# 09.26.24 Virtual Forum Summary: Seth Keller, Brian Chicoine and Amy Kolb Tucker

1. Holly introduced herself and went over the IDD-TRANSFORM project.
2. Holly went over accessibility for the virtual forum.
3. Holly acknowledged the funding for the Down Syndrome/Trisomy 21, Aging and Alzheimer's workgroup project. It is funded by the Stanford Alzheimer's Disease Research Center.
4. Holly acknowledged that there were no conflicts of interest to be disclosed.
5. Holly shared that continuing medical education (CME) credit was available for the forum.
6. Holly went over the agenda for the forum.
7. Holly went over how to submit a question to the panelists.
8. Holly went over labels and how our workgroup has chosen to use the term Down syndrome/Trisomy 21.
9. Holly thanked the members of the Down Syndrome/Trisomy 21, Alzheimer's and Aging Workgroup.
10. Holly shared that resources would be listed at the end of the session as well as on the IDD-TRANSFORM website: https://med.stanford.edu/idd-transform
11. Holly introduced Dashiell Meier, a Special Olympics athlete and member of the workgroup.
12. Dashiell introduced Dr. Seth Keller, Amy Kolb Tucker and Dr. Brian Chicoine.

13. Megan transitioned into the panelist discussion (see panel questions below).
14. Holly thanked the panel for their time.

## Panelist Questions and Summary of Response(s):

**Amy, tell us about your and Kate's journey with Alzheimer's disease. What challenges did you face in the Alzheimer's diagnosis process for your daughter, Kate and what challenges and questions do you continue to face in Kate's journey with Alzheimer's?**

Amy: Kate was typically one of the higher functioning individuals with Down Syndrome. We were really naïve and caught off guard when we first got her diagnosis at the age of 40. Because I assumed she was on the higher end of the spectrum, she would not face it until she was much older.

**Where have you gone to find information about Alzheimer's disease and what support/resources have been helpful to you that you'd like other families to know about? [ie. Advocate Medical Group, EDSD from the NTG, NDSS]**

Amy: When we first suspected symptoms of Alzheimer's, the first resources we consulted were from the National Down Syndrome Society. They have two publications that we used: The Alzheimer's Disease in Down Syndrome publication and the Alzheimer's Disease in Down Syndrome

Guide for Caregivers. We also found the National Task Group on Intellectual Disabilities and Dementia.

**The NTG developed the EDSD. Can you tell us what that is (and what it's not)? Who can and should make use of it? What does it tell you?**

Seth: The EDSD was created a number of years ago when the NTG was first founded. It's a screening tool to document particular symptoms and if there's a change in someone as they get older. It is highly recommended to start using this tool at around age 40 and it is filled by caregivers.

**Could you tell us what you would advise for a patient with Down syndrome or their family member who may be worried about developing Alzheimer's, or even already showing possible symptoms? What are the symptoms in DS and how are they different from Alzheimer's in other people?**

Seth: In terms of changes, some of the symptoms involve behavioral changes as well as the inability to do some of the activities a person with Down Syndrome/Trisomy 21 used to do before. There are also elements of confusion. A major red flag is the beginning of new seizures.

Brian: Many people with Down Syndrome have what's called the groove which is a tendency to do things the same or repetitiously and it can be very functional throughout their life. With Alzheimer's, they become more groovy and get stuck in things and have a hard time adjusting or they lose

that groove. We also see a change in gait. Some of that is balance and some is depth perception.

**What do physicians and other healthcare providers need to know about the unique needs and possible comorbidities in adults with DS? How should that inform their approach to diagnosis and care? (ie. addressing environmental stimuli, higher frequency of seizures, differences in pain, challenges of completing advance directives, etc.)**

Seth: It's hugely important for primary and specialty care to appreciate that it might be a great issue of why change is occurring. Behavioral challenges and social issues are important to understand. It's important to look at the social cues and social history of the patient with Down Syndrome. Looking at hearing problems is also important.

Brian: As a primary caregiver, going through the differentials is more important than the diagnostics. The rule outs are really important in this population. The family support is also a really important piece of this.

Amy: I do know from parents I have talked to and my own experience, we really appreciate the health providers who talk to our loved ones as opposed to talking to us. Including the patient is really important.

Seth: Don't make false assumptions about what they can or cannot say to a health provider.

**A big barrier we are hearing about from families is lack of provider education – or lack of educated providers who know about the increased risk of Alzheimer's in adults with DS or know how to conduct a differential diagnosis. What does education currently look like for healthcare trainees about Down syndrome and IDD more broadly? What are they learning? How can it be improved? What are some of the challenges?**

Seth: The reality is there's no training deemed mandatory for most health care providers in adult-oriented coursework. In pediatric medicine, they will learn at some point about people with developmental disabilities born through the early life, but as they transition into their adult life, there's nothing within most medical schools that mandate that health providers have to learn about this. This results in a lack of quality services. To address this, make curriculum about developmental disabilities mandatory, take a look at the medical education curriculum. The American Academy of Developmental Medicine and Dentistry has created a number of medical school trainings around intellectual and developmental disabilities, so the future is brighter about education on developmental disabilities.

