17q12 Foundation NEWSLETTER

a resource for chromosome 17q12 deletion and duplication syndromes

JULY 2021

17q12 Family

To our 17q12 community,

We are thrilled to be celebrating our third annual awareness day with all of you. WIthin the last year we have welcomed many new members to the 17q12 family from around the world. After taking the needed steps to find the right platform for the 17q12 medical registry, we are now in the final stages of moving forward. We missed connecting in person at another family conference this year due to the pandemic, but we have begun discussing the idea of a virtual conference. Our goals for this year are to launch the medical registry, create a natural history study to better understand 17q12 duplications and deletions, and to continue to support and advocate for both our current and future members as we grow. Thank you to all of our supporters that have generously donated and supported us this last year!

Meet Margo, our newest board member!

My son Sebastian (8) was diagnosed with the 17q12 deletion when he was 5. The diagnosis answered important questions for us, but also made us aware of other health issues we didn't realize that Sebastian had until we screened him. We attended the 2019 Family Conference and it was a great relief to get more information and meet other families. We felt it was important to get more involved and subsequently hosted an art auction to raise money for the 17q12 patient registry, and have been working with Drs at Yale to help start a research project to study the immune system of patients with 17q12 deletion/duplication. I've worked in tech & marketing for 20 years and hope to use my skills to be of help to the 17q12 Foundation board and community.

GET TO KNOW US

Allaina Wellman
President

What does 17q12 Foundation mean to you? The Foundation means hope to me, hope for our children and future children. Hope for answers and research. Hope is everything, the foundation is everything.
What activity helped get your family through COVID quarantine? Arts and Crafts; we did it all!
If you could be any hero, who would you be? Hermione Granger, she's wicked smart, always has the answer and gets to run around with lots of books. What more could you want?

Liz Fourie
Vice President

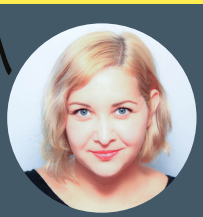
What does 17q12 Foundation mean to you? It means community and pushing forward for a better understanding of 17q12 disorders.
What activity helped get your family through COVID quarantine? Watching all of the caterpillars turn into butterflies in our yard
If you could be any hero, who would you be? Black Widow

Sherie Scott
Secretary

What does 17q12 Foundation mean to you? The foundation means community and a sense of belonging as well as knowing we can make a difference.
What activity helped get your family through COVID quarantine? Family Game Nights!
If you could be any hero, who would you be? Stormy from the X-Men

Mark Dempsey
Treasurer

What does 17q12 Foundation mean to you? The foundation is meaningful to me because it's a group of (mainly) parents working for the betterment of their children
What activity helped get your family through COVID quarantine? visiting the Buffalo at the Nature Center.
If you could be any hero, who would you be? Superman



Margo Casados
Board member

What does 17q12 Foundation mean to you? To me, it means wonderful possibilities - for research, answers, treatments, healing, community, and of a better life for all of us.
What activity helped get your family through COVID quarantine? Baking! We love to bake normally but we did a lot during quarantine.
If you could be any hero, who would you be? Captain Marvel because she has superhuman strength, healing powers, & I've always wanted to fly fighter jets!

WE REPRESENT TWO SEPERATE SYNDROMES

17q12 DELETION syndrome

- Neurodevelopmental: intellectual disability, learning disability, anxiety
- MODY-5 (diabetes)
- Kidney cysts
- Psychiatric Disorders
  - Macrocephaly
    - urinary tract abnormalities, abnormalities of the female reproductive system (most often an absent or abnormal uturus)
    - Hypomagnesemia
    - Elevated liver enzymes
    - Musculoskeletal
      - most often de-novo (new occurance)

BOTH syndromes

- Kidney abnormalities
- autism spectrum disorder
- developmental delay
- seizures
- Eye/Vision problems
- Heart Defects
- Hypotonia

17q12 DUPLICATION syndrome

- Neurodevelopmental: intellectual abilities ranging from typical to severe disability, behavioral concerns (aggression, compulsive disorders)
- Microcephaly
- feeding issues
- growth problems
- Skeletal differences
- Tracheoesophageal fistula
- most often inherited from a parent

