



Research Roundtable Community Summary – July 23, 2020

KIF1A.ORG hosted our first monthly Research Roundtable on July 23, 2020. Research Roundtables bring together leading KIF1A researchers to share updates, discuss challenges and exchange ideas. Our ultimate goal is to facilitate collaboration so we can accelerate the path to KAND treatment. The first Research Roundtable was largely an introductory meeting of our long-term Research Network participants. In that spirit, read on to learn more about key members of the Research Network who attended the inaugural meeting.

Attendance



16 RESEARCHERS



10 INSTITUTIONS



3 KIF1A.ORG REPS

“Scale of Research” Guide

Before we learn more about these researchers, here’s a quick overview of some terminology you’ll see below. Research is conducted in a range of settings: from clinical research in humans, to biophysical research at the molecular level. Often times, researchers tend to specialize in one or two scales of research.

- **Clinical:** Research conducted at the human level, often focused on specific diseases or disorders.
- **Mouse Model:** Research conducted on mice that can simulate components of human diseases or disorders.
- **C. elegans Model:** Research conducted on a type of nematode worm (yes, a worm!!!) that can simulate components of human diseases or disorders.
- **Cellular:** Research conducted on isolated cells that are grown in a petri dish.
- **Molecular:** Research conducted on the components *inside* of a cell.
- **Biophysical:** Often conducted at the molecular level, biophysical research uses the principles of physics to understand biological systems.

Meet the Researchers



Dr. Wendy Chung

Location: Columbia University | New York, NY, USA

Scale of Research: Clinical – Precision Medicine/Mouse Model/Cellular

- We all know Dr. Chung—to say we appreciate the work she does is the least we can do!
- Dr. Chung played an important role at this meeting by presenting on the clinical presentation of KAND, based on the most current data from the KIF1A Natural History Study. While many of these researchers study KIF1A, not all of them have KAND-specific projects. Thus, Dr. Chung’s brief introduction to KAND was very informative to many participants and yielded a lot of very good questions from the other researchers!



Dr. Cat Lutz

Location: The Jackson Laboratory | Bar Harbor, ME, USA

Scale of Research: Mouse Model

- Dr. Lutz has been a long-time member of our Research Network and is the Director of the Mouse Repository and the Rare and Orphan Disease Center at The Jackson Laboratory.
- Dr. Lutz was an essential player in the development of our KIF1A mouse models.
- Dr. Lutz also serves as the Director of In Vivo Pharmacology and Efficacy Testing Program in Bar Harbor, which interfaces with biotechnology and pharmaceutical companies to pursue novel therapeutics across a variety of therapeutic areas.

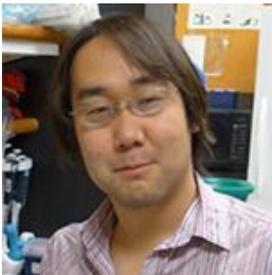


Dr. Richard McKenney

Location: University of California, Davis | Davis, CA, USA

Scale of Research: Molecular/Biophysical

- Dr. McKenney's research is focused on understanding how the movement of molecular motor proteins like KIF1A is balanced and coordinated (and how this balance and coordination is altered in certain diseases).
- In 2019, Dr. McKenney and Dr. Niwa (who you'll learn about below) published a paper on a handful of KIF1A mutants. This was one of the first studies that investigated the functional implications of certain KIF1A mutations. You can check out the paper and our Research Simplified summary [here](#).
- Fun fact: Dr. McKenney received his PhD in Dr. Richard Vallee's lab. Dr. Vallee is at Columbia University, and has collaborated with KIF1A.ORG since our first family meeting in New York City in 2017. The kinesin world is very, very small!



Dr. Shinsuke Niwa

Location: Tohoku University | Sendai, Japan

Scale of Research: Cell Biology/*C. elegans* Model

- Dr. Niwa's lab is focused on understanding how kinesin motors influence a process called cellular morphogenesis. In other words, his lab wants to know why certain cells end up being one shape, while other cells end up being a totally different shape.
- Dr. Niwa uses a *C. elegans* model to understand the impact of certain KIF1A mutations in the nervous system.
- Dr. Niwa has been studying KIF1A for over a decade.
- Dr. Niwa and Dr. McKenney regularly collaborate on research projects. In fact, Dr. Niwa's work on KIF1A is what got Dr. McKenney thinking about KIF1A in the first place!
- Fun fact: As Dr. Niwa is located in Japan, his attendance at this meeting meant he was joining at us 4:00 a.m. Japan Standard Time—we thank him for his dedication and willingness to start his day with this meeting!



