

## Efforts to cure rare genetic diseases target people who conquered them

*Biologist Stephen Friend is president and co-founder of the nonprofit research organization Sage Bionetworks in Seattle, and co-director of the Resilience Project, which starts in September. He is searching for exceptional people who carry genes for serious childhood disease but have never gotten sick—are you one of them?*

### **Why do you want to find people who have genes for serious diseases?**

We aim to find relatively healthy adults who have somehow escaped having the classical symptoms, because although they carry the faulty gene that would normally mean they get the disease, they also have something else protecting them—possibly another mutation. We know people who have made it to adulthood without any symptoms are going to be very rare. That's why we are referring to them as unexpected heroes.

### **Why is finding them so important?**

We know that those who avoided getting sick when young may harbor protective genes. The assumption is that finding these protective factors is a very direct path to developing new therapies.

That's because almost all of the genetic alterations that cause disease are due to a loss of function, say in a protein that the faulty gene codes for. Equally, a loss of function caused by the second mutations our unexpected heroes carry is probably what's providing protection. The vast majority of drugs we have use small molecules, which are extremely poor at restoring function, but very good at disrupting it. So if you found a dozen of these protective factors, there is a high chance you could build drugs to mimic most of them by recreating the loss of function.

**Read the full, original story: [Unexpected genetic heroes](#)**