17q12

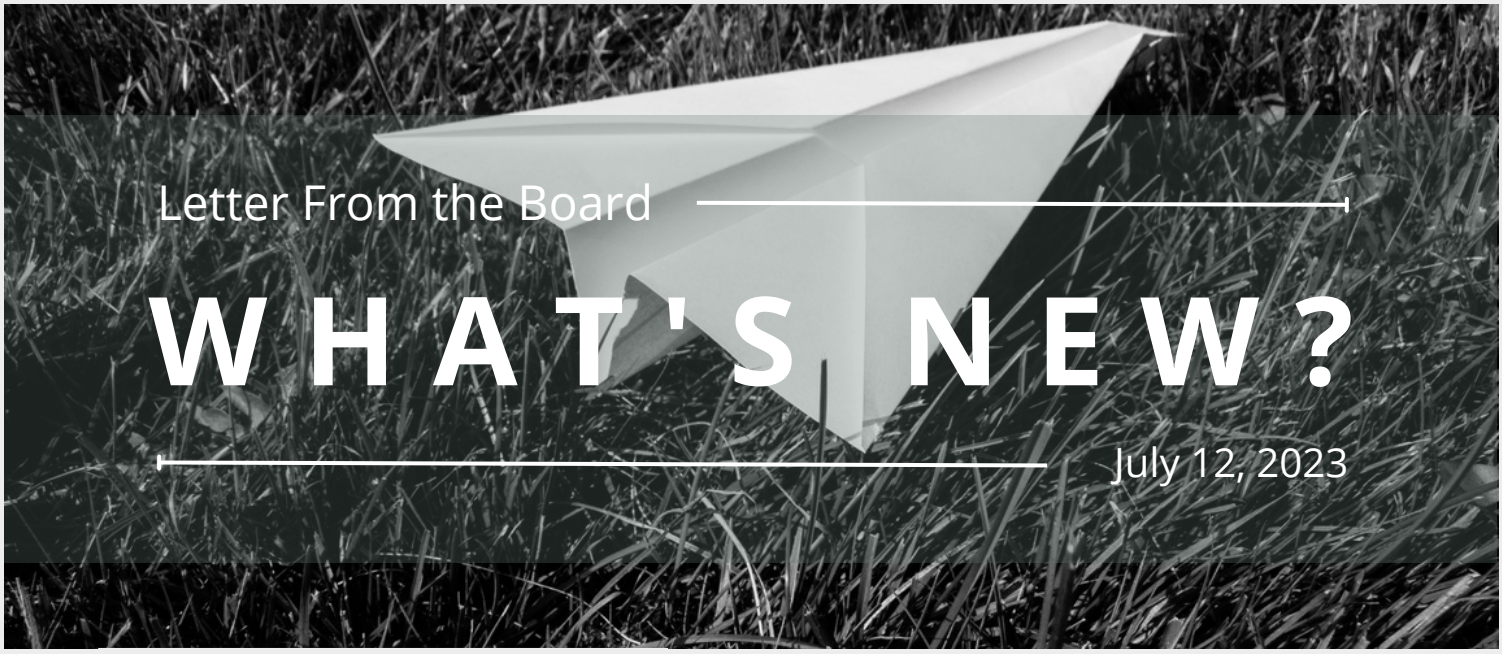
NEWSLETTER

A Resource for Chromosome 17q12 Deletion and Duplication Syndromes

CHROMO17Q12.ORG

17q12 FOUNDATION

JULY 2023



Letter From the Board

WHAT'S NEW?

July 12, 2023

Dear 17q12 Community,

Thank you for helping us celebrate our fifth annual 17q12 awareness day! We have catapulted into an exciting chapter for individuals with chromosome 17q12 syndromes. This past year we have been working tirelessly on building our medical registry. We've made great progress while also building connections in the rare community. Our member community has grown, and we have welcomed new families and individuals from numerous countries. We had the opportunity to hold more virtual meet-ups, as well as launching a new program, 17q12 Near You, where we matched verified members that were geographically close. To say we are excited for the future is an understatement. Thank you for supporting the 17q12 Foundation and our community as we continue to learn, grow, and advocate together!

Allaina Wellman
President

Liz Fourie
Vice President

Sherie Scott
Secretary

Mark Dempsey
Treasurer

Margo Casados
Board Member

We stick together like peanut butter & jelly!

The 17q12 Foundation represents two separate syndromes on Chromosome 17q12. Each syndrome presents differently, but put together we are stronger.

