THE 10 THINGS YOU NEED TO DO ONCE YOU HAVE A DIAGNOSIS

When Matt Wilsey daughter's Grace was diagnosed with NGLY1 deficiency, it came only after a three-year odyssey to find an answer to what was ailing her. Even with a diagnosis in hand, Wilsey had few alternatives. Grace was one of only a handful of people worldwide known to have the ultra rare genetic disease.

NGLY1 is part of a family of congenital disorders of glycosylation. In these disorders, the cells of the body are unable to synthesize N-glycanase, an enzyme that plays a critical role in the recycling of certain misfolded glycoproteins. In the absence of the enzyme, these misfolded proteins accumulate in cells and can cause harm in a variety of ways. This includes developmental delays, movement disorder, and liver problems.

A serial Silicon Valley entrepreneur, Wilsey and his wife formed the Grace Wilsey Foundation and set off to connect to leading researchers around the world to engage them in finding answers about the disease and hopefully a cure.

During a fireside chat at the Global Genes 2015 RARE Patient Advocacy Summit, Wilsey discussed his experiences, what he's learned in his efforts to enlist a multidisciplinary group of scientists into working on NGLY1 deficiency, and how he's brought a Silicon Valley entrepreneur's approach to this work.

> "Silicon Valley wants to disrupt. They want to disrupt the status quo," said Wilsey, pointing to examples such as Uber and how it has challenged the taxi industry. He also echoed Facebook CEO Mark Zuckerberg's motto "move fast and break things."
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> "That's what we're trying to do—we're trying to break the system," said Wilsey. "We don't have 50 years to cure this disease. We have five years to cure this disease. The deck is stacked against us. The probability of success is quite low, but the fight is still worth fighting."

Drawing from his experiences, Wilsey offered conference attendees the following ten things to do after getting a rare disease diagnosis and building an organization to find treatments:

1. CELEBRATE

Getting a diagnosis is a major accomplishment and people often don't take time to appreciate that moment. Wilsey said you should be present with your child and significant other and take time to appreciate this development before getting to work.

2. CONTINUE TO BUILD RELATIONSHIPS

In order to arrive at a diagnosis, most people have had to do a fair degree of relationship building. But Wilsey says don't be blinded by friendship and gratitude. The team you will need after a diagnosis will most likely be different than the team that provided the diagnosis. If you are building an organization to support research, it will be important to find and recruit and key scientist to help establish a translational beachhead and be a source of unbiased advice.

3. LEARN FROM OTHERS WHO HAVE GONE BEFORE YOU

There are many issues you will confront that others have already had to work through. Reach out to people who have gone through the issues and benefit from their experiences. Wilsey says not a week goes by that he is not emailing other people who have built organizations like his to see how they handled specific problems. "It's not rocket science," he said. "There are a lot of smart people who have been doing this."

4. START SUPPORTING RESEARCH

There's no reason to delay getting involved in supporting research. "Dive into the deep end," says Wilsey. "Don't wait." It doesn't take a multi-million budget to have an impact. Raising $80,000 is enough to support the work of two

post-docs for a year. The important thing is to set objectives and milestones and check in quarterly to monitor progress. Though it's not always possible to predict where science will take you, it's important to understand where you are going and to have some way of measuring progress because you will have limited capital and need to be efficient in deploying it.

5. FOCUS

The key is to focus on the patients. Every decision should be made with them in mind. It's not a popularity contest. You are going to ruffle some feathers—with parents, scientists, clinicians, or the National Institutes of Health. "If you are not ruffling some feathers," he said, "you are not pushing hard enough."

6. STAY FLAT AND FAST

Everyone is worried about failing, but in Silicon Valley it is not a bad thing. "It's almost like a badge of honor," said Wilsey. He said it's important to create an organization that's flat and create an environment of open collaboration.

7. BUILD A DREAM TEAM

Seek out the best scientists in each field and leverage diversity in building a team. There are many fields in science and medicine that are needed to treat or cure a rare disease. Seek out the best in each field. The scientist should be learning from each other.

8. RARE UNLOCKS THE COMMON

Wilsey thinks it's important for rare disease advocates to carry the message that rare diseases play a critical role in unlocking more common diseases. It's an important argument to attract funding and engage researchers. He argues hitting a problem head on seldom works and that for big ticket diseases, where we are spending billions of dollars and making little or no progress, rare disease research can provide breakthroughs. In the case of NGLY1 deficiency, Wilsey said researchers are finding shared pathways with cancer, Alzheimer's disease, and Parkinson's disease.

9. KEEP THE FAITH

Things are going to get challenging. This is the biggest challenge of your life. You will need to be strong and confident. Find faith in something to help give you strength, whether it's your religion, family, or hometown baseball team—anything. "I have a bad day a couple of times a week," said Wilsey. "You have to keep fighting."

10. BE OPEN TO SERENDIPITY

Be open to the unexpected. The best ideas often come from the least expected places.

CONTENT SPECIALISTS

Matt Wilsey, Parent Advocate, NGLY1 and President, Grace Wilsey Foundation
Matt is a Silicon Valley angel investor and advisor. In addition to consumer products and services, Matt invests in and advocates for biomedical research, drug development, and genetic sequencing technologies.

Before moving to the investment side, he spent many years as a front-line operator. Most recently, Matt was Co-Founder and Chief Revenue Officer of CardSpring, a payment infrastructure company that was acquired by Twitter. Previously, Matt ran West coast sales and business development for Howcast.com. He was responsible for building Howcast's instructional content library, distribution network, and strategic relationships.

He started his career serving in various roles at the White House and the Department of Defense. Matt became a rare disease hunter and advocate after his daughter Grace was born with NGLY1 Deficiency. He

has since funded 19 research teams accounting for over 50 scientists in three countries with the sole purpose of curing the disease.

Matt holds a B.A. from Stanford University and a M.B.A. from Stanford's Graduate School of Business. In addition to Global Genes, Matt sits on the Board of Directors of the Grace Wilsey Foundation where he is President, The Lucile Packard Foundation for Children's Health, the EveryLife Foundation for Rare Diseases, and Perlstein Lab.

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Daniel Levine is an award-winning business journalist who has reported on the life sciences, economic development, and business policy issues throughout his 25-year career. Since 2011, he has served as the lead editor and writer of Burrill Media's acclaimed annual book on the biotech industry and hosts The Burrill Report's weekly podcast. His work has appeared in The New York Times, The Industry Standard, TheStreet.com, and other national publications. He also is the host of RARECast™ podcasts.