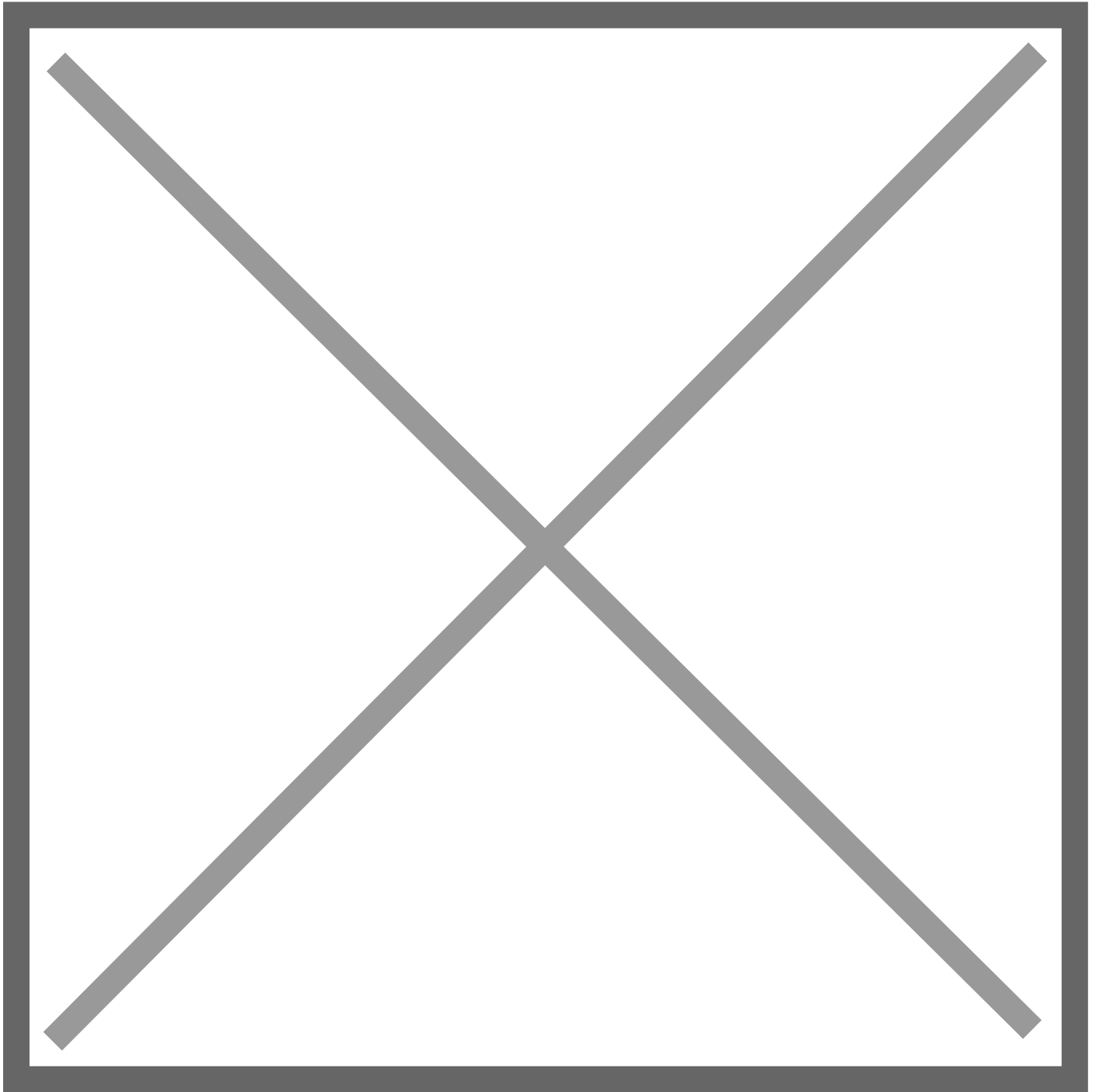


Pfizer Logo

Empowering People with ‘Super Genes’ to Help Unlock New Treatment Pathways

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Living among us today are people who carry “super genes” with variants that make them resilient to certain diseases. Some successful medicines mimic these genes, such as a new bone-builder that is based on families with genetically strong bones. If scientists could find patients with genetic resilience, to say, advanced cancer or [liver disease](#), studying these individuals could unlock novel treatments for some of the most critical conditions of our time. But until recently, finding these research participants has been like searching for a needle in a complex and highly regulated haystack.

