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: ____

INTRODUCTION

This consent form describes a research study and is designed to help you decide if you would like to be a part of the research study. This study is taking place at the National Institutes of Health (NIH) and other sites across the United States and is called *Clinical and Genetic Evaluation of Patients with Undiagnosed Disorders Through the Undiagnosed Diseases Network* (UDN).

You are being asked to submit your medical records and other information about your health to see if you qualify for additional evaluation as part of the larger research study. Taking part in research at NIH or a participating site is your choice.

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.

If the individual being considered for enrollment is a minor then the term “you” refers to “you and/or your child” throughout the remainder of this document.

If the individual being considered to participate in this research study is not able to give consent for themselves, you, as the Legally Authorized Representative, will be their decision-maker and you are being asked to give permission for this person to be considered for this study. For the remainder of this document, the term “you” refers to you as the decision-maker and/or the individual being asked to participate in this research.

☐ I have read and understand this section.

ABOUT THIS 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 this 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 sex. 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 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 about updates of the testing.

In the future, we may re-contact UDN participants to ask if they are interested in participating in activities like surveys and interviews.

☐ I have read and understand this section.

RISKS AND BENEFITS

What are the risks and discomforts 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:* There is a potential risk of loss of privacy and confidentiality. Every effort will be made to protect your privacy and confidential information, but this cannot be guaranteed. 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 identifiable 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 submit your case to the UDN 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. You do not have to submit your case to the UDN if you do not want to.

What if I change my mind?

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

☐ I have read and understand this section.

PAYMENT

Will I receive payment for being in this study?

You will not receive any payment for submitting your case to the UDN.

☐ I have read and understand this section.

CONFIDENTIALITY

Confidentiality Protections Provided 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. We will do our best to make sure the information you share with us will be kept confidential, meaning no one outside of the UDN will have access to your responses.

Information gathered for this study is protected under a Certificate of Confidentiality and the Privacy Act.

Certificate of Confidentiality

To help us protect your privacy, NIH has a Certificate of Confidentiality (Certificate). With this Certificate, researchers may not release or use information about you except in certain cases. NIH researchers must not share information that may identify you in any legal proceedings, such as if a court requests it with a subpoena.

The Certificate does not protect your information when it:

1. is shared with people connected with the research. For example, information may be used for internal reviews by NIH; or
2. is required by law to be disclosed. For example, information may be shared with the FDA or with public health agencies.
3. is for other research if allowed by other regulations;
4. is shared with your consent.

Researchers may provide your information when you say it is okay. The Certificate does not keep you from sharing your own information.

The Certificate will not prevent telling authorities about harm to yourself or others. Examples are child abuse and neglect.

Privacy Act

The Privacy Act helps keep your NIH research information confidential. In some cases, it is different from the Certificate. Under 42 U.S.C. § 282, NIH is authorized to collect the data for this study. This data is covered by an NIH Privacy Act System of Records Notice: 09-25-0200, Clinical, Basic and Population-based Research Studies of the National Institutes of Health (NIH). Sometimes the Privacy Act allows sharing your information without your permission. An example is if Congress requests it.

Information may also be shared for some research. It can be given to some federal and state agencies. It can be used for HIV partner notification, or for infectious disease, abuse, or neglect reports. It may be shared with tumor registries, or for quality or medical reviews. It may also be shared if NIH is involved in a lawsuit or event of a suspected or confirmed breach. However, NIH will only release information about you if allowed by both the Certificate and the Privacy Act. If you do not want to share your information with us, then you cannot participate in this study.

☐ I have read and understand this section.

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)

☐ I have read and understand this section.

CONSENT DOCUMENT

Please keep a copy of this document in case you want to read it again.

☐ I have read and understand this consent form.

☐ 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 (select one): ☐ Male ☐ Female ☐ Neither

If neither, please describe: _________________

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):

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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.

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____
____

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.

____
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____

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.

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____
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____

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 <u>all</u> 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?

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____
____
____

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.