The U.S. has 25 million overlooked rare disease sufferers. Here's how to change that.

By Doug Lindsay,
Co-Chair, Rare Disease Advisory Panel
For the Patient-Centered Outcomes Research Institute (PCORI)

The U.S. Centers for Disease Control say 25 million Americans suffer from a rare disease. I am one of them. There are 7,000 identified rare diseases and 95% of them have no approved treatment.

Rare disease is an overlooked world of sadness and despair. The way the National Institutes of Health funds research leaves most rare disease sufferers with no hope for an effective treatment in our lifetimes.

Dr. Chris Austin, former head of NCATS (National Center for Advancing Translational Sciences), says at the current rate it would take another 2,000 years for each of these diseases to get a treatment. Most stats associated with rare disease are this bleak.

The average rare disease patient spends seven years just trying to get a diagnosis. For my mother Barbara and Aunt Jeanne, it took nearly 30 years each. Ravaged by muscle disease and homebound for much of their adult lives, they got diagnosed only after I uncovered what was wrong with me, and only after I presented at an international medical conference a proposed treatment for my own condition – a rare, heritable autonomic-adrenal problem.

I got sick at 21. I was a disabled, bedbound college dropout with no medical training, but when I realized no one was coming to rescue me or my family, I worked tirelessly to do it myself. During those 14 years sick, 11 of them homebound, I developed new uses for five existing prescription drugs and developed the surgery doctors used to fix me.

No rare disease sufferer should have to do what I did just to have a treatment.

In years past, we've lacked tools to do dramatically better for rare disease patients. Today, however, we don't.

Some ways to change rare disease research's trajectory are complex. But one way is simple, powerful, and free: Within healthcare we need to explicitly identify "rare disease" as an equity group.

In 2010, The U.S. Department of Health and Human Services (HHS) released a report called "Healthy People 2020" listing six equity groups for whom they sought to rectify health

disparities: Race/ethnicity; Gender; Socioeconomic status; Disability status; Lesbian, gay, bisexual, and transgender status; and Geography.

One cannot miss that these communities have been prominent cultural conversation topics in the intervening years. Neither "Healthy People 2020" nor "Healthy People 2030" lists rare disease as an equity group. But to change things for rare disease sufferers, our government should designate us an equity group now. Waiting ten years for "Healthy People 2040" to do it is not an answer.

I'm Co-Chair of a Congressionally-mandated Rare Disease Advisory Panel. It's part of an institution called the Patient-Centered Outcomes Research Institute (PCORI).

Obamacare created PCORI, and PCORI's given out $2.4 billion in research grants since 2010. PCORI funds research to help doctors and patients make treatment decisions. We put patients and patient outcomes at the center of our focus and then fund studies accordingly.

Yet even within PCORI, with its rare disease panel Congressionally mandated, rare disease is the odd man out. Just 5% of PCORI's grant dollars have gone to rare disease studies.

If we at PCORI and the leadership at HHS and NIH explicitly identify rare disease as an equity group, then common realities like seven-year diagnostic journeys and 19 of 20 rare conditions without an approved treatment become health disparities. And if rare disease outcomes are an acknowledged health disparity, then we have a way to talk about it and a way to pursue changing it.

These days HHS, NIH, PCORI, and major hospitals and health systems are all aiming to remedy health disparities. Without this designation, those efforts will not even intend to help rare disease patients.

The disabled are an acknowledged equity group, and many rare disease sufferers are disabled by their conditions. Yet we rare disease sufferers are seeking research, treatments, and cures – healthcare solutions – not merely better access to a world still devoid of effective treatments for us.

Any progress will come too late to help my mother Barbara. She passed away in 2016, too sick to undergo the surgery that gave me my life back. But millions of others are still waiting for our help.

The path to designating rare disease outcomes as a health disparity is clear. In an October 2016 release, the director of the National Institute of Minority Health and Health Disparities (NIMHD) explained: He has the power to define a health disparity population in consultation with the director of HHS's Agency for Healthcare Research and Quality (AHRQ).

So, I call on PCORI, along with HHS and NIMHD officials to serve the American people by explicitly defining rare disease sufferers as an equity group within healthcare. This is how we make the invisible problem of rare disease visible and change the future of rare disease research to help 25 million Americans.

(799 words)

Corroboration of claims made:

1. CDC 25,000,000 stat (pulled quote): https://blogs.cdc.gov/genomics/2019/04/04/introducing-the-rare-diseases/
    a. NIH confirms with 25-30 million range: https://rarediseases.info.nih.gov/diseases/pages/31/faqs-about-rare-diseases
2. 7000 identified rare diseases (paragraph 3): https://nihrecord.nih.gov/2021/04/16/rare-diseases-are-public-health-issue
3. 95% with no approved treatment (paragraph 3): https://www.thelancet.com/journals/landia/article/PIIS2213-8587(19)30006-3/fulltext
4. Dr. Austin says 2000 years to get treatments at this rate (paragraph 4): https://nihrecord.nih.gov/2021/04/16/rare-diseases-are-public-health-issue
5. 7 year diagnostic journey (paragraph 3): https://www.thelancet.com/journals/landia/article/PIIS2213-8587(19)30006-3/fulltext
6. Image of my abstract from Clinical Autonomic Research, from the conference proceedings of the American Autonomic Society's 2002 meeting: attached as PDF
7. CNN story on my medical journey and surgery: https://www.cnn.com/2019/07/27/health/doug-lindsay-invented-surgery-trnd/index.html
8. "Healthy People 2010": https://web.archive.org/web/20101219081349/http://healthypeople.gov/Document/html/uih/uih_2.htm
9. Healthy People 2020: https://www.healthypeople.gov/sites/default/files/HP2020_brochure_with_LHI_508_FNL.pdf
10. Healthy People 2030: https://health.gov/healthypeople/objectives-and-data/browse-objectives#populations
11. Co-Chair of RDAP, Rare Disease Advisory Panel: https://www.pcori.org/people/doug-lindsay
12. Slide from RDAP's public meeting in December 2021 on $2.4 Billion: attached
13. Slide on rare disease portfolio and funding total (roughly 5% of the 2.4 billion), also December 2021: attached
14. Link to PCORI: https://www.pcori.org

15. Link to the NIH's National Institute on Minority Health and Health Disparities: https://www.nimhd.nih.gov
16. Link to Director's Message quoted in the piece: https://www.nimhd.nih.gov/about/directors-corner/messages/message_10-06-16.html
17. Article on my public health and translational research advising work with Washington University in St. Louis: https://icts.wustl.edu/icts-community-advisory-board-co-chair-featured-in-people-magazine-regarding-rare-disease-journey/?utm_source=rss&utm_medium=rss&utm_campaign=icts-community-advisory-board-co-chair-featured-in-people-magazine-regarding-rare-disease-journey