Dr. Arne Gennerich

Location: Albert Einstein College of Medicine | Bronx, NY, USA
Scale of Research: Molecular/Biophysical

- Dr. Gennerich is most widely known for his work on understanding the force production of kinesin motors like KIF1A. Essentially, he seeks to understand how “strong” of a machine certain kinesin motors are, and what properties of kinesin motors can make them “stronger” or “weaker”.
- Dr. Gennerich collaborates with many of our KIF1A Research Network members and has been a fantastic scientific advisor for our organization over the past few months.
- Fun fact: Dr. Chung, Dr. Vallee (see below), and Dr. Gennerich have received a “multi-PI” (PI = Principal Investigator) grant from the National Institutes of Health (NIH). This means that these three labs have teamed up to tackle a common project on KIF1A/KAND. This is the largest grant provided for KAND-specific KIF1A research to date!



Dr. Kristen Verhey

Location: University of Michigan | Ann Arbor, MI, USA
Scale of Research: Molecular/Biophysical

- Dr. Verhey has been a pioneer molecular KIF1A researcher since the early 2000s. Much of what we know about KIF1A’s function on microtubules is based on her lab’s work.
- Some of Dr. Verhey’s work on KIF1A has focused on understanding the specific steps needed to turn KIF1A from the “off-position” (unable to transport cargo), to the “on-position” (able to transport cargo). This is an area that is currently being investigated by many researchers and is viewed as a potential therapeutic target in the lifecycle of a KIF1A protein.



Dr. Richard Vallee

Location: Columbia University | New York, NY, USA
Scale of Research: Mouse Model/Cellular

- Dr. Vallee's lab has been investigating KIF1A for many, many years.
- In the past Dr. Vallee's lab has researched the role of KIF1A in brain development, using a mouse model.
- More recently, Dr. Vallee has started researching KIF1A in the context of KAND to understand the effect of KIF1A mutations.
- If you attended our 2019 KAND Conference, you probably saw a presentation from his lab about the relationship between KIF1A and a protein called BDNF. You can watch the presentation [here](#).



Dr. William Hancock

Location: The Pennsylvania State University | University Park, PA, USA
Scale of Research: Molecular/Biophysical

- Dr. Hancock's work is focused on understanding the intricate mechanics of the KIF1A machinery. You can think of his lab members as being part KIF1A engineer and part KIF1A mechanic!
- Having spent many years studying other kinesin motors, his lab is now newly focused on studying KIF1A.
- Fun fact: Dr. Hancock is well known for his guitar playing skills and has been known to create parody science songs that he debuts at scientific conferences.

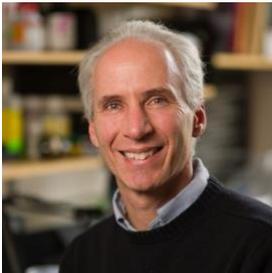


Dr. Erika Holzbaur

Location: University of Pennsylvania | Philadelphia, PA, USA

Scale of Research: Molecular/Biophysical/Cellular/Mouse Model

- Dr. Holzbaur's lab studies the role of motor proteins across many levels and projects. Most pertinent to KAND, her lab focuses on how mutations or defects in molecular motor proteins like KIF1A can lead to neurodegenerative diseases.
- In 2019, Dr. Holzbaur's lab published a paper investigating the effect of over 15 KIF1A mutations—the most comprehensive study at the time. Read the paper and our Research Simplified summary [here](#).



Dr. Christopher Berger

Location: University of Vermont | Burlington, VT, USA

Scale of Research: Molecular/Biophysical

- Dr. Berger's lab is focused on understanding how KIF1A cargo transport is regulated. In other words, what helps control the KIF1A flow of traffic in our cells?
- Dr. Berger's lab mainly focuses on how KIF1A and other kinesin motors respond to obstacles, or roadblocks, while engaging in cargo transport.
- Fun fact: KIF1A.ORG's Science Communication Director, Dr. Dominique Lessard, conducted her PhD dissertation in Dr. Berger's lab!

What's Next?

- Scheduling August Research Roundtable
- Expecting additional KIF1A researchers to join future meetings, including researchers we're engaging from outside the U.S.
- Collecting feedback from Research Roundtable participants on:
 - Suggested topics of discussion for future meetings (in addition to sharing general updates)
 - Ideas for the KIF1A Research Hub, a digital collaboration platform we're building for researchers as part of the CZI Rare As One grant awarded to KIF1A.ORG