17q12 community spotlight

Deniz (6 months), Australia, Duplication

My son has the 17q12 duplication and has had a lot of challenges since he was born. The biggest problem we have faced since his birth is his feeding.
He has dysphasia and has coordination difficulty swallowing.
His biggest strength is that he is such a happy baby and no matter how much of a painful procedure he endures, the moment after I settle him, he gives me a great big smile like nothing happened. He is my hero

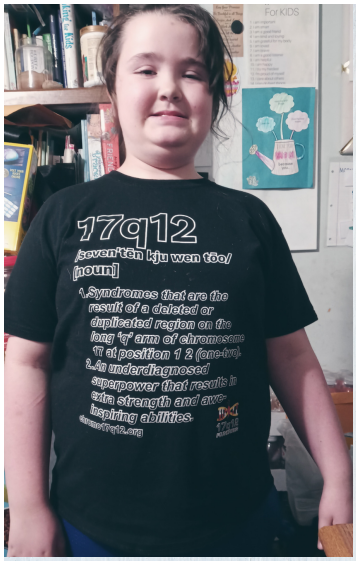
.

Joyann (8), UK, Duplication

This is Joyann, nearly 8 years old. She has 17q12 duplication syndrome. She has secondary microcephaly, learning difficulties, hypermobility, asthma, and is short-sighted. She tries very hard with everything, and is absolutely amazing

Luca(15), Imara(12), Nova-Mae(8), & Alrieke (42); The Netherlands, Duplication

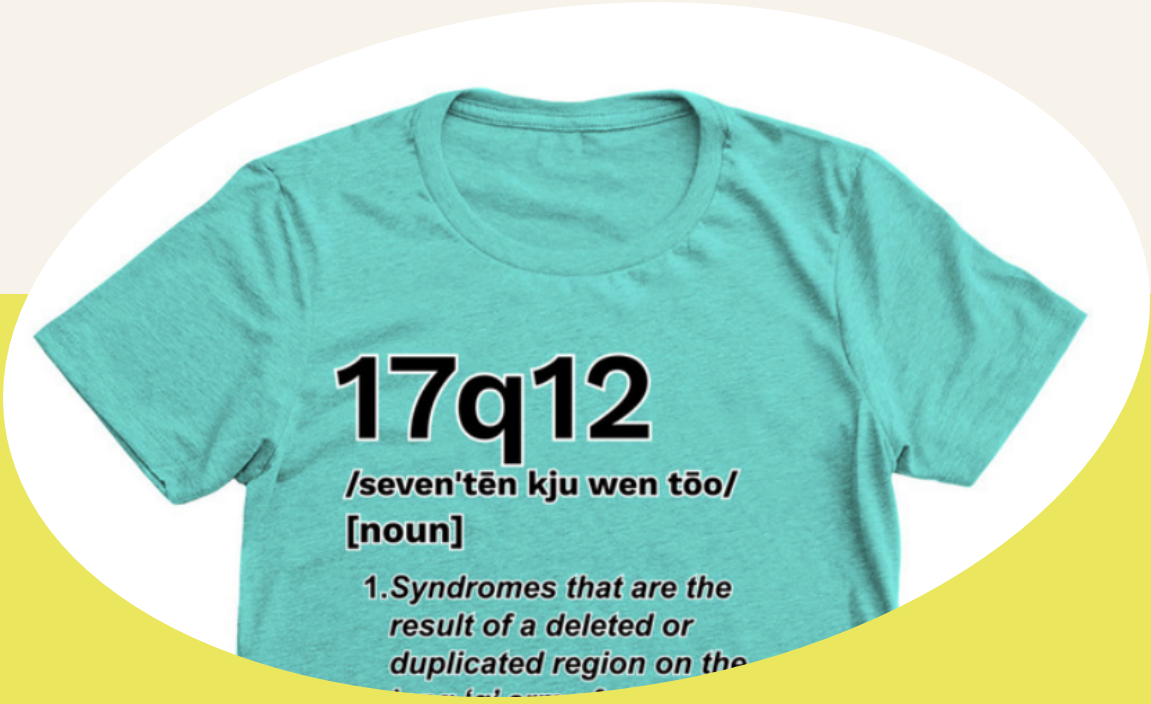
All 4 of us have 17q12 microduplication syndrome. We have known since 2019. It expresses itself with us with anxiety disorders, autism traits, scoliosis, and mild intellectual disabilities in Imara; learning problems in Nova-Mae; kidney problems, reflux, kidney cysts in Alrieke; intestinal problems, eye abnormalities, and hip abnormalities in Luca. I am grateful as a mother that I have it too so that I can better understand my children. We are proud of who we are!



