17q12 FOUNDATION

2022 NEWSLETTER

A RESOURCE FOR CHROMOSOME 17Q12 DELETION AND DUPLICATION SYNDROMES

WHAT'S NEW?

LETTER FROM THE BOARD

To our 17q12 community,

Thank you for helping us celebrate our fourth annual 17q12 awareness day! This last year has been a year of growth and connection for the 17q12 Foundation. In this past year we have welcomed new members from various countries, became members of NORD (National Organization for Rare Disorders), signed our registry contract, received our first grant, were nominated for a RARE Champions of Hope Award by Global Genes, and were welcomed as members of Global Genes Advocacy Alliance. We are thrilled to announce that we begin the onboarding process for the 17q12 registry this month! It is expected to launch in early 2023, and we will then be enrolling participants to better learn about chromosome 17q12 deletion and duplication syndromes. We are also looking forward to having another opportunity to connect through our virtual meet-ups. We can't thank you all enough for your support! We couldn't have made it to this next chapter without the 17q12 community. We hope you enjoy this year's newsletter where you can find details and our announcement about our registry partnership. Thank you for helping us represent the 17q12 community!

Allaina Wellman
President

Liz Fourie
Vice President

Sherie Scott
Secretary

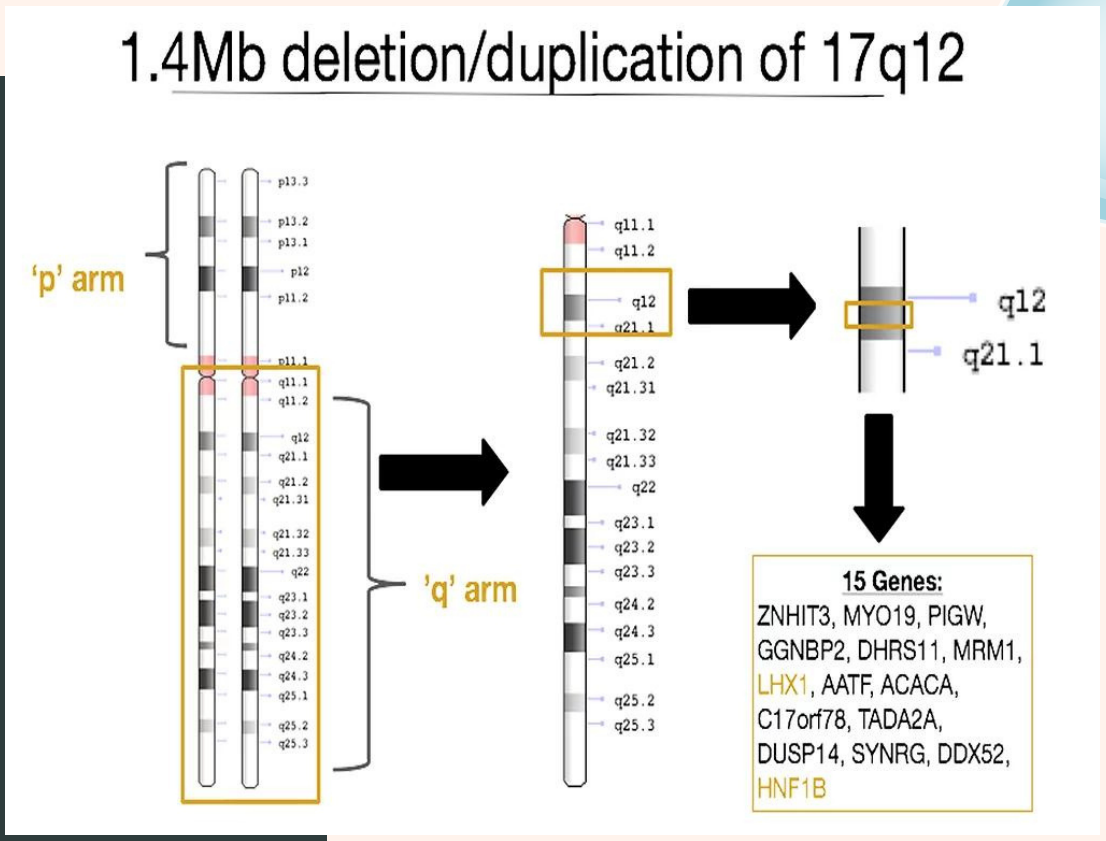
Mark Dempsey
Treasurer

Margo Casados
Board Member

WHAT IS 17Q12?

