JULY 2024

17q12 Foundation

NEWSLETTER

A RESOURCE FOR CHROMOSOME 17Q12 DELETION AND DUPLICATION SYNDROMES

JULY 12, 2024

WHAT'S NEW?

LETTER FROM THE BOARD

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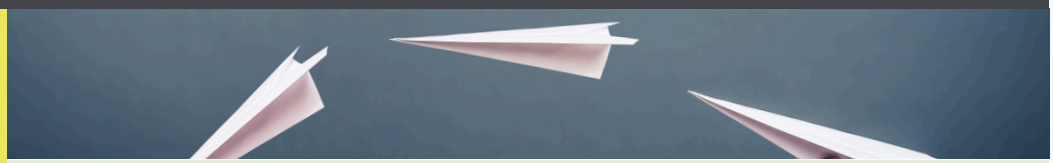


To our 17q12 community,

We are delighted to mark our fifth annual awareness day with all of you. Back in 2017, we began as a small group with ambitious goals that seemed distant. Today, we take pride in the progress made as a dedicated community of individuals, parents, caregivers, and family members united by the common aim of providing support where needed and advocating for increased research. After extensive fundraising and nearly two years of hard work, we have successfully launched the much-anticipated registry. While our primary focus was on creating the registry, we also continued to strengthen ties within the rare community. Representing the 17q12 community at NORD's annual Rare Summit, fostering connections in 19 different countries, and linking more families through our 17q12 Near You program have been key highlights. We look forward to organizing more virtual meet-ups soon, while also striving towards our next big goal: arranging a long-awaited in-person conference. Your support for the 17q12 Foundation's mission of supporting individuals with chromosome 17q12 syndromes is greatly appreciated.

17q12 foundation

17q12
FOUNDATION

The 17q12 Foundation in partnership with the National Organization for Rare Disorders (NORD) has launched the 17q12 Patient Registry, a study with global reach to study chromosome 17q12 deletion syndrome and chromosome 17q12 duplication syndrome. Both syndromes affect various organs and systems, have different features, and may vary from person to person. Some features between the two syndromes may include MODY5 (diabetes), kidney disease, heart issues, and seizures to name a few.

The 17q12 Patient Registry creates a platform for patients around the world to strengthen their voices and provide critical information about their experiences living with chromosome 17q12 deletion or duplication syndrome. Its purpose is to build an international resource to be used by scientists in future research.

"The patient information that the registry collects will help give direction to scientists on areas in which to focus their research efforts, and to hopefully allow clinicians to see trends in patient responses to treatment for chromosome 17q12 deletion or duplication syndrome," said Liz Fourie, Vice President of the 17q12 Foundation. "The hope for the future is to better understand these syndromes and illuminate treatments that have had some success that may become the standard of care for chromosome 17q12 deletion and duplication syndromes."

For more information, visit 17q12registry.iamrare.org.

About the 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 whom 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®)
With a 40-year history of advancing care, treatments and policy, the National Organization for Rare Disorders (NORD) is the leading and longest-standing patient advocacy group for the 30 million Americans living with a rare disease. An independent 501(c)(3) nonprofit, NORD is dedicated to individuals with rare diseases and the organizations that serve them. NORD, along with its more than 330 patient organization members, is committed to improving the health and well-being of people with rare diseases by driving advances in care, research and policy. For more information, visit rarediseases.org.

Media Contacts:
Liz Fourie, info@chromo17q12.org, 17q12 Foundation
NORD Marketing and Communications, media@rarediseases.org

17Q12 PATIENT REGISTRY FAQS

How is the data collected?

Data is collected through a secure web-based application (that can be accessed by computer, tablet or phone) developed by the National Organization for Rare Disorders, Inc. (NORD®), Study participants respond to questions grouped within a series of surveys developed per study standards and in collaboration with syndrome specific experts.

What types of data will be collected in the 17q12 Patient Registry?

The data collected includes but is not limited to:

- Socio-demographics
- Medical and diagnostics
- Treatment and syndrome progression
- Management of care
- Quality of life

Who can join the study?

This study is open to anyone who has a chromosome 17q12 deletion or duplication syndrome diagnosis and meets the study inclusion criteria for participation.

Is there a cost to participate?

There is no cost to the patient to join this study.

Can data be collected worldwide?

The registry uses an online platform which allows participants to contribute data from anywhere in the world. Individuals from other countries who enter data into the registry should be aware that data and privacy laws are different in the U.S. from other countries. This U.S. based registry will protect data and privacy according to U.S. requirements.

