____

Spoken communications (phone, in-person): _____

Parent/guardian name (if applicable): _____

Parent/guardian's preferred language (if applicable):

Please select the language that the parent/guardian is most comfortable using to communicate with UDN study staff.

Written communications (emails, letters): _____

Spoken communications (phone, in-person): _____

Participant Demographics

Date of birth: _____

Sex Assigned at Birth (select one): ☐ Male ☐ Female ☐ Neither

If neither, what sex was assigned at birth? _____

Gender Identity (select one):

How do you describe your gender identity? If your gender identity is not listed, select additional identity.

☐ Man/boy
☐ Transgender man/Trans man
☐ Non-binary
☐ Additional identity
☐ Woman/Girl
☐ Transgender woman/Trans woman
☐ Prefer not to say

If additional identity, what is the participant's gender identity?

_____

Race (please select all that apply):

☐ American Indian or Alaska Native
☐ Asian
☐ Black or African American
☐ Middle Eastern or North African
☐ Native Hawaiian or Other Pacific Islander
☐ White
☐ Other

If other, please explain: _____

If you would like to specify your race with more detail, please provide that here (optional):

_____
_____
_____
_____

Ethnicity (please select one of the following):

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

If you would like to specify your ethnicity with more detail, please provide that here (optional):

_____

_____
_____
_____

What kind of health insurance or health plan do you have now? Select all that apply.

- ☐ No health insurance or health plan
- ☐ Indian Health Service
- ☐ Medicaid, Medical Assistance (MA), CHIP or kid's state insurance
- ☐ Medicare
- ☐ Military health care (e.g., Tricare)
- ☐ Private health insurance (through a job, school, or bought directly)
- ☐ Insurance from outside the US
- ☐ Other

If other, please explain: _____

How many family members <u>under 18 years old</u> are currently living in your household?:

_____

How many family members <u>18 years or older</u> are currently living in your household?:

_____

Participant Symptoms

Category of Participant's Primary Symptoms

Please select the symptom category from the list shown below that BEST describes the participant's primary symptoms.

- ☐ Allergies and disorders of the immune system
- ☐ Cardiology and vascular conditions (heart, artery, vein, and lymph disorders)
- ☐ Dentistry and craniofacial (bones of the 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 (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)
- ☐ 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
☐ Other (please explain: _____)

When did the participant's symptoms start? If you are not sure about the month, take your best guess. _____

Does the participant 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 participant. If there is more than one relative with the same or similar symptoms, please indicate this.

_____
_____
_____
_____

Is there exposure to anything in the environment that 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.

_____
_____
_____
_____

Were there any exposures during the parents' lives that may have caused the participant's undiagnosed condition? Were there any accidental or chronic exposures during their infancy, childhood, or young adult life? Were there any exposures during pregnancy? 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.

_____

_____

_____

_____

Study Information

Is the participant currently involved in any other research studies such as a clinical trial?

☐ Yes ☐ No

Has the participant previously been seen by health care providers at one of the UDN clinical sites?

Visit https://undiagnosed.hms.harvard.edu/udn-sites/ for a current list of sites or contact the UDN Helpdesk (email: UDN@hms.harvard.edu phone: 1-844-746-4836).

☐ Yes ☐ No

If yes, which UDN clinical site was the participant previously seen at?

_____

_____

_____

May we automatically assign your case to one of the UDN clinical sites?

☐ Yes ☐ No

If no, please list the clinical site you would like your case to be assigned to.

Visit https://undiagnosed.hms.harvard.edu/udn-sites/ for a current list of sites or contact the UDN Helpdesk (email: UDN@hms.harvard.edu phone: 1-844-746-4836).

_____

Please explain why you want to be assigned to the site listed above.

_____

_____

_____

_____

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

- ☐ None
- ☐ Bed bound
- ☐ Permanently hospitalized
- ☐ Ventilator dependent
- ☐ Wheelchair dependent
- ☐ Partially ambulatory
- ☐ Continuous intravenous infusion
- ☐ Other

If other, please describe.

_____

_____

_____

To your knowledge, have other family members of the participant submitted their case to the UDN or NIH UDP?

☐ Yes ☐ No ☐ Unknown

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

How did you hear about the UDN?

- ☐ Advocacy/support organization
- ☐ Clinicaltrials.gov
- ☐ Conference
- ☐ Facebook
- ☐ Friend/family member
- ☐ GARD
- ☐ Genetic testing lab
- ☐ Internet
- ☐ News
- ☐ NIH
- ☐ NORD
- ☐ Online ad

☐ Provider
☐ Twitter
☐ Other

If other, please describe.

_____
_____
_____
_____

Provider Information

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

Health care provider first name: _____

Health care provider last name: _____

Health care provider email address: _____

Health care provider phone number: _____

Health care provider fax number: _____

Health care provider street address: _____

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

Health care provider city, state, zip code: _____

Health care provider country: _____

Household Demographics

This information will be used to support clinical evaluations and UDN research only.

What is the highest education level completed by someone in the participant's household?

☐ Less than high school
☐ High school graduate

☐ Some college or associate's degree
☐ Bachelor's degree
☐ Graduate or professional degree
☐ Prefer not to answer
☐ I don't know

What was the total income in the participant's household in the last calendar year? Please provide the best estimate of the total income of all family members living in the household.

☐ Less than $10,000
☐ $10,000 to $24,999
☐ $25,000 to $39,999
☐ $40,000 to $54,999
☐ $55,000 to $69,999
☐ $70,000 to $84,999
☐ $85,000 to $99,999
☐ $100,000 to $149,999
☐ $150,000 to $200,000
☐ More than $200,000
☐ Prefer not to answer

Is there any additional information about your household you feel is important to share or that you want the UDN team to know?

_____
_____
_____
_____

Attachments

Required attachment

Study recommendation letter: Please attach a study recommendation letter from one of the participant's healthcare providers (for example: specialist, primary care physician, nurse practitioner). The letter must be written in English, signed, and on letterhead. A relative cannot write the letter. Please contact the UDN Data Management 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: Participants are welcome to provide a summary (500 words maximum) of the undiagnosed condition from their perspective. This narrative summary may be helpful for the UDN team to learn more about the participant's experience with the undiagnosed condition. If you would like to provide a narrative summary, please attach it to this case submission.

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