Brian: Seth mentioned the Down Syndrome Medical Interest Group and I encourage you to take a look at it. It's very inexpensive and has webinars. There's also a panel of experts. I'm also working with the National Down Syndrome Society on developing a short course on caring for people with Down Syndrome.

**While there are some centers that specialize in caring for adults with Down Syndrome, we know most places don't have such a specialty clinic. How would you advise clinicians and families in areas without these focused clinics, especially those who live in more rural areas with less access to care? How do families get better outcomes from the resources and physicians available to them?**

Seth: Family advocacy, community support advocacy, is key to bringing information to doctors in a friendly manner. Help the medical professionals help you and it helps you understand as to whether that medical professional is open-minded. It's mostly due to not having the information.

Brian: Most people are willing to learn. It does fall onto family members to bring information on Down Syndrome into the clinic.

Amy: It's a delicate balance. We don't want to come in and be "those" parents and the doctor rolls their eyes and sets it aside. This is a collaboration. We're coming in to help each other.

**Do you have any other suggestions about empowerment or educational materials?**

Seth: I provided some resources in the chat from the NTG website that talk about resources and information that you would learn for yourselves and to bring to the provider. (All resources have been added to a resource document, found here.)

Brian: Learning from each other is also important. Every person with Down Syndrome is unique.

**What can healthcare providers do right now to learn more about this topic and begin to improve their clinical care for patients with Down syndrome? Where can they go to find educational tools and resources? (ie. Project ECHO, AADMD chapters?)**

Seth: It's a double issue. What can a healthcare provider learn now and are they in any area in which they are partnering with a memory center or cognitive center? It's really about partnerships in terms of that.

## Questions and Answers:

**If people are suspecting an Alzheimer's diagnosis in their family member with Down Syndrome and are having trouble getting it, how important is it to see a neurologist and what should they do? What kind of doctors should they look for if your son or daughter already has a diagnosis?**

We want the individuals to have a medical home and a primary doctor to take care of that. A neurologist might be useful if there's a complicated seizure disorder or if there's something that's being missed. An orthopedic surgeon might be involved if there's a question about a gait or something more than Alzheimer's. Urologists can also come into play.

**Is there a registry of neurologists who are knowledgeable about Down Syndrome or interested in that?**

There is not.  The challenge of that is you can get a name of someone you know, but you have to verify if they're good at it.

**Can you talk about biomarkers and treatment?  How can people get their loved ones with Down Syndrome involved in clinical trials?  Who is the right person to do treatment for Alzheimer's disease in someone with Down Syndrome?**

Biomarkers are research that will make a change in the accuracy of the diagnosis of Alzheimer's disease.  There are drugs out there that have just been released and it's for Alzheimer's Disease.  The problem is that with the research that's been done, there is a risk with swelling and bleeding of the brain with treatment.  People with Down Syndrome might be more disposed to serious complications.  Safety studies have not been done yet.

**Do you have suggestions for people who want to get involved in research and other advocacy efforts to improve research in this area?**

The LuMind and the Alzheimer's Biomarker Consortium for Down Syndrome sites have portals that you can enter information and they will give you information about clinical trials and epidemiological study sites. There's also a site called Clinical Trials.gov where anyone who's doing a study like that has to register the study.

**Can you give people advice about asking providers about behavioral challenges in caring for a loved one with Down Syndrome and Alzheimer's?**

The Adult Down Syndrome Resources Center has a tremendous resource library of visual aides to assist in everything from reminders of the process of brushing your teeth, social stories to deal with emotional problems, graphic organizers on taking charge of medical appointments and other things. It has ways of speaking with your loved one with Down Syndrome who may be less verbal.

**What has the role of the regional center been and some of the gaps in the community resources, both provided by the regional centers and housing organizations?**

California has a fairly decent regional center that coordinates care for all individuals with intellectual and developmental disabilities. In an ideal world, there would be some way for regional centers to be educated and knowledgeable on this area ahead of time. Ideally, there would be information handed out at the person's age of 35 to families so they know what they need to get for their loved one. Regional centers are the perfect agency to deliver that information.

**What do we need government services providers to do as well as the challenges of finding residential options for people with Down Syndrome who have Alzheimer's disease?**

You want to make sure that whoever you talk to understands that they have Down Syndrome as well as Alzheimer's disease. In general, we're pushing aging people with Down Syndrome into a system that doesn't do a great job. If it is not possible to provide care at home, we need to find what care

facilities can help people with Down Syndrome and Alzheimer's disease. It goes back to the regional centers.

**What can you tell families about what we know now and what we might learn new in the coming years about prevention of Alzheimer's disease?**

The way we take care of ourselves will have an impact on our aging bodies and aging brains. Being mentally calm and relaxed, as well as eating well and sleeping well, are really important. Sleep apnea is very common in people with Down Syndrome and it is encouraged that they get that treated as soon as possible. Exercise and weight management are important too.

**What are the challenges with normal pressure hydrocephalus?**

It is a real phenomenon. You have to show that you have excess water on the brain. People with Down Syndrome might be born with this and have larger ventricles. If you want to know if they actually have it, reduce the amount of fluid in the brain and a Lombard puncture or spinal tap is performed.