Is the data safe?

The registry follows strict government guidelines to ensure patient information is protected. The platform is served over HTTPS, which means that the data is encrypted when being sent from the user's browser to the NORD servers. The data is also kept encrypted in the NORD database. Communications between the registry platform application server and the database are also encrypted. As with any information you provide electronically, there is a very rare chance that your privacy could be compromised. However, the registry and the security measures minimize the chance of this occurring.

Learn more at 17q12registry.iamrare.org/faq/

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

Both Syndromes

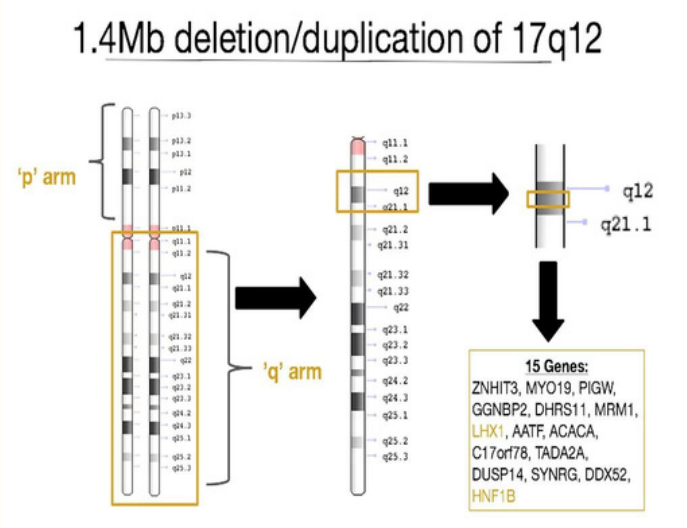
- Kidney abnormalities
- Autism Spectrum Disorder
- Developmental delay
- Eye/vision problems
- Heart defects
- Hypotonia

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
- Hypomagnesemia
- Elevated liver enzymes
- Musculoskeletal
- Most often de-novo (new occurance)

What is 17q12?

The 17q12 Foundation represents two separate chromosome disorders: chromosome 17q12 duplication syndrome and chromosome 17q12 deletion syndrome. Both syndromes have their own symptoms and features and vary person to person.



Celebrating Achievements

Kyle, Age 14
UK (DUP)

Kyle was finally diagnosed with ASD in January 2023. He suffered with his mental health for 18 months. It is heartbreaking watching your child suffer and there was nothing we could do but be there for him, guide him, and help him with medication to ease his torment. He has now come out of the other side our bright beautiful boy. Every year he achieves something new, and his confidence is growing. He's so compassionate and caring and has us laughing with his bluntness. Everyone who meets Kyle can't help but fall in love with him.

Robert, Age 3
USA (DEL)

He is working hard in OT and playing more with toys. He likes to close the lids on containers.

Sebastian, Age 11
USA (DEL)

Sebastian graduated from Elementary School and is moving on to Middle School in the fall. He recently completed his first season playing baseball in the Miracle League of CT, where he is able to play using his wheelchair and hit some great home runs! Sebastian also had his acting debut starring in a commercial!

Delilah, Age 8
USA (DEL)

Delilah has become an amazing artist at just 8 years old and has been doing different events since May. Here she is at one of her events. Her business is called Pretty Lit Creations. She even has a website prettylitcreations.com

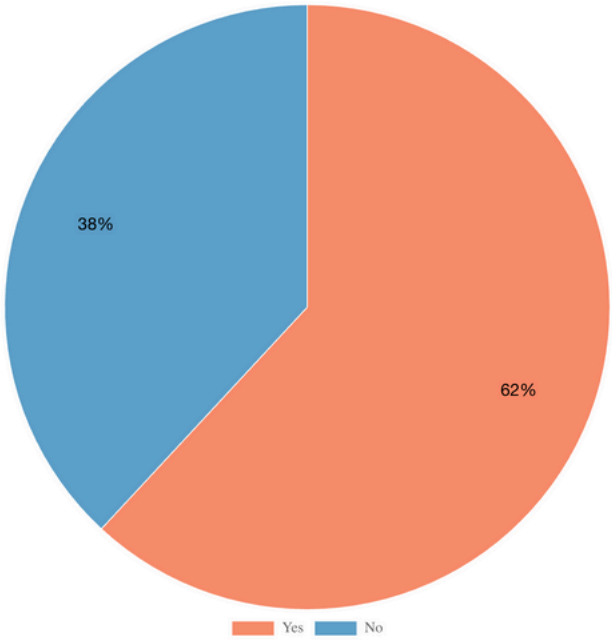
Alina, Age 5
USA (DUP)

This summer Alina learned how to write her first name and was discharged from OT for the first time in her whole life!

