

AN INTERVIEW WITH

Robert Thompson Stone, MD

Grant Recipient
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Can you give a brief summary of the project you'll be working on with your grant funding?

The project is titled “GenoKrabbe: A Comprehensive Genotype-Phenotype Database for Krabbe Disease”. It’s a project I'm working on with the University of Minnesota. They were funded initially, and then we were funded after them. The aim of the project is to create a multi-institutional platform that is accessible to our institutions together, but also to the Krabbe community; patients, families, advocacy groups, etc.

As Krabbe newborn screening grows, which we're really excited about, there is obviously data that shows clear emergent cases of infantile Krabbe, which we know how to move forward with. But there is also a large amount of individuals, which will only grow as newborn screening increases, for whom the data is a little bit unclear. It seems like they're likely at risk, but not for the infantile form of what we call late onset, or later onset Krabbe disease. And our goal is to understand what that risk is, or precisely when the age of onset might be, when we should be screening, and how aggressively we should be screening at any given age. We have some ideas about those questions, but there's still a lot to be learned.

The thing that really inspired me to be a part of this project is that I'm a member of the Krabbe Newborn Screening Council, where we talk about issues regarding Krabbe newborn screening and also go through cases that are coming in from the different states. Time and time again, at the end of discussions about a complex case, this group comes to the conclusion that we really need to understand these genotypes better, how the disease evolves over time, and what we should be doing for these patients.

In the past, there have been databases that have some information, but they are fairly siloed without a lot of accessibility for families or clinical providers. Our goal is to not only try to add to the knowledge base of what this data means and how we can use it, but also to make sure that this is something that is very accessible to the people who need it. And that's why it was important for us to work together in a multi-institutional collaboration, and for us to choose a third party platform that wasn't within just one individual institution.

How did you become involved in Krabbe disease research?

In medical school I was interested in neurology, and then within neurology, I became interested in child neurology and more complex central nervous system diseases, eventually settling on white matter disorders. That field interested me because of the connections that you can make with people who have complex needs, being able to work with them through those needs and developing some specialty expertise that can provide assistance that families might not be able to get otherwise.



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New York state was the first state to screen for Krabbe, many years ago. There were a lot of challenges to that process, though because we didn't have psychosine and all these other things that help us to understand how to classify it. A previous individual at our institution had been a part of a lot of the newborn screening discussions, on the front lines trying to help families. They ended up leaving and going to a different institution that was out of state, and I took over where they left off, which helped me to fast track some of those connections and get going in this field, which I didn't necessarily expect.

What about your research and the research of others in this field gives you hope?

I think there's a lot of reason to hope for the Krabbe disease and leukodystrophy communities. Within this research, there is hope that with the expansion of newborn screening we can help with the specificity of our prognostic information and recommendations for families. There is also hope that in the future, parents won't accidentally be told that their child has some form of this disease that isn't true; that we're not under or over-testing babies or toddlers based on their risk profile. There is also great preclinical therapeutics research going on, giving us hope around gene therapy and improvements in stem cell transplant techniques and mortality rates.

What are both the best and the most challenging parts of your job?

The best part of my job around taking care of these kids with leukodystrophies is forging connections. Getting to know them really well over time is a great part of what I do. I also really enjoy thinking through the best way to communicate. Communication is so critical, from that first phone call when you have to tell a family that there was a newborn screening positive, and what that means, to communicating what a complex genetic result means, and what to expect. It's important to do that in a way that's respectful of patient families and what they want to know, and making it a conversation.

I also enjoy going above and beyond to help people who are struggling or trying to work through a challenging situation. For example, I just diagnosed a neonate with vanishing white matter disease, which is a different leukodystrophy. There's a clinical trial currently going on for that disease, but this patient doesn't qualify. So the challenge in this case is trying to figure out if there's there a way that we can get treatment to this baby outside of a clinic. It's a problem that's more complex, but really worth it.

At the same time, one of the biggest challenges is exactly that: the logistics of complex processes. So how to get treatment for that baby outside of a clinical trial is a big challenge, because there's probably a lot I don't understand about those processes, and they're all unique. Because of this, it can be difficult to build up a lot of experience for each specific scenario.



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Advocating for what the families and kids need can also be a big challenge. Even if we do all the hard work of understanding what they need, there are a lot of barriers to implementation, and moving through the red tape - whether it's issues with insurance, or understanding hospital processes, or the ways that companies might interface with our institutions, or travel needed because treatments might only be available in a few different places around the country - can be pretty challenging.

How do you spend your time outside of the clinic?

I'm married, and I have two kids and two very cute goldendoodles. We spend a lot of time outside, and my favorite hobby is pickleball, which I play two or three times a week to have fun and stay active.