



Research Roundtable Community Summary – October 22, 2020

KIF1A.ORG held our fourth Research Roundtable meeting, “Think Tank: Solving the KIF1A Maze” on October 22. Our goals for this meeting were to:

- 1) Crowdfund information and ideas regarding outstanding therapeutic development gaps in knowledge
- 2) Speak with researchers to help identify which gaps in knowledge we should focus on to move forward

Attendance



8 INSTITUTIONS



21 RESEARCHERS



3 KIF1A.ORG REPS

Big Takeaways

Commonalities between rare neurological diseases

- The meeting kicked off with a discussion about rare neurological diseases that present similarly to KAND on a clinical level.
- In the basic research world, disease/disorder-related gene mutations are commonly investigated at a molecular level (small scale research). Findings from these types of studies can be used to help us understand a specific part of the clinical level of a disease/disorder.
- However, another approach is to look at a disease/disorder from a clinical level and try to identify similarities and differences between other diseases/disorders. The similarities and differences can then point us to new targets that are worth investigating at the molecular level.
- This is also known as the “bottom-up” vs. “top-down” style of investigation.

Proteins that could mediate KIF1A cargo transport

- KIF1A's main role in our nervous system is to transport cargo throughout the neurons of our nervous system.
- Like any sort of trafficking, this process must be efficiently regulated to avoid things like traffic jams or cargo lost in transit.
- We spent time in this meeting discussing proteins that we already know regulate KIF1A cargo transport. From this, we began to extrapolate on other proteins that have not yet been shown to regulate KIF1A cargo transport but are likely candidates based on what we know about them.

The role of KIF1A in neuronal health

- We spent a large part of this meeting discussing the processes in our bodies that keep neurons and our nervous system healthy.
- Furthermore, we discussed what we know about KIF1A's role in neuronal health and how KAND-specific mutations may alter these processes.
- One process discussed that may be a new concept for some is called "autophagy." This is the body's way of cleaning out damaged cells that may be harmful if not disposed of properly. Currently, we do not know how KAND-specific mutations may alter this process.

There is merit in digging deeper into what we know

- In this meeting we discussed the importance of "digging deeper" into what we already know about KIF1A.
- While it is extremely helpful to identify new information, it is also important to reflect and identify the massive gains in KIF1A knowledge that the scientific community has made over the past 10 years.
- With strategic follow-up investigation of certain areas of KIF1A knowledge, we could learn a lot that could help drive therapeutic discovery.

The "fanciest" experiments are not necessarily the answer

- Scientific technology is evolving around us at a rapid pace. As is such, it can become easy to get wrapped up in the "newest" or "most sophisticated" techniques for scientific investigation. However, in this meeting we discussed how just because a form of investigation is older or more simplistic does not mean it should be off the table as a potential investigative tool. While we encourage and are enthused by the discovery of new scientific tools, we also must remember that we have so many useful and accessible tools at our fingertips already!