THE 17Q12 PATIENT REGISTRY

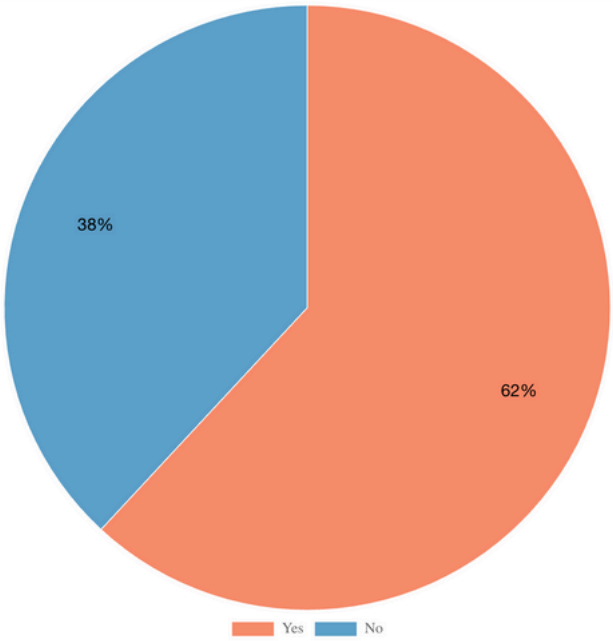
is run by the 17q12 Foundation but it is for the 17q12 community.

Has the Participant ever received occupational therapy?



Responses: 21

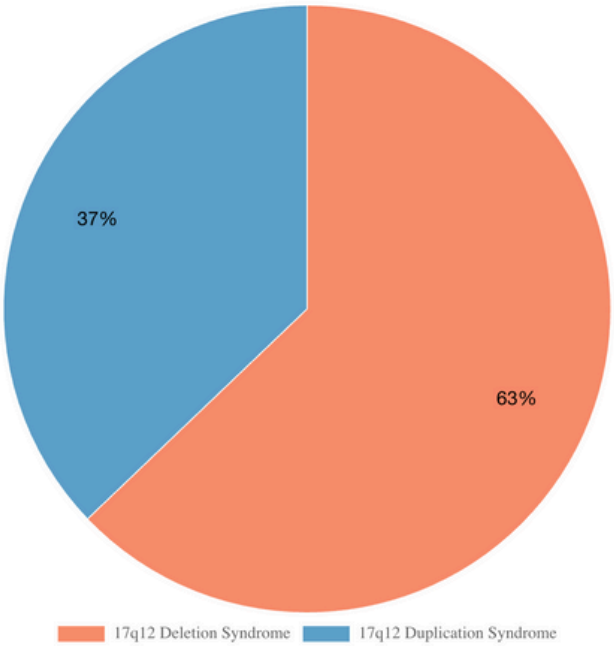
Has the Participant ever received physical therapy?



Responses: 21

Combined 17q12 Duplication and Deletion data pulled from the 17q12 Patient Registry from July 2024. Graph data is based on the number of responses.

What is the Participant's rare disease diagnosis?