BOTH SYNDROMES:

eye/vision problems

developmental delay

Autism Spectrum Disorder

seizures

hypotonia

HEART DEFECTS

kidney abnormalities

Community SPOTLIGHT

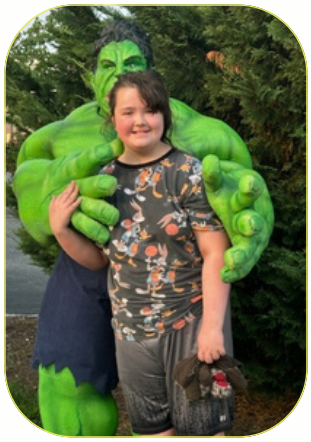
Chloe (4), USA, Deletion

Chloe was born with cysts on her kidneys and she sees a nephrologist once a year, along with a team of other doctors. Joint laxity limits a lot of her physical activities. She was diagnosed with 17q12 deletion syndrome at 6 months old. Her personality is her greatest strength. Chloe is funny, outspoken and loves to sing and dance.

Wesson (15 mos), USA, Duplication

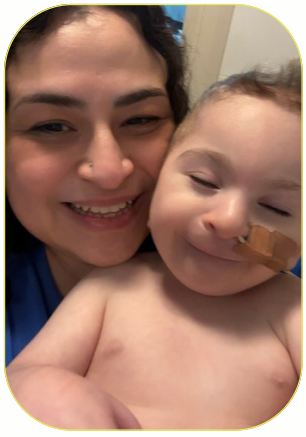
Wesson has GDD, Hypotonia, Tracheomalacia, Eczema, language disorder, behaviors, feeding difficulties, Oropharyngeal Dysphasia, abnormal MRI, sleep issues. along with a few more issues. He is awaiting an Autism eval. It's very hard some days but both him and his mom are strong willed, and he is her heart. He was 9 months old and she was 36 years old when diagnosed. His greatest strength is that he is a warrior and strong willed.

Victoria (11), USA, Deletion



The deletion mostly affects Victoria with autism, ADHD, behavioral issues, skin issues, sensory disorders, hypotonia, and a weakened left leg, which makes her prone to tripping/falling a lot. Victoria was almost 2 years old when she received her diagnosis. Her greatest strength is that she is persistent. If there is a way to get it done, she will do it.

Deniz (2), Australia, Duplication



The duplication has affected Deniz with a number of disabilities: feeding dysphasia, Coloboma, hearing loss, microcephaly, global developmental delay, thoracic scoliosis, undescended testes, and a PDA device in his heart. At 16 weeks pregnant his mom's ultrasounds showed low fluid. The results of the amniocentesis showed 17q12 duplication. Deniz has a high pain threshold, is always happy, and barely cries. He is his family's little miracle.

17q12 Patient Registry

BUILDING PROGRESS

July 2022

The onboarding process began with NORD®(National Organization for Rare Disorders). Three 17q12 Foundation board members have met weekly with NORD and other rare organizations that are going through the same process.

September 2022

We formed our Registry Advisory Board that oversees the conduct of the study. It is comprised of a team of individuals with 17q12, parents, doctors, and researchers working with 17q12 syndromes; all of whom are donating their time and expertise.

October 2022

The first meeting with the Registry Advisory Board was held with the 17q12 Foundation where we began to collaborate and assigned important roles.

November 2022

We began the process of creating all of the important documents and surveys that make-up the 17q12 Patient Registry. This included months of reviewing and revising the documents with both NORD and various specialists. The 17q12 Foundation collaborated with doctors from across the country, such as Yale, Baylor College of Medicine, UCLA, and Connecticut Children's Medical Center.

June 2023

NORD began the building process of the 17q12 Patient Registry through their registry platform, IAMRARE®. Once it is completed we will begin a very thorough testing phase.

July 2023

The 17q12 Foundation registry team is continuing to meet weekly while we work hard on the remaining steps needed to launch; including completing Good Clinical Practice Training. Working with NORD has enabled us to continue to keep the 17q12 community in the forefront as we approach this next chapter. We can't wait to launch!

CELEBRATING ACHIEVEMENTS

DELILAH BELLE, AGE 7

USA (DEL)

Delilah Belle was the youngest member elected into her school's chorus. She just finished Kindergarten. She was also the only student to have a solo performance at the end of the year recital singing "Big Dreams".

STEPHEN, AGE 29

USA (DEL)

Stephen had a blast at the Summer Bash Party put on by the Therapeutic Recreation Department for the young adult social club that he's in. Twice a month he attends social outings like bowling, going to a movie, a park, out to dinner, and more. He LOVES his Social Club!

JOYANN, AGE 9

UK (DUP)

Joyann just recently moved up a level in both Math and Spelling!

