Wall Street Journal: ***Hard Choices in Pursuit of Rare-Disease Cures; Patients Urge More Data Gathering as Tighter Federal Budget Crimps Research***
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Sarah Kucharski has been diagnosed with fibromuscular dysplasia and is among patients seeking more rare-disorder research. Brooks Kraft for The Wall Street Journal

But first, they wanted to deliver a petition, and a message, to Francis Collins, the NIH's director.

Late last year, the National Institute on Aging, which is part of NIH, said a long-running observational study of fibromuscular dysplasia and four other rare diseases was no longer collecting data or enrolling patients, and that the study's goals had been met. Sufferers, arguing that fibromuscular dysplasia's cause or cure still isn't known, mobilized. One participant in the research, Kari Ulrich, set up an online petition that when printed out was a phonebook-size document containing names and comments of about 11,000 people.

"It's heavy," Ms. Kucharski said, fanning the pages. The 33-year-old, who lives in Canton, N.C., founded a patient-advocacy group in 2011 after her diagnosis. She had a stroke at 28, an event that might be connected to her underlying condition. Today, there is no obvious physical sign of her ordeal. "Nobody knows unless I tell them," she said. That is part of the problem with drawing attention to the disorder, she said: "Patients usually look fine."

The petition, which covers all five rare diseases in the study, is part of a broader effort by patients who don't want to see the data collection stopped. Patients have spoken to their representatives in Congress, reached out to rare-disease advocacy groups and taken the cause to social media. NIA officials have sought to reassure patients that the data collected will be put to use.

The fight to continue the study exemplifies tensions that often arise between researchers and patients over which efforts yield the most valuable science. The research in this case is a "natural history" study, intended to document the progress of little-understood diseases but not necessarily to discover treatments. Both NIH and the Food and Drug Administration have said such studies are a critical early step toward drug development. But the studies are expensive and don't always lead to new trials, and they are harder to sustain at a time of budget cuts when difficult choices must be made.

Luigi Ferrucci, scientific director of NIA, said he led an assessment of the agency's research program that looked at many factors, including a study's relevance to NIA's mandate to study aging. At the time, there were 49 active clinical studies approved at the NIA program for research done by or at the institute. That is now down to 18, a reflection of budget constraints, among other concerns.

"In a way, it is an issue in every observational study," Dr. Ferrucci said. "When do you stop? At some point, the data you collected needs to be analyzed and made into science." He noted that when it comes to data collection, "If you don't stop, you don't have the resources to study anything else."

It isn't known how many people suffer from fibromuscular dysplasia, which is said to cause one or more arteries in the body to have abnormal cell development in the artery wall. The goal of the NIA study—which began in 2003 and focuses on five disorders including Ehlers-Danlos and Marfan syndromes—was to collect data, imaging and biosamples for the disorders from as many as 1,385 participants, create a repository where samples could be analyzed, and follow some patients every five to 10 years to learn about the course of the diseases. No end date was set.

Emil Kakkis, who is using natural-history information in a different disease to guide drug development at Ultragenyx Pharmaceutical Inc., where he is chief executive, said there is always competition for resources and that it is natural that common diseases that generate high public awareness garner more funds. He said that if data collection needs to stop, steps should be taken so that "the data collected so far is not lost."

The NIA's Dr. Ferrucci said information from about 1,000 participants had been gathered at the time data collection was halted. About $200,000 in funding will be used to help manage the existing information.

At the Rare Disease Day event, Ms. Kucharski and Ms. Saplis, 54, a former nurse, corralled NIH's Dr. Collins after his remarks. "Wow," he said when they handed him the 2-inch-thick binder of signatures. He lauded their advocacy and said he would consider the issue.

Later, Dr. Collins said he takes the concerns seriously. "Patients are our most important partners in research," he said in an email, while citing constraints caused by the budget squeeze. NIA said it hopes to begin meeting with patients and their representatives to address their concerns.

Afterward, the women called Ms. Ulrich, 46, who was too ill to travel and present the petition herself. When she heard that Dr. Collins got the document, she cried.

Then the women headed back to their brochures and pens, one more rare disease vying for attention. "This is one study," Ms. Kucharski said, as visitors stopped by, "but there are implications for patients who might participate in studies in the future. If we give data and nothing happens, what is the point?"