15 GENES

There are 15 genes involved in the two 17q12 syndromes that we represent: chromosome 17q12 duplication syndrome and 17q12 deletion syndrome. Some members may have a few more or a few less genes deleted or duplicated. Out of the 15 genes, only two of the genes' function is currently known. This is why research is so important. There is still so much more to learn about when one copy of these genes is missing or extra.



SYNRG

DUSP14

HNF1B

DDX52

PIGW

MYO19

DHRS11

C17orf78

MRM1

GGNBP2

ACACA

ZNHIT3

AATF

LHX1

TADA2A

Genes with a Known Function:

LHX1: Mayer-Rokitansky-Küster-Hauser syndrome

HNF1B: renal cysts and diabetes (RCAD) (diabetes only when deleted)

Celebrating Achievements

Kyle, age 12

UK

Kyle has been learning how to swim and has done absolutely amazing. He can swim with no aids at all!

Victoria, age 10

NJ, USA

Victoria just finished up a great soccer season with her other peers with special needs. They worked on teamwork, communication, and mobility. Victoria did a great job!

Davey, age 4

MS, USA

Davey is talking in full sentences!!

17q12 Foundation Partners with NORD® to Initiate Natural History Study of Chromosome 17q12 Deletion Syndrome and Chromosome 17q12 Duplication Syndrome

Research study is planned for participants worldwide to advance understanding and treatments for rare syndromes causing MODY5, kidney disease, heart issues, seizures, and the many other multisystem features and symptoms

July 12, 2022 — 17q12 Foundation has partnered with the National Organization for Rare Disorders (NORD) to plan and build a study to research chromosome 17q12 deletion and chromosome 17q12 duplication syndrome.

This study, designed by researchers, medical experts and patients, will provide the opportunity for patients around the world to share information about chromosome 17q12 syndromes to be used by scientists in future research. Both Chromosome 17q12 deletion syndrome and 17q12 duplication syndrome are rare disorders. Based on current research, about 1 in 2,500 individuals in the general population have chromosome 17q12 duplication syndrome and about 1 in 14,500 people in the general population have the deletion syndrome. Both syndromes affect various organs and systems, have different features, and may vary person to person. Some features between the two syndromes may include MODY5, kidney disease, heart issues, and seizures to name a few.

“17q12 Foundation was formed out of the need for research and understanding of both these rare chromosome disorders. We often see families and individuals in the 17q12 community searching for answers to unexplained symptoms and struggling to access important resources, only to hit roadblocks due to the lack of understanding and data that currently exists for chromosome 17q12 deletion/duplication syndrome. We raised the funds together as a community, and now it is time to advocate and push for the information and understanding that our community needs! The only way to move forward with research is together. We aim to keep the 17q12 community as our focus as we leap into this next chapter.” - Liz Fourie, Vice President, 17q12 Foundation

This natural history study will be hosted on NORD’s IAMRARE® online platform. It is designed to track the course of chromosome 17q12 syndromes over time in a set of electronic surveys. Patients, or their caregivers or guardians, will enter information from anywhere in the world.

“The success of the registry is dependent on asking the chromosome 17q12 community for their participation in the design of the study. Combined with the input of experts, this will ensure that our surveys collect the information necessary to advance research and tell the stories our community needs to share,” said Liz Fourie.

The data is confidential and stored securely on the platform. One goal of the 17q12 Foundation is to share the collected data with individuals or institutions conducting research or clinical trials in Chromosome 17q12 Deletion Syndrome and Chromosome 17q12 Duplication Syndrome.

The 17q12 Foundation is launching this study in collaboration with NORD, an independent nonprofit that built its natural history study platform as part of its mission to help identify and treat all 7,000 rare diseases. The 17q12 Foundation is a member of NORD, and these two organizations will work together to understand the challenges and identify opportunities to advance research for this rare disease patient population.

For more information, visit chromo17q12.org.

About 17q12 Foundation

The 17q12 Foundation represents individuals and families with either chromosome 17q12 microdeletion syndrome or microduplication syndrome. After meeting at a family conference for 17q12 syndromes in 2017, a group of four parents and a genetic counselor came together with the same vision, to begin a non-profit organization for chromosome 17q12 syndromes. 501(c)(3) status was gained in 2018, and today the 17q12 Foundation is run by five motivated parents, all who are volunteering their time. We represent a growing community of individuals who share the same vision: to improve the quality of life of people with chromosome 17q12 syndromes by increasing awareness, advancing research, and providing support.

