

# The Undiagnosed Diseases Network International



[www.udninternational.org](http://www.udninternational.org)



## Principles

- Patients enrolled in the UDNI are selected for the unique characteristics of their disorder and for its potential to inform new aspects of cell biology, pathogenetic 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, 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, metabolomics) 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.
- 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.



## Background and Aims

The UDNI was established in Rome in 2014 during the first international scientific conferences and endorsed in 2015 during the second international meeting in Budapest **to provide diagnoses to patients, implement diagnostic tools and foster research on novel diseases and their mechanisms.**

The Network aims to fill the knowledge gaps that impede diagnosis, particularly for ultra-RD, and to foster the translation of research into medical practice, aided by active patient involvement. The specific aims of the UDNI are to work collaboratively and internationally to: i) provide diagnoses for patients that have eluded diagnosis by clinical experts; ii) contribute to standards of diagnosis by implementing additional diagnostic tools; iii) foster research into the etiology and pathogenesis of novel diseases; and iv) disseminate those research results broadly and rapidly.



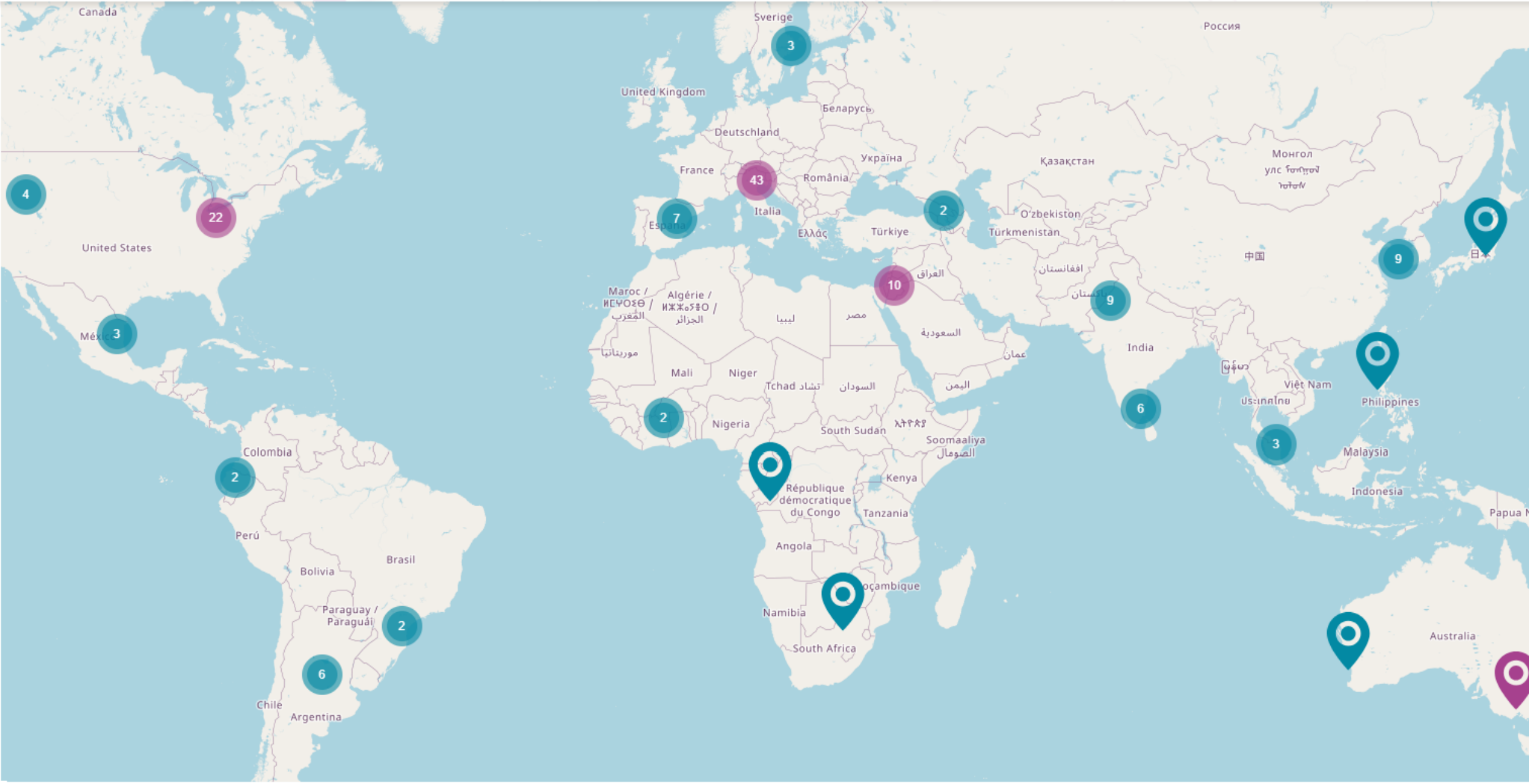
## UDNI Working Groups and Participating Countries