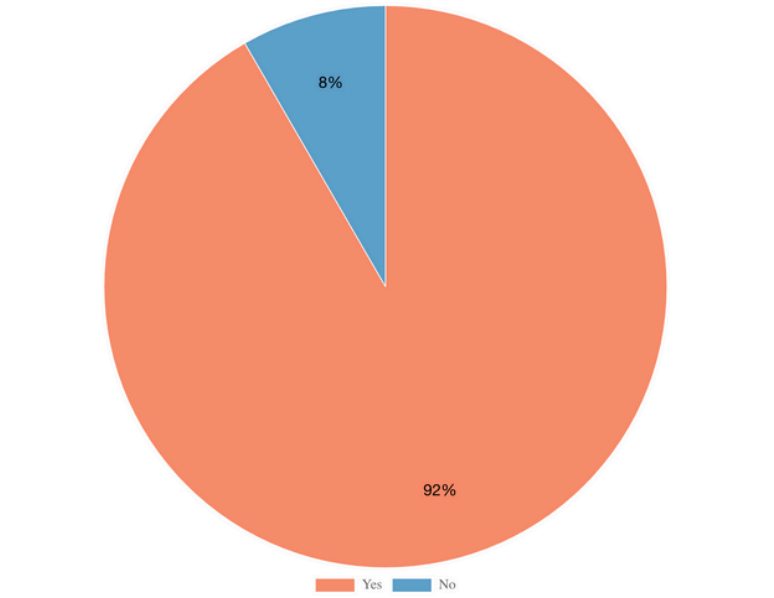
Responses: 35

chromo17q12.org/registry

Chromosome 17q12 Deletion Syndrome

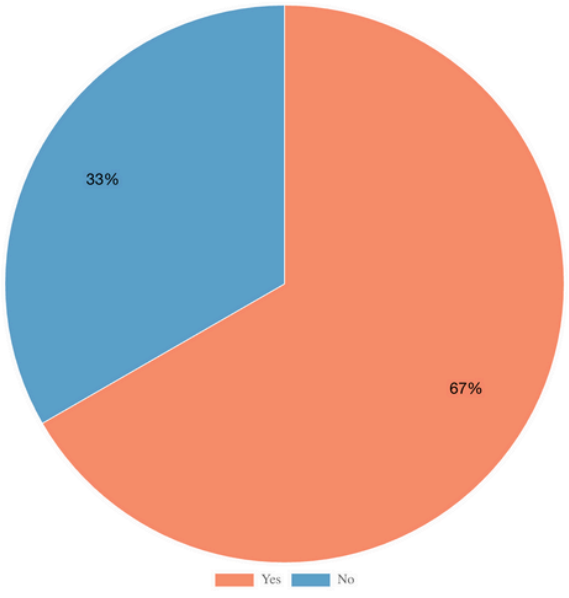
chromo17q12.org/registry

Has the Participant ever been diagnosed with any kidney or urinary tract abnormalities?



Responses: 12

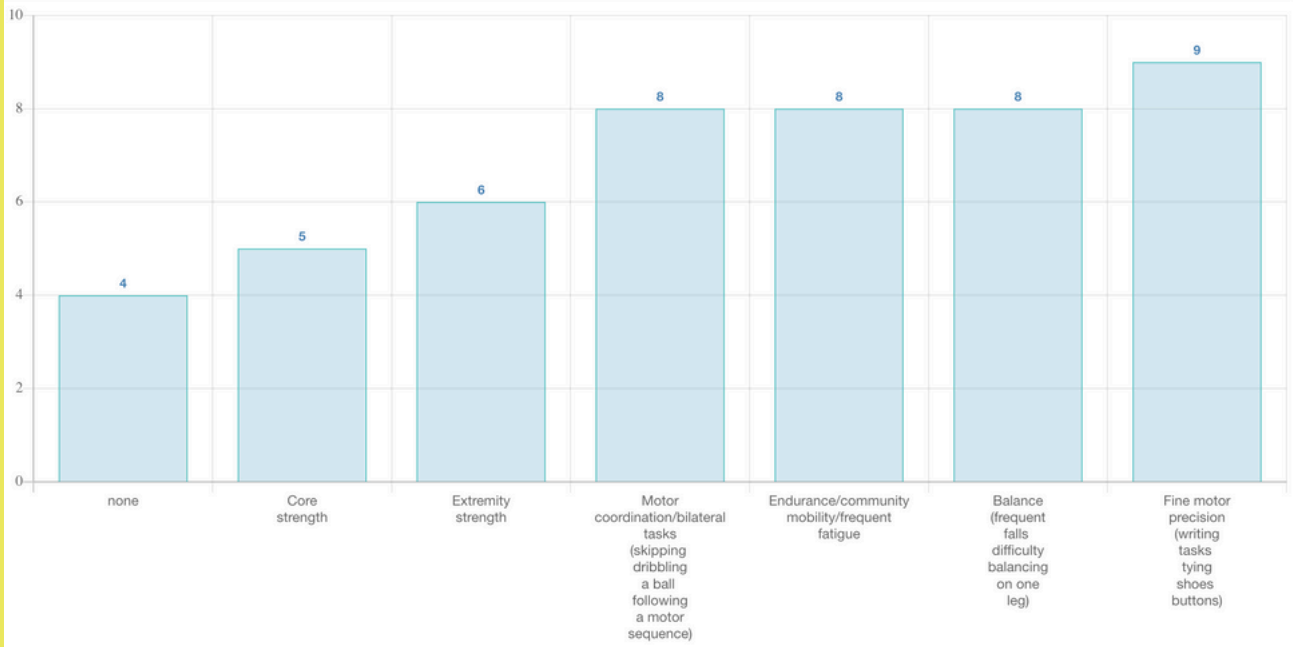
Has the Participant ever been diagnosed with hypomagnesemia (low serum magnesium level)?