About National Organization for Rare Disorders, Inc. (NORD®)

The National Organization for Rare Disorders (NORD) is the leading independent advocacy organization representing all patients and families affected by rare diseases in the United States. NORD began as a small group of patient advocates that formed a coalition to unify and mobilize support to pass the Orphan Drug Act of 1983. Since then, the organization has led the way in voicing the needs of the rare disease community, driving supportive policies, furthering education, advancing medical research, and providing patient and family services for those who need them most. Together with over 300 disease-specific member organizations, more than 17,000 Rare Action Network advocates across all 50 states, and national and global partners, NORD delivers on its mission to improve the lives of those impacted by rare diseases. Visit rarediseases.org.

Media Contact:

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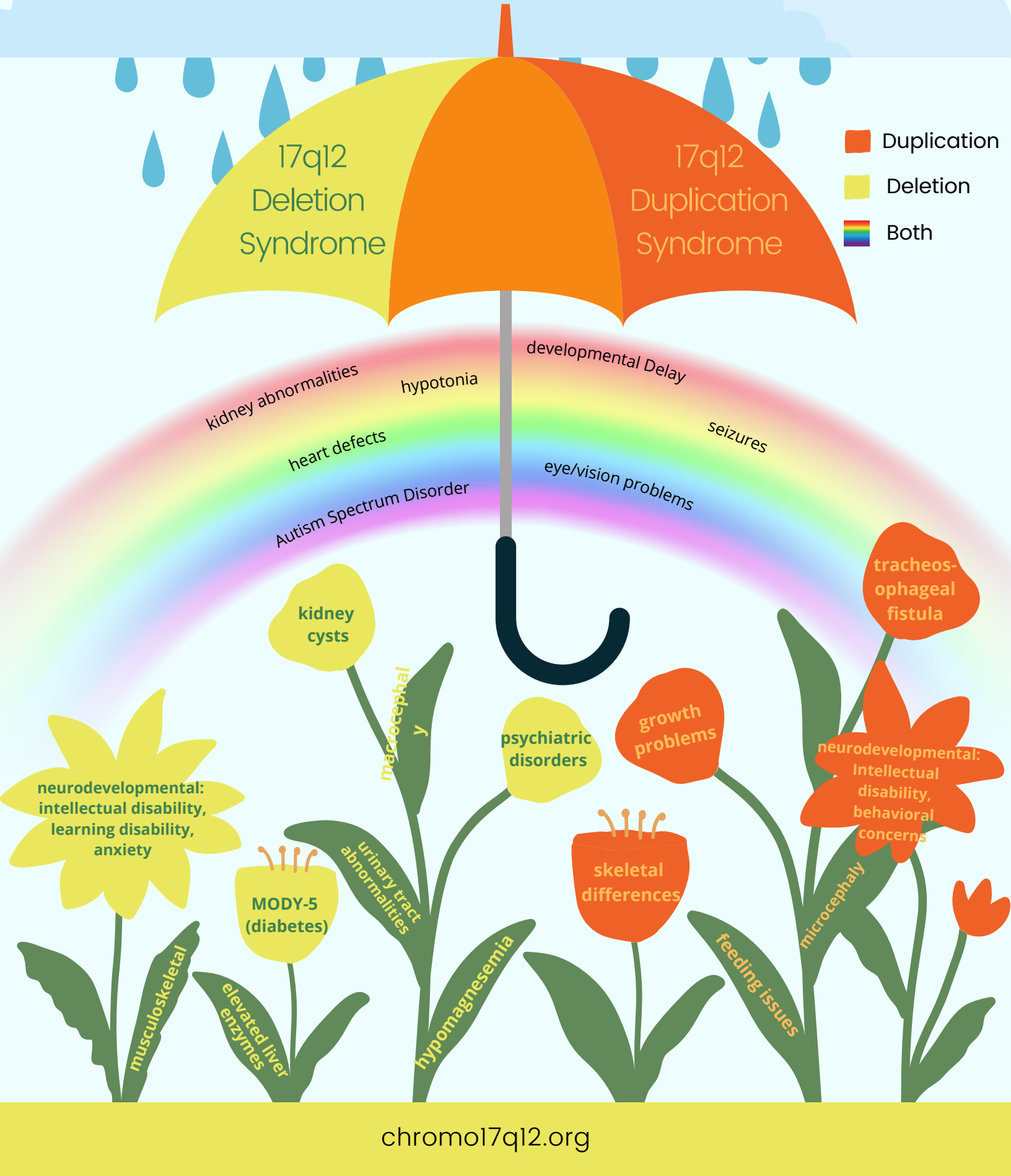
Liz Fourie, liz@chromo17q12.org, 17q12 Foundation

