

GeNeEvent - The Seventh International Conference on Rare and Undiagnosed Diseases

The Seventh International Conference on Rare and Undiagnosed Diseases was hosted at Delhi NCR from April 13th – 15th 2019. It was the official meeting of the Undiagnosed Diseases Network International (UDNI), in conjunction with an initial meeting to address patient needs for Rare Disorders in India. It was a collaborative meeting of the National Institutes of Health Undiagnosed Diseases Network (NIH UDN), Institute of Medical Genetics and Genomics, Sir Ganga Ram Hospital New Delhi, Organization of Rare Diseases India (ORDI), CSIR-Institute of Genomics and Integrative Biology (IGIB), and the Wilhelm Foundation, Sweden.

The meeting highlighted the current status of diagnosis of rare disorders, newborn screening, research collaborations, and patient groups and their importance in the arena of rare disorders. The inaugural talk on the vision and way forward of the UDNI was delivered by Dr William Gahl, Director of the NIH UDN. A unique aspect of this conference was its linking with the meeting of the UDNI. The UDNI network (<http://www.udninternational.org/>), of which India is also a member, is concerned with diagnosis and research related to patients who remain undiagnosed despite extensive clinical evaluation. The program aims to work collaboratively and at an international scale for patients who do not get a diagnosis despite expert evaluation. The purpose of the discussion within the UDNI agenda was to solidify plans to share data as a consortium of international undiagnosed diseases programs, to promote the diagnosis of rare diseases and the discovery of new diseases, and to foster the elucidation of new mechanisms and pathways. To this effect diagnosed and undiagnosed cases were presented to elicit suggestions for diagnosis. The undiagnosed cases are required to be submitted to Phenome Central keeping with the charter of the UDNI.

The meeting was well attended, with more than 250 registrations, by students, young scientists, and senior and young geneticists of the country. Many international experts working in the area of rare disorders participated in the conference. The meeting concluded on a very positive note with a hope to enhance efforts for diagnosis and treatment of patients with rare, undiagnosed disorders in India.

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