Responses: 12

17q12 Deletion data pulled from the 17q12 Patient Registry from July 2024. Graph data is based on the number of responses.

Does the Participant experience difficulties in any of the areas listed below?



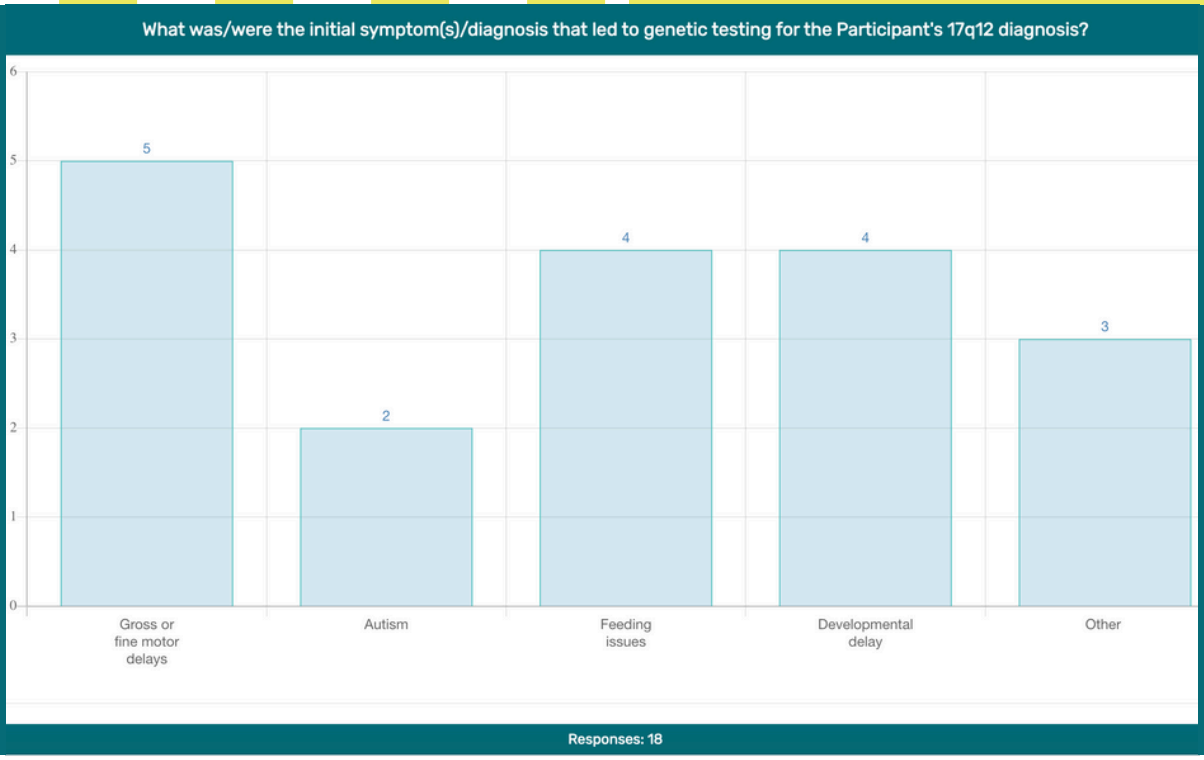
Responses: 48

Chromosome

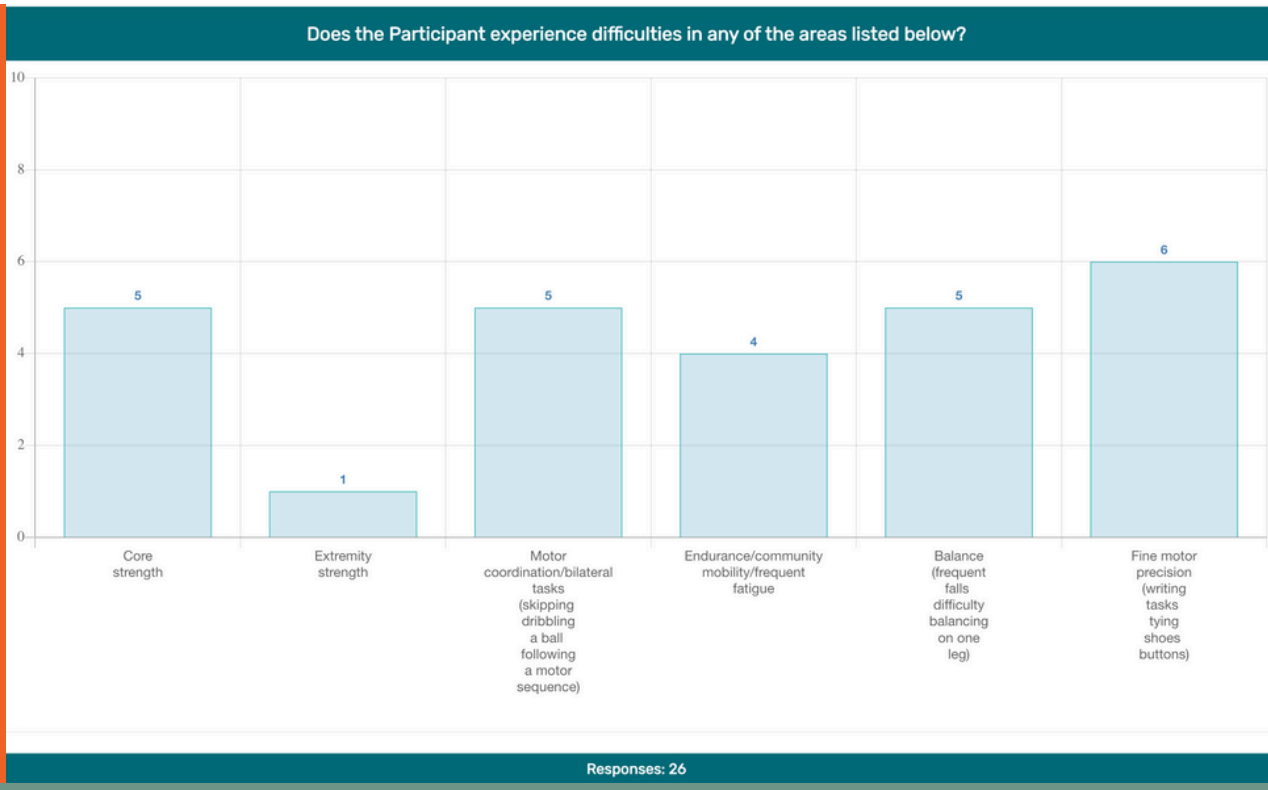
17q12 Duplication

Syndrome

chromo17q12.org/registry



17q12 Duplication data pulled from the 17q12 Patient Registry from July 2024. Graph data is based on the number of responses.



Community spotlight

Wesson, Age 2, USA (DUP)

Wesson was diagnosed with 17q12 duplication at 9 months of age. He has hypotonia, global developmental delay, autism, hypoglycemia, sleep apnea, restless leg syndrome, feeding difficulties, orphangeal dysphasia, swallowing dysfunction, GERD, and anemia. He was fed by ng tube then Gtube, and now he can eat orally with thickened liquids. Wesson also has speech and language disorder, ear tubes, chronic infections, Marcus Gunn jaw winking syndrome, abnormal MRI, eczema, abnormal gait, other disorders of physical development, lack of coordination, diarrhea, constipation, disruptive behavior, and genetic disorder. He also has 16p13.11 microduplication. Wesson continues to see doctors and has tests to follow; it's a journey. He is a determined smart, fun, loving little boy full of life despite his struggles!

Eden, Age 21 Months, UK (DEL)

Eden has 17q12 deletion which has caused her to have cysts on both her kidneys which were detected antenatally. This means Eden has regular visits, which include blood tests and ultrasounds at our local children's hospital. Eden's development is delayed due to the deletion and she is awaiting a diagnosis of global development delay and autism. Eden received the diagnosis just after her 1st birthday after a long 6-month wait for the results. Despite struggling with communication and sensory issues, along with her health issues, Eden is full of determination and will find a way to do whatever it is she desires! Eden has recently learnt how to say 'mama, dadda and babba' and absolutely loves nursery rhymes and bubbles!!

Cole, Age 3, USA (DEL)

Cole was diagnosed at 2 years old with 17q12 deletion when we saw a developmental pediatrician who referred us to a genetic counselor after I thought he may have had some autism markers. He isn't affected as of yet, outside of some gross motor delays and very mild developmental delay. We have seen a lot of specialists and although things are good now, we just need to continue to monitor him throughout his life. Cole's biggest strength is the glow he brings to our mornings. He is always so sweet and happy to see his family when he wakes up, making sure we know he loves them and misses them when they're away. His little smile is just the cutest thing ever and he is such a lovable and cuddly boy. He is truly one of a kind!