17q12 Registry FAQs

The registry will launch in early 2023!

- The registry is a secure database that will help researchers and clinicians learn more about chromosome 17q12 deletion syndrome and 17q12 duplication syndrome.
- Enrolling in the 17q12 registry is optional and participants can opt out at anytime.
- The 17q12 Foundation will have 100% data ownership, which means we can keep the 17q12 community in the forefront.
- 17q12 individuals are welcome to participate from anywhere in the world.
- The more participants that enroll the more we will learn about 17q12 syndromes.

There are several features and symptoms that fall under 17q12 syndromes. Individuals may have more diagnoses in addition to a 17q12 diagnosis.



17q12 Community

SPOTLIGHT

Joyann is kind, caring, and tries hard at everything. She is very lovable. She also has learning difficulties, balance and coordination issues, hypermobility, and asthma, amongst other things.

Joyann (8), UK
Duplication

Brax has an intellectual disability and has had a lot of support with his education along with an adjusted curriculum. He sees a speech therapist and went from being non verbal til 3 1/2 to having a broad vocabulary with a mild language impediment and lisp. He also works with an occupational therapist as well as a physiotherapist to increase his muscle tone in his legs and torso. Brax wears glasses for his long sightedness and asymmetrical vision. He is currently awaiting results of kidney scans, blood results for diabetes, and testing for epilepsy. After his latest Dr appointment he is also being referred for behavioural therapy, psychotherapy and to a surgeon to correct his umbilical hernia. He is a beautiful boy with a huge heart and oh how that smile melts my heart!

Brax (6), Australia
Duplication

Jason has had a tough year, between transitioning to a new area and struggling to get through the day at school due to pain; he had ended the year well! I am so excited to share that Jason has gone three months without being in a pain cycle! And this is the first time in a while he was able to attend summer camp and make it through without being in immense amount of pain! Thanks to his incredible medical team strides have been made! Also thanks to his drive and not ever willing to give up! We are so proud of him and beyond excited as we take on this next year! Jason has two older siblings at home who continue to support him and cheer him on. Jason does a great job in doing the same for them!We know there will be struggles along the way, but we also know we have an incredible God, an amazing medical team and a phenomenal Team Jason to cheer him on!

Jason (11), USA
Duplication

Zevyn (5), USA, Deletion

Zevyn doesn't need to go back to her urologist and her kidney cysts were not seen on her last ultrasound! She still has grade 1 bilateral hydronephrosis but it doesn't affect her day to day from what we've noticed. She has the okay from her pediatrician to start kinder this fall! This year she did tap, ballet, and soccer and has done so well. Aside from being on the small side, the deletion doesn't affect her. If she had never been tested we never would have known. And I think that just goes to show how wide of a spectrum there is for 17q12 syndromes!

Adaptive Supports

that help the 17q12 community fly!

Zander (11), Deletion
To manage his pain and fatigue, Zander uses a power chair to help conserve his energy and to access his community.

Jason (11), Duplication
Jason is graduating to inserts so they can start working on his ankle strength.

Aksel (5), Duplication
Aksel is non-verbal and has autism. He uses a LAMP words for life device for communication.