QUINN, AGE 22

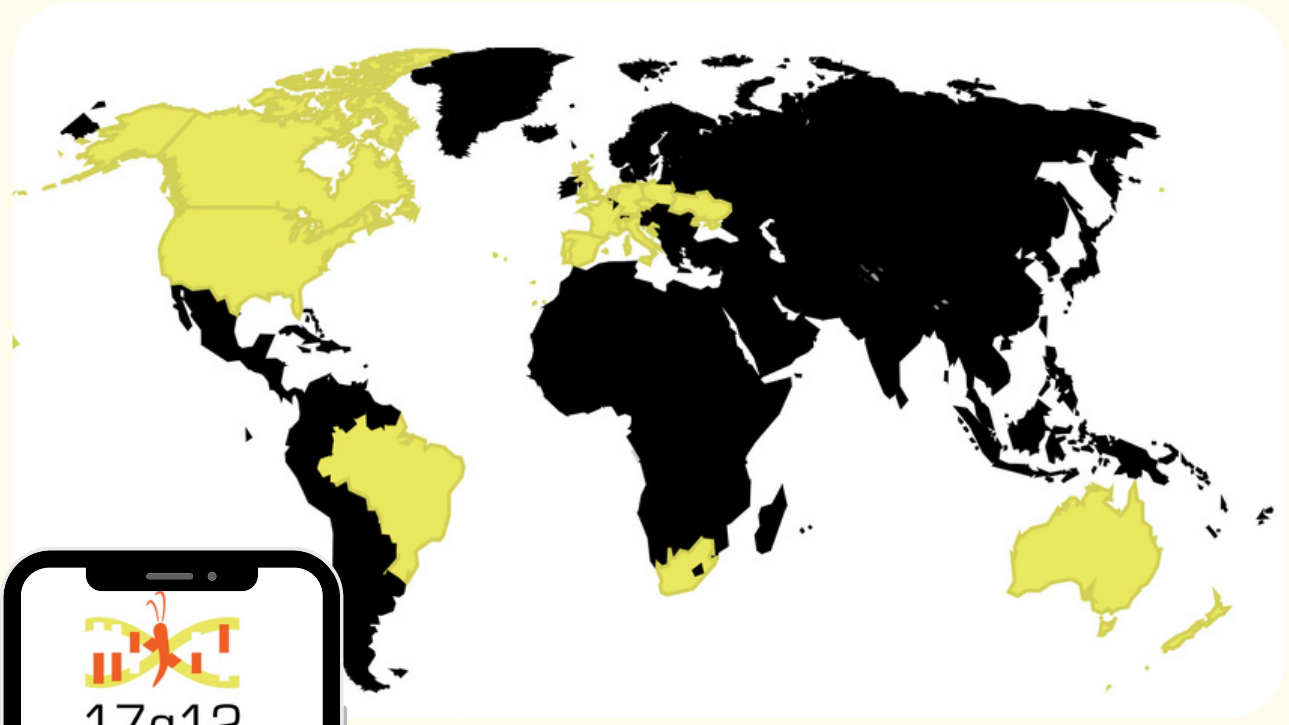
CANADA (DEL)

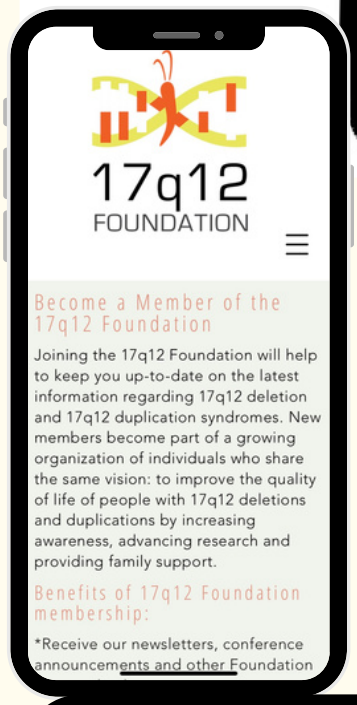
Quinn has started fundraising for charities, and is making great progress with her first fundraising project.

AKSEL, AGE 6

USA (DUP)

Aksel got his feeding tube out this year after 6yrs of having one. For several years in the beginning, He was tpn and j tube fed. A few years ago he graduated to just a g tube. He has overcome intestinal failure after two intestinal surgeries. He has maintained his weight for over a yr. He graduated from three of his specialist doctors this year and is soaring to a healthy quality of life. He is pictured here with Dr. Rueven Zev Cohen from CHOA GI clinic, a doctor that helped save his life over 4yrs ago. Aksel is 17q12 strong!