The UDNI involves centers with internationally recognized expertise, an international Governing Board, and several interacting Working Groups and Committees (see **table 1**). Active patient participation is achieved through the Patient Engagement Group comprising 19 patient's organizations (see **figure 1**). The UDNI now involves 41 countries (see figure 1) in all continents including *Argentina, Australia, Austria, Belgium, Brazil, Bulgaria, Canada, Chile, China, Ecuador, France, Georgia, Germany, Ghana, Hong Kong, Hungary, India, Israel, Italy, Japan, Korea, Kuwait, Mali, Malta, Mexico, New Zealand, Pakistan, Philippines, Saudi Arabia, Serbia, Singapore, South Africa, South Korea, Spain, Sri Lanka, Sweden, Switzerland, Thailand, The Netherlands, Turkey and USA.*

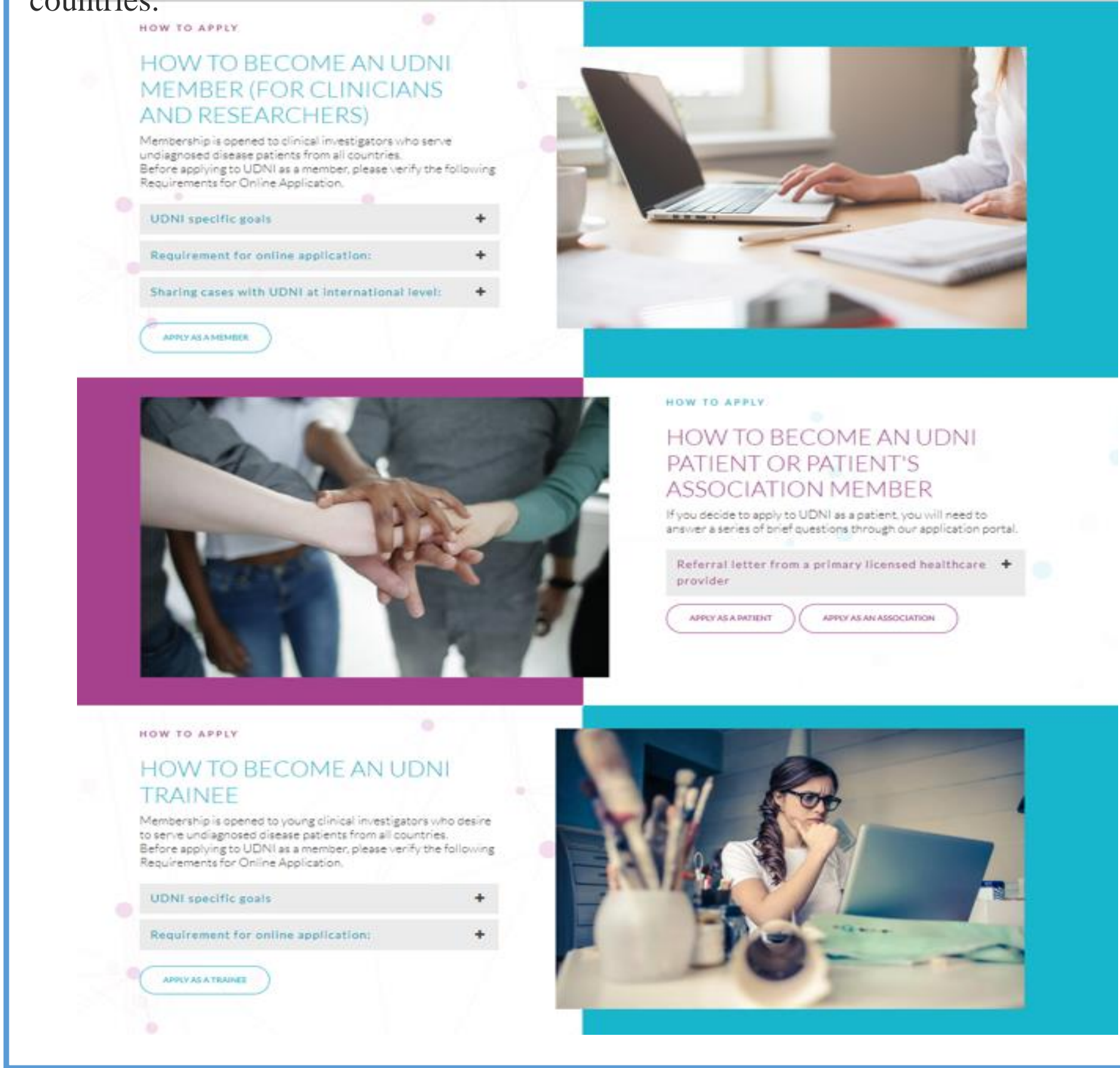
Table 1. UDNI Working Groups and Committees	In brief
<b>Program Committee</b>	Linking the Program to a scientific meeting is considered beneficial, in particular to justify participant travel. The UDNI has no objections to industry participation in its general meetings.
<b>Communication Group</b>	Coordinating UDNI information transfer. Recommendations for the current website include adding a patient section, changing the membership requirements on the website to conform to the non-clinician categories, providing information on how to apply to the UDNI, adding members' profiles and photos, and posting slide sets about the UDNI.
<b>Membership Committee</b>	Reviewing all applications for membership and accepting new members on a rolling basis. On a regular basis all applicants who meet the membership criteria are presented to the full membership for approval.
<b>Functional Study Group</b>	To gather the latest knowledge and publicly available resources related to functional studies, organize such information in simplified and accessible manner, disseminate useful information to all stakeholders, and facilitate collaborations between clinical and basic researchers world-wide.
<b>Patient Engagement Group</b>	To provide a patient perspective to issues like data sharing and to offer clarity and transparency. This requires mutual trust and involves the presence of a patient representative on the Governing Board.
<b>Genetic Counseling Group</b>	To address the genetic counseling needs of patients and their families who participate in an Undiagnosed Diseases Program.
<b>Educational Group</b>	To address the educational needs of the scientific community involved in undiagnosed diseases.
<b>Developing Countries Group</b>	To address the issues (difficulties and opportunities) for providing diagnoses to patients with undiagnosed and rare diseases in Developing Countries; to cooperate with national experts in the achievement of new diagnoses; to build local capacity so that correct diagnosis becomes the standard of care in these settings.
<b>Diagnostic Working Group</b>	To provide support and an opinion to local Undiagnosed Disease Programs to accelerate diagnoses for more families, support developing undiagnosed disease programs particularly in lower resource areas and share knowledge and skills, promote discovery and expand global medical knowledge.

