

CSAW NEWSLETTER

OCTOBER 2020

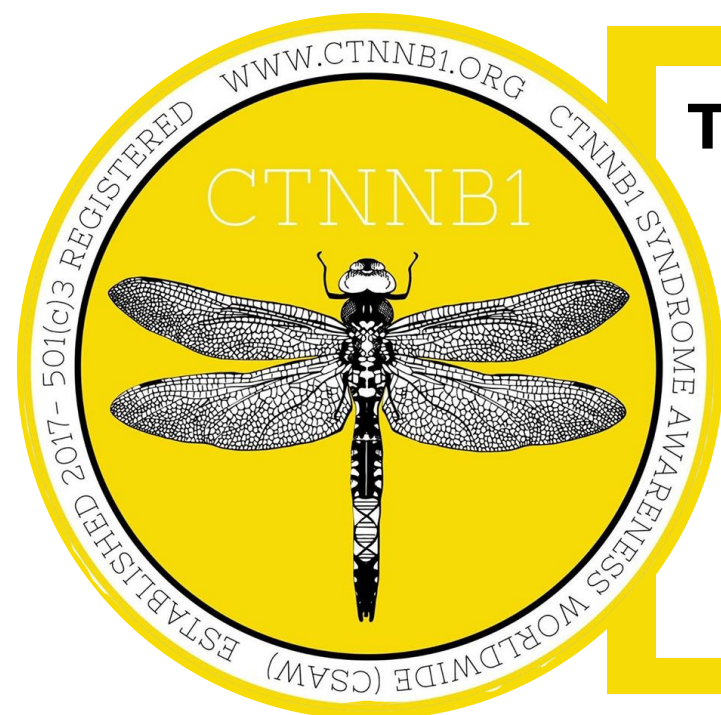


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Despite the many complications brought on by COVID, CSAW has had a busy year – and it's not over yet! Here's what we did, and what's to come.



CTNNB1 Day – July 25th

Last year we named July 25th as CTNNB1 Syndrome Day and adopted officially as our symbol the beautiful Dragonfly we use as our logo. Many of you shared pictures of your own dragonfly creations then and again this year.



CTNNB1 Day – July 25th continued

This year's celebration saw a remarkable collaboration with our sister organization, Advancing CTNNB1 Cures and Treatments ([ACCT](#)). Together we presented ReCon - CTNNB1 Syndrome Research and Connection, a half-day Zoom conference featuring Dr. Wendy Chung of Columbia University, and Dr. Michele Jacob of Tufts University. They spoke about the CTNNB1 Syndrome registry, natural history study, and research completed so far. (ACCT has issued its second grant to Dr. Jacob at Tufts. A new research update will be provided at the end of the grant agreement in February.)

More than 80 families attended the Zoom conference, and more than 700 have since viewed it on [YouTube](#). Doctors and researchers from 3 countries and 4 states representing institutions including the University of Washington, University of Hong Kong, Adelaide Medical School in Australia, Mt. Sinai in New York City, CHOP in Philadelphia, Johns Hopkins and Children's Hospital of New York Presbyterian, Columbia University, also attended.

The only disappointment was not being able to gather in person, which many of us, including Drs. Chung and Jacob, were looking forward to. If COVID restrictions are lifted by next year, we'll try for that again.






Knowledge in Numbers

One of the key takeaways from ReCon was the urgent need for our families to participate fully in the CTNNB1 Syndrome registry and natural history, administered through Simons Searchlight. And it's not just ReCon researchers underscoring the importance of collecting this vital information. In their recent virtual summits, both the National Organization for Rare Disorders (NORD) and Global Genes highlighted the same thing.

But with more than 7,000 rare disorders identified worldwide, CTNNB1 Syndrome is one of the smallest. Ours is an ultra-rare condition, most often defined as affecting fewer than one in one million people. That means we need to provide as much reliable data as possible for the medical, scientific, therapeutic, and pharmacological communities to become aware of and understand the syndrome if treatments, therapies, and, perhaps someday, a cure, are to advance.

CSAW recently published its first CTNNB1 Syndrome Survey Annual Report (available at www.ctnnb1.org/registry), a valuable resource for families, doctors, therapists, and researchers. We want to continue to update and expand the information, and that means each affected family needs to participate.



If your child has CTNNB1 Syndrome, please register with Simons Searchlight!

Here is what you need to do: Visit Simon's Search light:

<https://research.simonssearchlight.org/account/create>
and create an account. You will need to upload your child's genetic report. This is followed by a call with a genetic counselor to discuss your child's medical history and development. Families can email Simons Searchlight Coordinator with any questions on how to enroll (coordinator@simonssearchlight.org).

If you have any questions before you begin, don't hesitate to contact either CSAW or ACCT; we work together to capture and curate CTNNB1 Syndrome data for Simons.



CSAW – Connecting With Each Other

The usual challenges of parenting are compounded for parents and primary caregivers of children with needs that are special. The recent restrictions due to the COVID pandemic have made things harder for many. But we've also seen how powerful connecting virtually can be. So our creative and compassionate families have come up with some new ideas to help us help each other.

- The Parents' Place Moms and Dads sometimes need a place to go to for guidance and support. We want to provide help with the many challenges our parents face, such as coping with the emotional demands of full-time caregiving, advocating for appropriate school interventions and accommodations, and knowing what we need to do to be ready for when our children turn 18. The first topic, presented by CSAW VP and school counselor Annie Wood, is Organizing Your Child's IEP Binder. We welcome your ideas for future topics.
- Virtual Meetups The burden of stress is great for parents of children with needs that are special. Fear, worry, guilt, isolation, grief, and depression are all common. Often the most beneficial support parents receive is from other parents. Connecting in person is difficult right now, so we want to help make these connections happen virtually. Watch the parent Facebook page for virtual-meetup information, or contact Annie at annie.wood@ctnnb1.org if you want further information or if you just want to connect with another parent.
- CTNNB1 Grandparents and Family Facebook Page For many good reasons, the CTNNB1 Facebook page for parents is private – only parents are admitted. But we've all heard from other family members – including many grandparents – who yearn to understand more about their family's situation. So CTNNB1 mom Effie Parks – also host of the podcast Once Upon a Gene – set up a FB page for Grandparents and Family of CTNNB1 children.

It also is private, so if you have a family member who would like to join, they will have to request permission. In less than a week there already are almost 70 (very pleased) members.

Park Perks

Did you know that your CTNNB1 family member may be eligible for a free lifetime pass – one per car – to our amazing national parks? You can apply for the National Parks Access Pass for US Citizens at:

https://www.nps.gov/planyourvisit/passes.htm#CP_JUMP_5088581

A letter from your doctor is required. (Thanks to CTNNB1 Mom Jenny McMahon Blanchard for the great tip!)



Smile for CSAW!

Don't forget! Amazon Prime Day is October 13-14 (with some pre-deals already happening!) Be sure you've joined Amazon Smile and selected CTNNB1 Syndrome Awareness Worldwide as your charity of choice, and Amazon will donate a percentage of all your Smile purchases to that organization.

