Background

To address unmet needs of undiagnosed patients at global level, an international network was formed: the Undiagnosed Diseases Network International (UDNI). The UDNI was established following two International Conferences (Rome, September 29-30, 2014 and Budapest, June 26-27 2015) sponsored by the Common Fund, within the Office of the NIH Director, along with the Wilhelm Foundation, Sweden.

Principles

1. 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, pathogenetic mechanism(s) and therapy. Candidate patients should have been extensively examined already, so that obvious diagnoses have been eliminated.

2. Accepted patients should be thoroughly evaluated by the UDNI, preferably at no cost to the patient.

3. 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.

3. 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.

4. The -omics and phenotypic data should be shared among members of the UDNI.

5. Functional studies should be performed to substantiate causal relationships between a candidate gene and the phenotype and address novel therapies.

Practices

The **UDNI** will adhere to the following best practices.

1. Applications will be specific to member sites, but should include core information described in Appendix A.
2. Clinical site evaluations will be comprehensive and include clinical and basic research approaches, including specimen collection for future studies, as described in Appendix B.
3. A list of clinical experts will be created for advice and referral both within the **UDNI** and outside of it.
4. A list of basic research topic experts will be compiled to serve as a potential collaborator pool; de-identified cases can be shared with these authorities if they have variants in genes within a particular basic researcher's area of expertise.
5. The **UDNI** will maintain a website with information for physicians and patients.
6. The NIH UDN will make available its Manual of Operations to the UDNI.

Board members

The initial Board of Directors will consist of Gareth Baynam (Australia), William Gahl (USA), Stephen Groft (USA), Kenjiro Kosaki (Japan), Paul Lasko (Canada), Bela Melegh (Hungary), and Domenica Taruscio (Italy). Half the board members are expected to be replaced every two years. Board members can apply for re-election

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

Contact us

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References

D. Taruscio, et al., 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