JOIN US!

As of July 2023 we have verified members in 17 different countries around the world! Registering as a member of the 17q12 Foundation, allows the 17q12 community the opportunity to join forces. It enables the 17q12 Foundation to keep our community that is spread out around the world updated with important news; from research opportunities to registry updates. It will also help us form a support network by connecting the 17q12 family with other verified members in the same geographical locations. If you or your child have a diagnosis of chromosome 17q12 deletion or duplication syndrome please connect with us at **chromo17q12.org/join-us** .

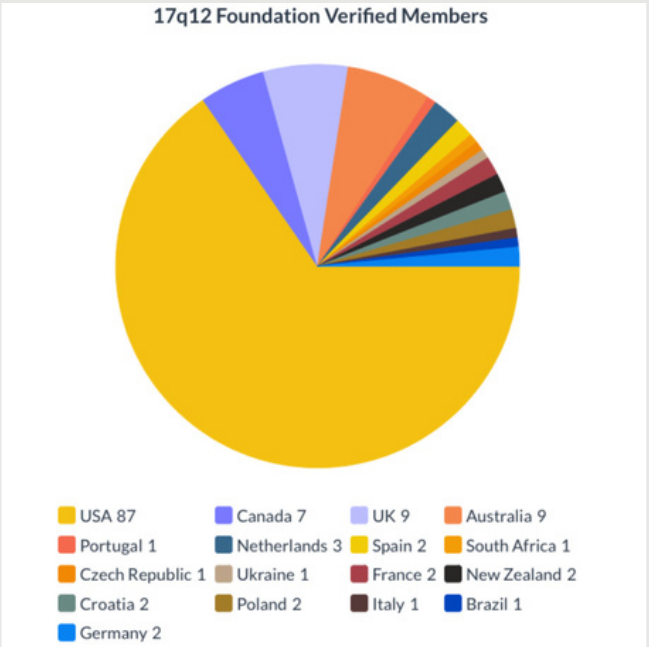
WHO ARE WE?

We currently have 133 verified members/families from the following countries:

75 verified members have 17q12 deletion.
58 verified members have 17q12 duplication.

280 Membership Registration forms have been submitted.

There are 256 verified members in our private support group on Facebook for parents, caregivers, and individuals directly affected by a 17q12 syndrome.



Research Announcements

Dr. Pengfei Liu's study

Pengfei Liu, Ph.d. Molecular and Human Genetics
Baylor College of Medicine

There is an IRB-approved study at Baylor College of Medicine led by Dr. Pengfei Liu that focuses on providing a comprehensive and precise diagnosis for the genetic condition of 17q12 in you or your family member. Participating involves collecting blood samples from you and your family member including your medical information. Genome sequencing will be performed on the samples collected. Analysis of the sequence data will be done to understand the cause of your genetic condition. If you are interested in study participation, please contact us at precisiongenome@bcm.edu. Please provide your full name, phone number, e-mail address, and good day and time for contact.

Enrollment requirements:

- Individuals with the 17q12 deletion.
- Individuals with the 17q12 duplication who have either renal, endocrine, or reproductive system issues.

17q12 Immunological Study

Junghee Jenny Shin, M.D., Ph.D.
Assistant Professor of Medicine
Yale University School of Medicine

The goal of our proposal is thus to test the overarching hypothesis that patients with the 17q12DS have altered T cell immune responses. I am studying the characteristics of T cells in patients with the 17q12 deletion or duplication syndrome using conventional and high-dimensional analyses and relate such immune alteration with genetic defects in these patients.

