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Position paper of the undiagnosed diseases network international with respect to the establishment of the journal *Rare-Open research in rare diseases*

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The Undiagnosed Diseases Network International (UDNI) welcomes the entrance of a new journal, *Rare-Open research in rare diseases*, into the rare disease community. The primary aspiration of both the UDNI and the journal is that the relationship of the two entities will evolve into a synergistic collaboration to advance awareness, investigation, and treatment of rare and undiagnosed diseases.

The UDNI, modeled after the USA's National Institutes of Health (NIH) Undiagnosed Diseases Program (UDP) [1,2] and Undiagnosed Diseases Network (UDN) [3,4], was established in 2014 with the support of the NIH Common Fund and the Wilhelm Foundation, an advocacy group for rare and undiagnosed disorders based in Sweden. The UDNI has held 11 international conferences from which have emanated a charter, best principles and practices, a Board of Directors, several Committees (Membership, Program, Communications), and a handful of Working Groups (Data Sharing, Functional Studies, Patient Engagement, Diagnostics, Developing Nations). A website is managed out of the Istituto Superiore di Sanità / Italian National Institute of Health in Rome (https://www.udninternational.org/). The establishment of the UDNI was announced in a 2015 White Paper [5] and a 5-year follow-up was published in 2020 [6]. Current initiatives include the Diagnostic Working Group's effort to provide a forum for discussion of undiagnosed cases that lack access to a UDP and a Developing Nations Working Group program to connect inchoate UDPs in Low- and Middle-Income Countries with other UDNI members' programs. Specifically, the Champions Initiative, a joint effort of the UDNI and the Wilhelm Foundation, has already identified local leaders in four nations, i.e., Mali, Democratic Republic of Congo, Ghana, and Pakistan to serve as examples of how the UDP concept can be globally expanded.

The UDNI can contribute to several of the new journal's aims. Network members have extensive expertise in genetic variant analysis, new DNA technologies, and clinical biochemical testing. As founders and leaders of undiagnosed diseases programs, they are at the forefront of rare disease diagnostics (Diagnostic Working Group), phenotypic expansions, new disease discovery, disease mechanism investigations, and gene function studies (Functional Studies Working Group). The UDNI's reach is international (Developing Nations Working Group) and patient-oriented (Patient Engagement Working Group). The UDNI now has more than 130 members representing over 40 nations throughout the world. The Network is dedicated to implementing the principles recently elucidated by the United Nations General Assembly in its Resolution on People Living with Rare Diseases (Addressing the Challenges of Persons Living with a Rare Disease and their Families) [7] recommending international collaborations to leave no person with a rare disease behind in the development of national legislation, regulations, policies, and responses to these actions and avoid limitations preventing individuals from reaching their full potential. The Primary mechanism for achieving this goal consists of sharing data, expertise, and resources. The UDNI now hopes to work with *Rare-Open research in rare diseases* to broadly disseminate information about rare and undiagnosed diseases and to implement the UDNI goals.

UDNI Governing Board (Olaf Bodamer, Helene Cederroth, William Gahl, Roberto Giugliani, Eric Klee, Manuel Posada De La Paz, Olaf Horst Riess, Domenica Taruscio).

Declaration of Competing Interest

The authors declare that they have no competing interests.

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