Victoria (9), US, Deletion

Victoria is affected by 17q12 in a multitude of ways. She is afflicted with Autism, ADHD, OCD, behavioral issues, hypotonia, severe anxiety, skin issues- very dry and "chicken bump" skin. There are medical issues as well. Victoria is very a funny child who loves music and art.

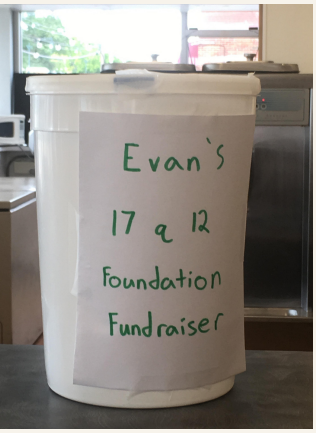
FUNDRAISE FOR 17Q12



DO YOU HAVE A FUNDRAISER IDEA?

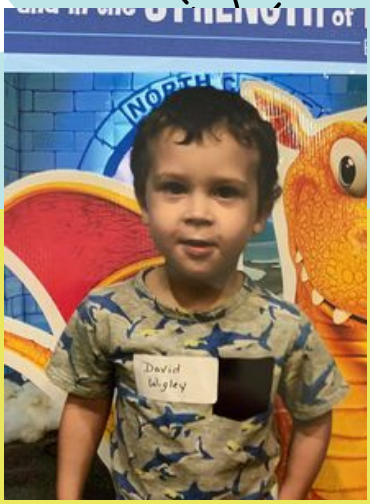
Contact us at chromo17q12.org and we can help you get started!

As our community grows, the need for support becomes greater. Currently, our biggest project is preparing to launch our medical registry. There will be annual maintenance costs that 17q12 Foundation will be responsible for. There is also a need for connection, now more than ever, and we hope to make a family conference possible in the future when it is safe for our community to gather. 17q12 Foundation is 100% volunteer run, parent and patient led, and focused entirely on supporting individuals and families with chromosome 17q12 deletions and duplications. Please consider fundraising for 17q12 to allow us to continue to support, advocate, and grow!



celebrating our achievements

#HowWeFly



Davey (3y)
MS, USA
Davey went to vacation Bible school and had a blast. For the first time, he said two words together, “ball kick!” His mom is so proud!

Joyann (8y)
UK
Joyann received a certificate from school because she is very kind.

Abigail (3y)
TN, USA
She’s doing really well with new words and phrases and is now her family's little mimic! She LOVES to sing!

James (15mo)
NC, USA
His cardiologist says he's recovering well after open heart surgery. He is starting to love walking while holding someone's hand!

Zevyn (4y)
CA, USA
Zevyn learned how to ride her tricycle!

Delilah Belle (5y)
NC, USA
For 6 months she practiced with her mom's help to climb the park obstacle, This past weekend she did it all by herself. Thanks to physical therapy and her determination to succeed.

Isaiah (2y)
MO, USA
Isaiah is nonverbal and just communicated with his family for the first time. He did his first sign and signed, "More!"

Researcher Q&A

Learn about 17q12 research studies & the researchers behind them

Pengfei Liu, Ph.d.

Molecular and Human Genetics

Baylor College of Medicine

What is your specialty or area of focus?

I am a clinical laboratory diagnostician by training. My research focuses on studying the cause and impact of genomic deletions and duplications to genetic disorders.

How did you become interested in 17q12 syndromes?

I work in a diagnostic lab where my clinical duty is to make molecular diagnoses for individuals with genetic disorders, including those caused by all kinds of genomic deletions and duplications. I also have past research experiences working with individuals carrying a few other deletions/duplications, for example, the Smith Magenis deletion syndrome, the Potocki-Lupski duplication syndrome, the CMT duplication syndrome, and the 16p11.2 deletion/duplication syndromes. What strikes me the most for the 17q12 syndromes is the high degree of clinical variability and unpredictability. This is a phenomenon observed in almost all genomic deletions/duplications, but the cause of this phenomenon is not understood. The 17q12 region is unique in that it is associated with perhaps one of the widest ranges of clinical manifestations among other genomic disorders. Despite this, relatively few research endeavors have been devoted to this genomic region. I believe studying the genetics of the 17q12 syndromes will not only benefit individuals carrying these deletions and duplication, but it will also help individuals carrying other deletions/duplications better predict their clinical outcomes and understand the variability of their conditions.