**Figure 1.** UDNI interactive map shows the 50 Countries, 170 UDNI members, 19 Patients Associations participating in the Network activities

## COUNTRIES PARTICIPATING TO THE UDNI PROJECT



Clinician, patient association and trainees can apply to become a UDNI member. Membership is open to everyone who serves undiagnosed disease patients from all countries.



## UDNI Patients Area

The UDNI together with the Wilhelm Foundation offer undiagnosed patients (adults and children), who have been evaluated at a UDP and still are undiagnosed to share their story. Patients who are undiagnosed can use this chance to show all the UDNI website visitors their medical history and photos. Many of the UDNI specialists may help patients in finalizing their diagnosis.

This area is under the Wilhelm Foundation responsibility. In this section Wilhelm Foundation will collect some information (phenotypic, genotypic, picture) from undiagnosed rare patients. For this purpose a dedicated ethics-Informed Assent as well as a UDNI Parental Consent were developed and approved by all UDNI Board Members and participants. Before adding information on a patient these consents have to be downloaded and signed by the Responsible of patient.

Patients must have undergone a specific visit in a dedicated undiagnosed clinical site before being added in the present section.

## ATTACHMENTS (available at [www.udninternational.org/](http://www.udninternational.org/))

- **Ethic informed Assent**
- **Wilhelm Foundation Privacy**
- **International consent for publicaton of patient image**

Please, visit our web site to find out more information: <https://www.udninternational.org/>

## References

Undiagnosed Diseases Network International (UDNI): White paper for global actions to meet patient needs. Mol. Genet. Metab. (2015), <http://dx.doi.org/10.1016/j.ymgme.2015.11.003>

The Undiagnosed Diseases Network International: Five years and more! Mol Genet Metab. 2020;129(4):243-254. doi: 10.1016/j.ymgme.2020.01.004.

Statement: consent has been provided for the publication of patient's photographs. The personal data, processed through the production and subsequent publication on the website [www.udninternational.org](http://www.udninternational.org) of photographic surveys of the interested parties, are collected exclusively for purposes of study and scientific research as well as for the assistance of Rare Undiagnosed Patients with the aim to disseminate the photos among researchers around the world in order to help those suffering from a rare undiagnosed disease to find a diagnosis.



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