



**UDNI**  
Undiagnosed  
Diseases Network  
INTERNATIONAL

## UNDIAGNOSED RARE DISEASES

Undiagnosed rare diseases represent one of the most complex challenges in modern medicine. Although each rare disease affects a small number of patients, collectively rare diseases impact millions of people worldwide. A significant proportion of affected individuals remain without a diagnosis, even after years of medical consultations and clinical tests.

This prolonged and uncertain journey is often referred to as the “diagnostic odyssey,” a process that may span several years and is frequently characterized by misdiagnoses, delayed care, and considerable emotional strain for both patients and their families.

One of the main difficulties lies in the rarity and variability of symptoms. Many conditions present with overlapping or nonspecific signs, making them hard to distinguish from more common diseases and also other rare diseases, which increases the number of misdiagnoses. In addition, limited access to advanced diagnostic tools, such as genomic sequencing, and a lack of specialized expertise can further delay or prevent accurate identification.

Even the most advanced genetic tests can provide answers for approximately 4 in 10 patients leaving the majority still searching for a diagnosis often for years, decades, or a lifetime, therefore the 60% remain People Living with Undiagnosed Diseases (PLWUD).

For these individuals, the absence of a diagnosis can result in years of inappropriate treatment, limited access to support services, and a profound sense of invisibility within the healthcare system. Without a diagnosis, patients often struggle to receive appropriate care, obtain the support they need, or gain recognition of their condition. In many cases, this situation can persist for years, decades, or even a lifetime.

The Undiagnosed Diseases Network International (UDNI, <https://www.udninternational.org>) was established in 2014 to address these challenges on a global scale.

The UDNI is an international network of clinicians, researchers and patients’ representatives working together to improve the diagnosis and understanding of rare and undiagnosed conditions. By connecting expert centers across different countries, the UDNI facilitates the sharing of knowledge, data, and advanced technologies.

A key strength of the UDNI is its collaborative and multidisciplinary approach. Complex patient cases are reviewed by teams of specialists from various fields, increasing the likelihood of identifying underlying causes. Moreover, the network contributes to scientific discovery by identifying new diseases and expanding knowledge of known conditions.

In conclusion, undiagnosed rare diseases highlight important gaps in healthcare systems and the UDNI represents a promising effort to bridge these gaps through international cooperation, research, and innovation. Raising awareness, advancing research, and advocating for equitable access to diagnostic tools are tangible actions each of us can take to help bring an end to the diagnostic odyssey faced by millions of people worldwide.