Research Anouncements

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

17q12 Immunological Study

As an Allergy and Clinical Immunology fellow at Yale, I had the privilege of providing care for a five-year-old patient with 17q12 deletion syndrome (17q12DS) since 2019. He presented with the well-known symptoms of the 17q12DS including renal cysts and autism. 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.

Subsequently, we recruited 37 patients with 17q12DS and collected their medical histories including allergies, infections, and autoimmunity. As expected, the majority of patient presented with well-known clinical phenotypes of 17q12 DS including renal diseases and neuropsychiatric disorders. Of interest, around 60% of patients presented with recurrent respiratory infections. Therefore, we investigated the alteration of T-cell characteristics and functions in 17q12 DS using bulk RNA sequencing, flow cytometric analysis, and multiplex cytokine array. Here we found that individuals with 17q12DS had a substantially decreased frequency of CD4+ T cells producing the T helper (Th) 1 cytokine IFN-γ, a major cytokine involved in defense against viral and bacterial infection, compared to age-matched healthy controls (HCs). RNA seq analysis of CD4+ T cells revealed decreased levels of TBX21, encoding the Th1 transcription factor T-bet, IFNG, and other Th1 chemokine encoding genes in subjects with 17q12DS compared to HCs. These findings were validated at the protein level using flow cytometry and multiplex assay. Our study is the first to demonstrate immune alterations in 17q12DS characterized by decreased T-bet and its downstream molecules such as IFN-γ. We are in the process of publishing this work. These findings warrant further investigations into the underlying mechanisms, which would inform precision therapy for individuals with 17q12DS.

Research Anouncements

Dr. Pengfei Liu's Study

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.

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

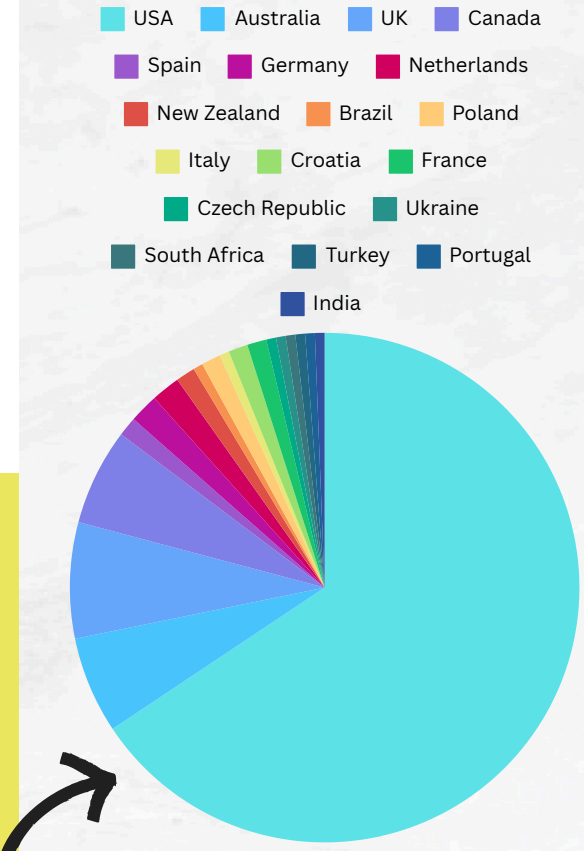
PRISMA Study

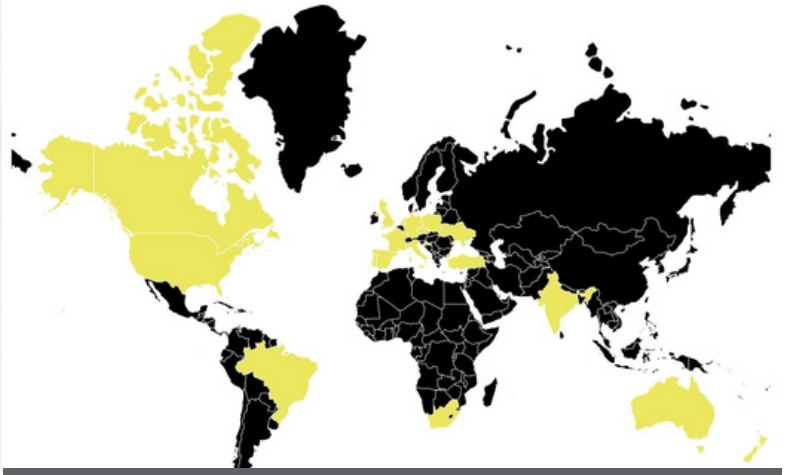
Daniel Moreno De Luca MD MSc
CASA Research Chair
Associate Professor & Principal Investigator
Precision Medicine in Autism (PRISMA) Group
Child and Adolescent, and Adult Psychiatry