What does your research project focus on specifically for 17q12?

Our long term research project aims to understand the molecular mechanism that determines the clinical variability of the 17q12 syndromes. Our current NIH-funded project focuses on individuals with the 17q12 deletions and individuals with the 17q12 duplication who have either renal, endocrine, or reproductive system issues.

How will your research project benefit the 17q12 community?

Our research project will directly impact the precision diagnosis of the 17q12 conditions. A long-term benefit is that a precise understanding of the mechanism of disease presentation will guide development of precision, personalized treatments in the future.

Researcher Q&A

Daniel Moreno De Luca MD MSc

PRISMA Research Group
Bradley Hospital
Brown University

What is your specialty or area of focus?

I am a physician and scientist specializing in child, adolescent, and adult psychiatry with advanced training in neurogenetics. I split my time between clinical care, where I see people with autism spectrum disorder, bipolar disorder, schizophrenia, or other mental health needs who also have an underlying genetic cause for these, and research on the rare genetic causes of mental health conditions and how we can use this information to provide more specific clinical recommendations.

What does your research project focus on specifically for 17q12?

At PRISMA, our research group at Bradley Hospital and Brown University, we are focused on understanding the mental health and medical "blueprint" that is associated with 17q12 CNVs, as well as other background genetic factors that may influence whether someone with a 17q12 CNV develops any of the clinical features that have been previously described in this population. We particularly want to focus on moving to "higher resolution" behavioral and medical diagnoses – for example, learning not only how common is a diagnosis like autism spectrum disorder in people with 17q12 deletions, but more importantly, how do people with 17q12 CNVs do on the social functioning scale regardless of whether they meet criteria for an autism diagnosis. Likewise, we want to understand how common a diagnosis like diabetes is, but also more importantly, where do the blood sugar values lie over time for people with 17q12 CNVs irrespective of whether they have a diagnosis of diabetes. This will ultimately allow us to move from a yes/no diagnosis, where a clinical condition is either present or absent, to a nuanced understanding of these clinical areas that tell us about people's areas of strength and challenges. A simple but useful example would be to compare this to boiling water for a nice cup of tea; moving from asking whether water is boiling at any given time (like a yes/no clinical diagnosis), to knowing the actual temperature of the water ("high resolution" quantitative, continuous traits like sociability or blood sugar levels). Lastly, we want to know the genetic factors in the rest of the genome that may contribute to someone developing these diagnoses, even if they all have a 17q12 CNV. Going back to our boiling water example, we want to understand why water boils more easily in some conditions like high altitude above sea level (background genetic factors, in our analogy), even if we have the same temperature for all (17q12 CNVs in this example).

How did you become interested in 17q12 syndromes?

I first became interested in 17q12 CNVs (copy number variants) when I was a postdoctoral fellow in neurogenetics at Emory University, when we started seeing that some people with autism or developmental disabilities that were sent for clinical genetic testing were being diagnosed with a 17q12 deletion or duplication. Up until then, these mental health diagnoses had not been previously described in people with 17q12 CNVs, and by launching a large international study that included over 70,000 people worldwide, we were able to show that 17q12 deletions increased risk for autism and schizophrenia. I had the pleasure of meeting several families with 17q12 CNVs, and this was the most rewarding aspect of this research and what got me most interested in choosing to work on understanding the clinical consequences of this rare genetic change; I very much treasure the life stories that they have allowed me to learn about and the long-lasting bonds we have established with a group of families that have now blossomed and grown into the 17q12 Foundation.

How will your research project benefit the 17q12 community?

