

LETTER TO THE EDITOR

Patient Voices on Diagnostic Research

Undiagnosed Diseases Network Foundation Participant Engagement and Empowerment Resource

The October 2025 issue of the *AMA Journal of Ethics* explored the ethics of diagnostic research, focusing heavily on the National Institutes of Health's Undiagnosed Diseases Network (UDN). One notable absence was the patient voice. We write as the UDN's affiliated patient advisory board, UDN Foundation Participant Engagement and Empowerment Resource (UDNF PEER). We are patients and caregivers living the diagnostic journeys examined in those articles. While we appreciate the emphasis on training students to care for patients thoughtfully and empathetically with undiagnosed conditions,¹ articles that called into question the value, ethics, and utility of diagnostic research did not reflect our experience with the UDN.

Some articles speculated about the value of participation without resulting treatments.^{2,3} For us, the UDN's value was not limited to a diagnosis or treatment. While some UDNF PEER members have found clear answers in the form of a diagnosis or treatment, others have not. But we benefitted from individualized attention to and comprehensive evaluation of our cases. Because of the UDN's benefit to us and our families, we want clinicians who are considering UDN referrals to understand its value.

Ethical concerns raised about informed consent did not reflect our experiences.^{2,3} Each of us clearly understood that the UDN is a research program, a diagnosis was not guaranteed, and we could withdraw at any time. We knew that the odds are stacked against us. While we hope for breakthroughs in our cases or our children's cases, we also hope that the UDN's findings help others.

Most concerning is one article's call to end the UDN's public funding.³ While the article raises interesting points about program access,³ we believe that increasing opportunities is a better answer than eliminating public programs. Critiques of the UDN's complexity and cost should also consider that the UDN's NIH funding for under- and uninsured patients translates to cost savings for the private health care system.⁴ Ending public funding would dismantle the only national model offering such coordinated investigation. Having walked this road, our hope is that the next patient who could benefit from the UDN's model has more opportunities to participate—not fewer.

We cannot speak for every participant; however, diagnostic research has benefited us as patients and caregivers, even those of us without diagnoses. As Hall et al write in this

issue, medical professionals' "duty of care is independent of the patient having an established diagnosis."⁵ We agree. When there is no standard of care for a patient's condition or symptoms, the availability of diagnostic research options is critical. Each discovery expands medical knowledge, informs future clinical practice, and shortens the diagnostic odyssey for others.

References

1. Massart MB, Dreikhorn EN. How should students and trainees be taught to support patients with undiagnosed conditions? *AMA J Ethics*. 2025;27(10):E733-E736.
2. Doyle TA, Conboy E. What are ethical merits and drawbacks of viewing "medical mysteries" as human subject research? *AMA J Ethics*. 2025;27(10):E718-E725.
3. Gordon G, Kearns L. Is the UDN N-of-1 enterprise ethically justifiable? *AMA J Ethics*. 2025;27(10):E737-E742.
4. Yang G, Cintina I, Pariser A, Oehrlein E, Sullivan J, Kennedy A. The national economic burden of rare disease in the United States in 2019. *Orphanet J Rare Dis*. 2022;17:163.
5. Hall, A, Webb, BD, Meyn, MS. What is the nature and scope of physicians' duties of care to patients without a diagnosis? *AMA J Ethics*. 2025;27(10):E726-E732.

Undiagnosed Diseases Network Foundation Participant Engagement and Empowerment Resource is an 11-member patient advisory group that advises Undiagnosed Diseases Network leadership and site teams on patient experiences and publishes materials to support the Undiagnosed Diseases Network Foundation mission of improving access to diagnosis, research, and care for all individuals living with ultra rare or undiagnosed conditions. More information about the Undiagnosed Diseases Network Foundation Participant Engagement and Empowerment Resource is available [here](#).

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