



Application form for UDNI Members (Clinicians)

Date of Application: _____

Organization: _____

Main Contact Name: _____

Main Contact e-mail: _____

The specific goals of the UDNI are to:

- (1) Improve the level of diagnosis and care for patients with undiagnosed diseases through the development of common protocols designed by a large community of investigators.
- (2) Facilitate research into the etiology of undiagnosed diseases, by collecting and sharing standardized, high-quality clinical and laboratory data, including genotyping, phenotyping, and documentation of environmental exposures.
- (3) Create an integrated and collaborative community across multiple clinical sites and among laboratory and clinical investigators prepared to investigate the pathophysiology of these newly recognized and rare diseases.

The UDNI is open to International Collaborators that agree to the criteria for participation described below.

Requirements for Online Application

Membership will be open to clinical investigators from all countries who serve undiagnosed disease patients. Members agree to adhere to the following principles:

- Patients enrolled in the UDNI should be selected for the unique characteristics of their disorder and for its potential to inform new aspects of cell biology, pathogenic mechanism(s), and therapy. Candidate patients should have been extensively examined already, so that obvious diagnoses have been eliminated.
- Accepted patients should be thoroughly evaluated by the UDNI collaborator, preferably at no cost to the patient.
- Patients should consent to share their data with other investigators within the group, according to the tenets of the Helsinki Declaration and/or of Good Clinical Practices. Patient phenotypes should be expressed using a standard ontology system in order to build up a highly integrated database.
- Next-generation sequencing and other -omics analyses (e.g., proteomics, glycomics, lipidomics) should be performed on enrolled families/patients (trios or quartets when possible), and analyzed with some uniformity and according to state-of-the-art protocols. New tools will be applied in some cases, including when a non-genetic undiagnosed disease (e.g., rare infections or poisonings) is suspected. Return of results will conform to site-specific consents.
- The -omics and phenotypic data should be shared among members of the UDNI.
- Functional studies should be performed to substantiate causal relationships between a candidate gene and the phenotype and address novel therapies.

Application Form should to include:

- a) The CV of the main clinical investigator (Annex 1)
- b) Description of the main activities performed by the organization (Annex 2)

Applications will be reviewed by the UDNI Board Members; a Membership Committee may be formed in the future.

Each Member should have 5 undiagnosed cases to contribute to the UDNI.

Acceptance *may* be limited to one-year, after which an assessment will be conducted for continuation.

Please return your application form, including CV (Annex 1) and the description of activities performed by the organization (Annex 2) to: UDNI@iss.it