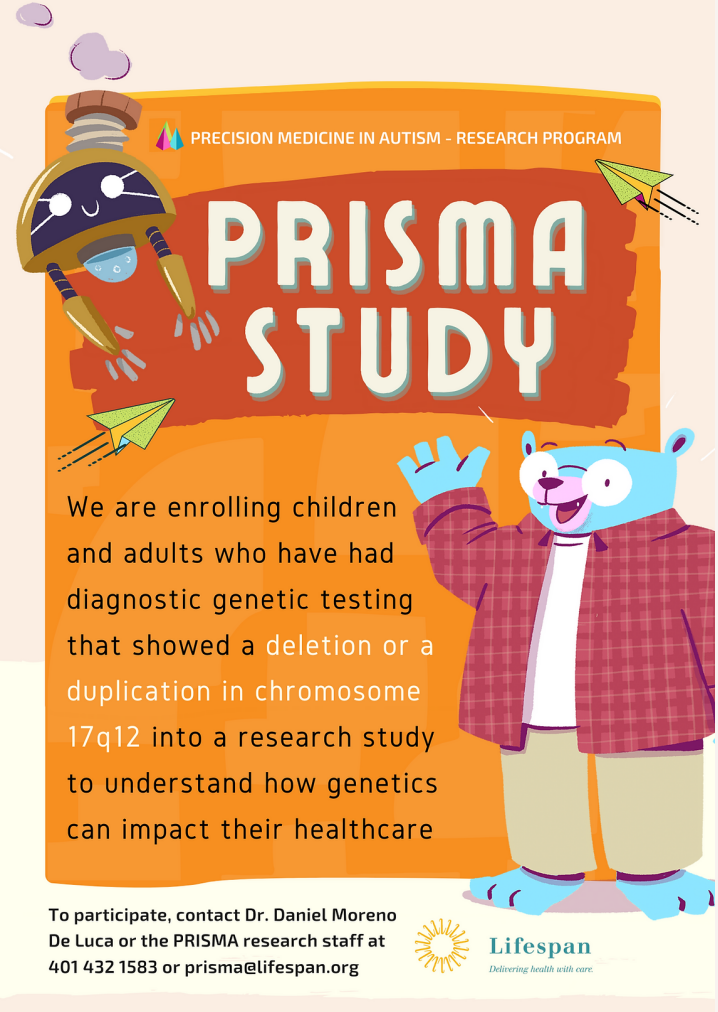
Some of the adaptive supports the 17q12 community uses:

Communication:
- tablet
- visual Aids & charts
- touchscreen
- laptop
- Zoom

Sensory
- fidget toys
- seamless clothing
- chewelry
- noise canceling headphones
- weighted blanket
- brushing
- gloves
- sunglasses
- trampoline
- slime
- hammock, sensory sack
- deep pressure therapy
- movement breaks

Education
- supportive desks/chairs
- timers
- tablets & laptops
- pencil grip
- visual aids/charts
- watch with alerts
- Snap Type Pro
- Voice to text
- Zoom

Mobility
- modified Footwear
- wheelchair/ power chair
- Kizik shoes
- push chair



17q12 Research News

Check out chromo17q12.org/research to read more about the current 17q12 research studies.

17q12 Immunological Study

Dr. Jenny Shin M.D. Ph.D. is an allergy-immunologist, in the laboratory of Dr. Insoo Kang, M.D. (Director of Allergy and Immunology) at the Yale School of Medicine. Her research focuses on the immune system of patients with 17q12 deletion and duplication. Proper immune system function is critical in the defense against bacteria, viruses, parasites, allergy, and inflammation. It is also critical for many other processes like the development and maintenance of the central nervous system, reproductive tract, and digestion. Dr. Shin hopes to understand any changes in function of the immune cells of 17q12 deletion/duplication patients, in relation to their clinical presentation. Participation is simple! You will only need to provide a detailed medical history and a blood sample. They are also looking for blood samples from unaffected children aged 4-18. If you or your child(ren) are interested in participating, please email us at immunodeficiency@yale.edu and fill out this form. Please be sure to include your name, phone number, and an appropriate time for us to contact you.

FUNDRAISING
for 17q12

17q12 Foundation is a registered 501(c)(3) organization and relies heavily on fundraising and community support. Any fundraising big or small makes an impact.