But now, thanks to a collaboration among Pfizer; Inspire, a social network for health; the Manton Center for Orphan Disease Research at Boston Children’s Hospital; and Citizen Genetics, a genetics research company; locating people with extremely rare genetic variations just got a lot faster. The initiative known as Patient Forward Access to Clinical and Technological Research (PFACTR), recently launched its first study looking for people who showed exceptional resilience to [lung cancer](#) in families with a history of autoimmune disease. Researchers want to explore whether certain genetic variants may have helped them survive cancer, and in a second line of inquiry, whether genetic variants may drive extreme autoimmune-related symptoms.

Normally, discovering a small cohort or one or two families who meet such criteria might take years — if you could find them at all. But with the Inspire platform, which includes nearly two million members and volunteers, researchers were able to identify over a hundred participants who fit the criteria in a few weeks. “This novel partnership enables us to proactively find families who are possibly carrying very rare and very interesting genetic variants. And it lets us do it quickly, yet while working within an appropriate clinical regulatory pathway,” says Stefan McDonough, Executive Director of Genetics, based at Pfizer’s Kendall Square Cambridge, Mass. research site. “It’s these very rare genetics that can show how to have a big effect on people’s health and in some ways are most useful to develop drug targets.”

Enabling research opportunities

A key challenge in finding study participants is that most people with protective genetic variants don’t even know they have them. If a variant is protecting you from illness, it means you’ve had an unexpectedly good prognosis, or perhaps aren’t even getting sick despite having risk factors. In these cases, you’re unlikely to get a genetic analysis, since your health is good on its own. Finding people who are resilient to disease by combing through medical records offers limited data and often only a single snapshot in time.

The Inspire platform provides an opportunity to address this challenge. The social media network has over two million members affected by serious health conditions, including [autoimmune disorders](#), cancers and rare diseases. These patients and caregivers can give detailed descriptions of their health and patient journeys. Members have control over their health information and privacy, and can consent to participate in research. And by being able to quickly sort for patients that meet certain criteria, researchers are able quickly able to screen for these rare genes. “Enrolling patients with rare diseases and conditions is something we do very efficiently,” said Alan Beggs, Director of the Manton Center. “But finding them when you are based at just a single hospital is a bigger challenge. Ascertaining rare families through the Inspire community provides a transformational opportunity to scale up the process to find the very rarest of the rare.”

And perhaps most important, members are engaged in learning more about their health and forming patient communities. “You have a combination of a large population and motivated patients, making for a powerful environment to help patients through their health journeys,” says Brian Loew, CEO of Inspire. “When you invite patients to help enable medical progress, a lot becomes possible.”

Being patient-forward

For the current lung cancer study, members who meet the criteria are invited to participate — and upon their consent — are referred to Boston Children’s Hospital’s Manton Center and Citizen Genetics, which are jointly conducting the virtual study under clinical trial guidelines. Participants submit a saliva sample for genetic analysis, and some may have phone interviews with investigators to gather additional health information. All participants are given the option to receive a full report of their genetic data and have access to a certified genetics counselor through Boston Children’s Hospital.

Another factor driving this novel research model is the recent advances made in [genomic technology](#), says Joe Monforte, a principal at Citizen Genetics, who together with Pfizer researchers is conducting the genetic analysis. If a genetic mutation is indeed driving a large effect, with new tools scientists have the ability to almost always identify the causal gene that's driving a protective mechanism or a disease. "We've reached a new plateau of performance with regard to genetic analysis," says Monforte. "It changes the whole dynamic of how we interpret genetic data and use these insights for developing potential therapies."

And with the growing numbers of active patients who want to participate in research and help contribute to new treatments for their conditions, the initiative partners hope to continue this study model going forward. "We're really trying to be patient forward and give back to them as much as we can, as we drive new therapies for their conditions," says Monforte.

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