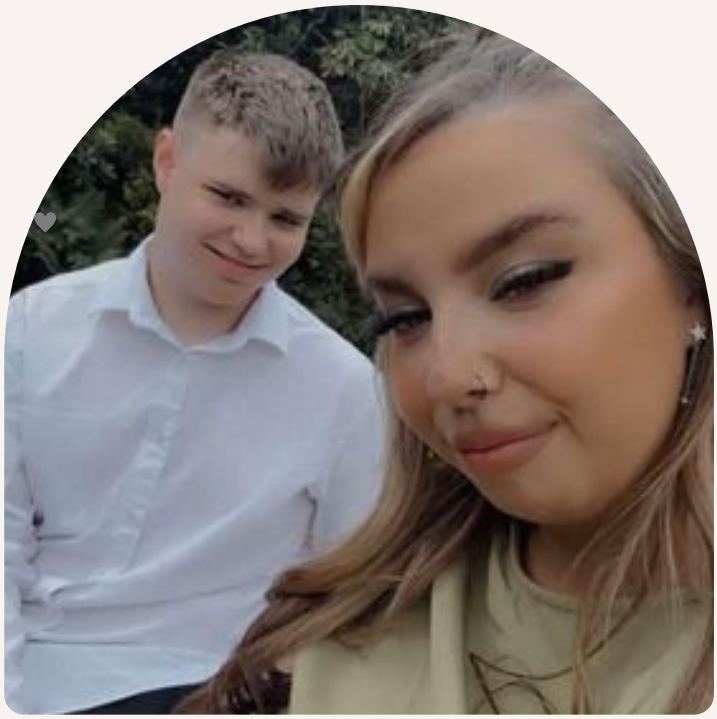
We recruited 36 patients with 17q12 DS as of now and collected their medical histories including allergies, infections and autoimmunity. As expected, majority of patients presented with well-known clinical phenotypes of 17q12 DS including renal diseases and neuropsychiatric disorders. Of interest, around 60% of patients presented with recurrent infection. We are investigating alteration of T-cell characteristics and functions in 17q12 DS using single cell RNA sequencing, flow cytometric analysis and multiplex cytokine array.

The proposed study may uncover a new immune regulatory mechanism broadly applicable to immune-mediated disorders while directly informing precision therapy for patients with 17q12 deletion or duplication syndrome.

Read more about research opportunities available to the 17q12 community here:

chromo17q12.org/research

Soaring Siblings



KYLE & CAITLIN

Meet Kyle and Caitlin. Kyle is 13 and has 17q12 duplication syndrome. His sister, Caitlin, is 21. Kyle loves spending time with Caitlin, listening to music, and a good gossip. Kyle has taught Caitlin patience is a must. She now works with children with disabilities.

DELILAH BELLE, JUNIOR, & JA'SIAH

Meet Delilah Belle (7) and her brothers, Junior (9) and Ja'Siah (5). Delilah Belle has 17q12 deletion syndrome. Their favorite thing to do together is wearing matching outfits and going to the Sonic Sand Park and trampoline park together. Her brothers have taught her how to use the YouTube microphone to look up her favorite videos.

OUR FIVE YEAR JOURNEY

Thank you for helping us reach our goals in five short years.
Let's see what we can accomplish together in the next five!

WHERE IT ALL BEGAN
JULY 2017

Four families and a genetic counselor met at a family conference for 17q12 syndromes in Chicago, IL. We all were motivated to form an organization for 17q12.

IT'S OFFICIAL!
JANUARY 2018

In 2018 we became a registered 501(c)(3) non-profit organization, and the 17q12 Foundation was born!

FIRST AWARENESS DAY
JULY 2019

We celebrated our first annual 17q12 Awareness Day on July 12, 2019, and launched a #HowWeFly campaign to help put a face to 17q12 syndromes.

FAMILY CONFERENCE
AUGUST 2019

The 17q12 community had the opportunity to gather at our first family conference as an official group in Providence, Rhode Island.

REGISTRY PROCESS BEGAN
JULY 2022

After much deliberation in choosing the right platform, we began the onboarding process to build a much needed registry for 17q12 syndromes.

REGISTRY GOAL MET
FEBRUARY 2020

We met our goal of raising $15,000 to fund a medical registry for 17q12 thanks to fundraisers such as the Casados family art auction, Josh's L2H hike for 17q12, and generous donations from the Artamenko family and the Palisades Foundation.

A YEAR IN REVIEW

A look into the funds we raised and spent in 2022

HOW WE RAISED

We received $4782 in donations through:

- Facebook fundraisers
- Direct donations
- Our annual Bonfire shirt campaign
- AmazonSmile

HOW WE SPENT

We spent $9660.22 on administration and registry costs that included:

- Zoom plan
- Annual website costs
- Google Voice
- insurance
- NORD annual membership fee
- Registry costs

Gold Transparency 2023

Candid.

Fundraising for 17q12

Every dollar raised helps us support the chromosome 17q12 community.

Our fundraising goal for this year is to cover the annual maintenance costs for the 17q12 registry and to fund a much needed family conference. With the registry getting closer to launching and our community growing, connection is needed now more than ever. If we work together we can fundraise enough to reach this goal.

Do you have an idea for a fundraiser? No fundraiser is too small. 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

- Birthday Fundraisers on Facebook
- Run your own Bonfire shirt campaign
- Plan a silent auction
- Organize a bake sale
- Ask a local restaurant about Dine & Donate
- Hold a car wash or garage sale

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!

#HOWWEFLY

Happy 17q12 Awareness Day!