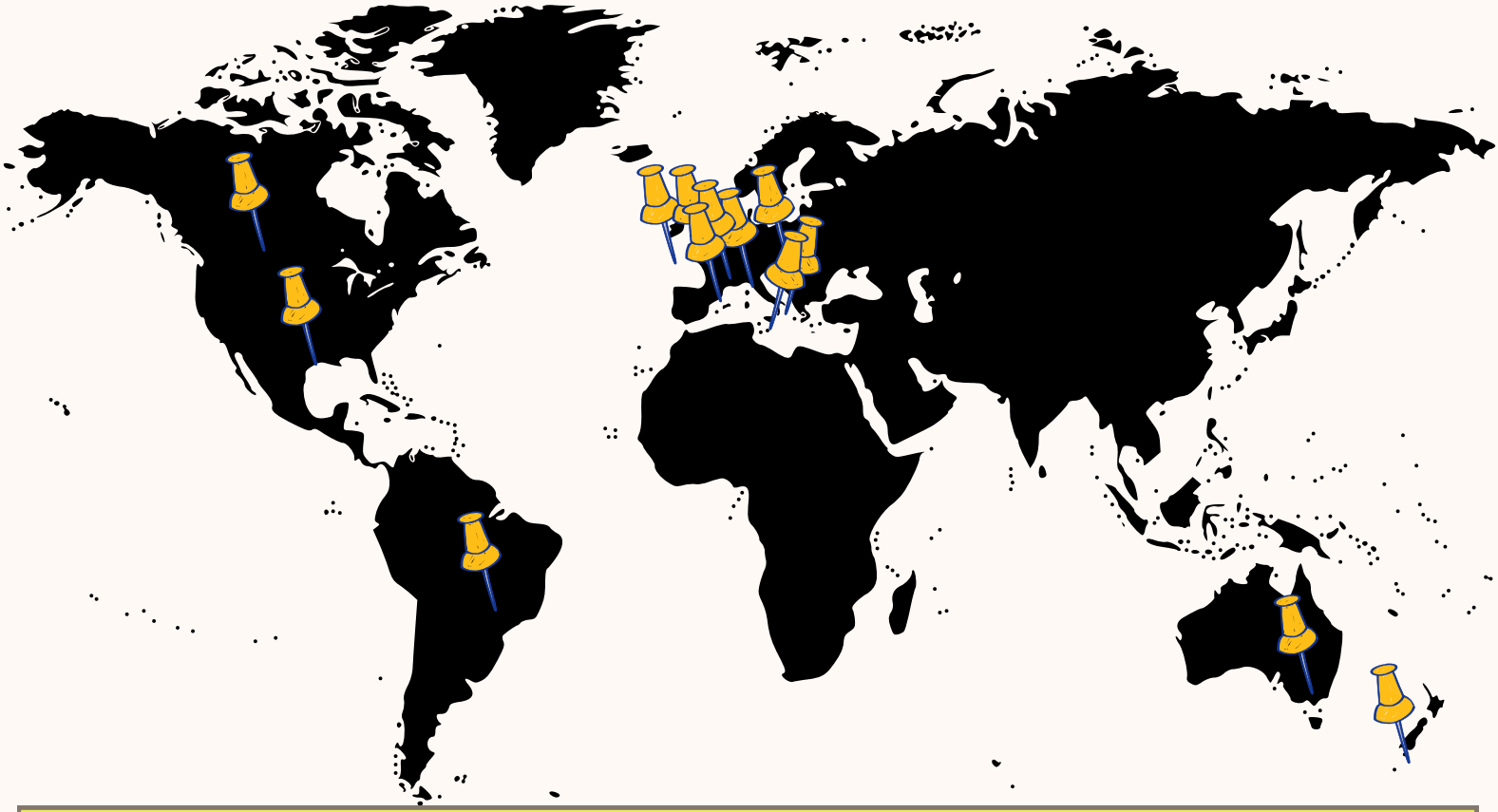
fundraising ideas:

- Dine to Donate: A restaurant (fast food or sit down) has a day and hours set aside and a portion of all food sales will go to the foundation.
- Silent Auction: Ask companies or friends to donate items to sell tickets to win donated items.
- Facebook Fundraisers: "Giving Tuesday" around Thanksgiving and Birthday donations

- garage sales
- lemonade stands
- bake sales
- car washes for 17q12

Please email us at info@chromo17q12.org if you would like to organize a fundraiser. We can provide our tax ID number, advertising, and support your efforts!

WHERE ARE WE FROM?



The 17q12 community is represented around the world

INDIVIDUALS AND FAMILIES WITH A CHROMOSOME 17Q12 SYNDROME CAN JOIN US AT CHROMO17Q12.ORG/JOIN-US

We currently have 93 Verified members from the Following countries:

213 Membership Registration forms have been submitted.

There are 231 members in our private support group on Facebook.