Our hope is that by understanding the areas of clinical strengths and challenges regarding mental health and other medical diagnoses and their additional genetic influences in people with 17q12 CNVs, we will be better able to individualize treatment for those who need it, a prime example of what we call Precision Medicine. In fact, we have created the Genomic Psychiatry Consultation Service at Bradley Hospital and Brown University, where we see people with mental health conditions who have had genetic testing that revealed a genetic cause for this (including 17q12 CNVs, and other genetic changes) to provide actionable clinical recommendations based on this genetic information. The more we learn with studies like the one we are carrying out, the more specific the knowledge and medical help we can then transmit back to families in the future! If you or anyone in your family has a 17q12 CNV and is interested in learning more, please don't hesitate to contact us at daniel_moreno_de_luca@brown.edu or at prisma@lifespan.org

Researcher Q&A

Junghee Jenny Shin, M.D., Ph.D.

Assistant Professor of Medicine

Yale University School of Medicine

What is your specialty or area of focus?

I completed Allergy and Clinical Immunology fellowship training (2017-2020) at Yale and joined the Yale Section of Rheumatology, Allergy and Immunology as a faculty in 2020. As an Allergy and Immunology specialist, I care for patients with a wide range of allergic and immunologic diseases. I also study the immune system of patients with immune deficiency and/or immune dysregulation using in-depth immune profiling techniques. My overall research goal is understanding how immune alterations occur and affect the pathogenesis of such disorders in relation to their genetic defects, clinical presentations and comorbidities.

How did you become interested in 17q12 syndromes?

As an Allergy and Clinical Immunology fellow at Yale, I had a privilege to provide care for a five-year old patient with the 17q12 deletion syndrome (17q12DS) since 2019. He presented with the well-known symptoms of the 17q12DS including renal cysts and autism but he also presented with frequent anaphylactic reactions to multiple foods, recurrent infections, and joint pain/laxity limiting his physical activities suggesting the possible dysregulation of the immune system, especially T cells which play a critical role in allergies, microbial defense and autoimmune diseases. Therefore, I started to investigate T cell immune responses in patients with 17q12 deletion or duplication syndrome.

What does your research project focus on specifically for 17q12?

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.

How will your research project benefit the 17q12 community?

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.

If you are interested in learning more about these research studies or would like to sign up please head over to chromo17q12.org/research

Become a 17q12 Foundation Member

REGISTERED MEMBERS

There have been 159 membership forms submitted as of July 2021. From the following countries: US, Australia, UK, Canada, The Netherlands, New Zealand, Israel, Poland, Italy, Spain, Canada, Germany, Austria, and Brazil.

VERIFIED MEMBERS

We currently have 67 members with a verified 17q12 deletion or duplication diagnosis. Verifying our members makes it possible for us to tell interested researchers how many deletion/duplication members we represent, and it helps us manage our private support group on Facebook for those directly affected by the same 17q12 syndromes.

SUPPORT GROUP

Our private support group on Facebook currently has 210 members. Through our private support group we are able to offer each other support in a supportive and positive space. The group is managed by all of our board members, that are also 17q12 parents, and we recently have had the opportunity to connect via zoom for virtual meet-ups.

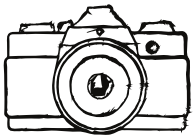
BENEFITS OF BECOMING A MEMBER

Registering as a member with the 17q12 Foundation, allows the 17q12 community the opportunity to join forces. Becoming a member makes it easier for the 17q12 Foundation to keep our community 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 members in the same geographical locations. If you or your child have a diagnosis of chromosome 17q12 duplication or deletion syndrome please connect with us at **chromo17q12.org/join-us**

IEP One-Pager

learn more at chromo17q12.org

name: _____



Place your 17q-tie's photo here

Vision statement:

_____

_____

I'M WORKING ON:

-
-
-
-
-

MY STRENGTHS:

-
-
-
-
-

WHAT WORKS FOR ME:

-
-
-
-
-

MY FAVORITES:

WHAT DOESN'T WORK FOR ME:

17q12 FOUNDATION

Parent contact info:

_____

_____

17q12

Awareness Day Scavenger Hunt

How many can you find?

- [ ] a butterfly
- [ ] the number 1
- [ ] something that starts with a q
- [ ] 2 clouds
- [ ] something that flies
- [ ] something green
- [ ] something orange
- [ ] the number 7
- [ ] a friend
- [ ] your favorite book
- [ ] a 17q12 shirt
- [ ] a bird
- [ ] a plane in the sky
- [ ] a yellow flower
- [ ] a pair of blue "genes"
- [ ] something that makes you smile