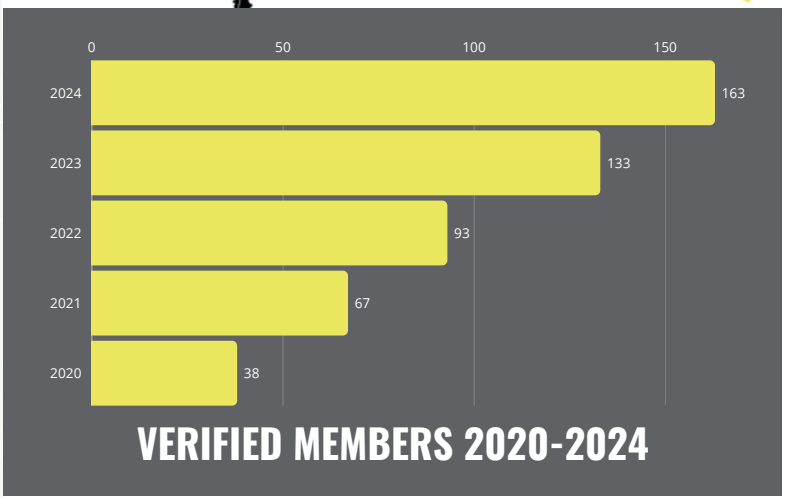
We are delighted to share some exciting updates from our PRISMA group – our team moved to the University of Alberta in Edmonton as part of Dr. Daniel Moreno De Luca's appointment as the inaugural CASA Research Chair! This move translates into exciting opportunities to double down on our efforts to work with the 17q12 CNV community as we aim to understand how this genetic change may lead to neuropsychiatric and medical conditions, and design precision medicine interventions informed by genetics for people who need them. We welcome the participation of anyone with 17q12 CNVs, with or without any psychiatric diagnoses. If you have already joined our previous US-based study, please reach out to us as we'd love for you to continue participating in this effort, which is considered a new study here in Canada (which means we would need a new consent from you). Moreover, we are delighted to share that this study can now be carried entirely in Spanish, aligned with our goal of obtaining the broadest and most diverse perspectives from our participants. Please check out our study flyer, and contact us at prisma@ualberta.com and visit www.precisionmedicineinautism.org to learn more.

JOIN US!

CHROMO17Q12.ORG/JOIN-US







WHO ARE WE?

We currently have 163 verified members/families from the following countries:

71 verified members have 17q12 duplication. 92 verified members have 17q12 duplication.

343 Membership Registration forms have been submitted.

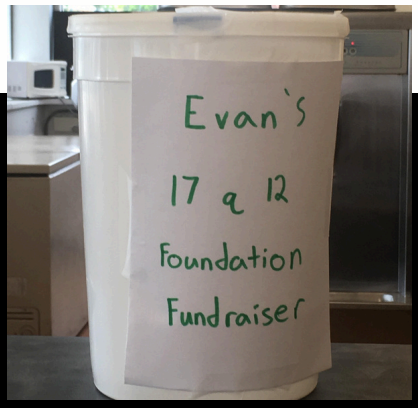
There are 282 verified members in our private support group on Facebook for parents, caregivers, and individuals directly affected by a 17q12 syndrome.

BECOME A 17Q12 FOUNDATION MEMBER

As of July 2024, the 17q12 Foundation has confirmed members spanning across 19 countries worldwide! By becoming a member of the 17q12 Foundation, individuals join the 17q12 community to collaborate effectively. This membership grants access to important updates, ranging from research opportunities to conferences and virtual meetups. Verified members can also participate in our private Facebook support group for those directly impacted by chromosome 17q12 syndromes. Members have the option to register with 17q12 Near You, an initiative that aims to establish a supportive community by linking families and individuals in their respective regions. If you or your child has been diagnosed with chromosome 17q12 deletion or duplication syndrome, we invite you to connect with us at chromo17q12.org/join-us.

FUNDRAISE FOR 17Q12

Thank you to the siblings and individuals with 17q12 who created this year's shirt design!



Our aim for fundraising this year is to finance a crucial family conference in 2025. By joining forces, we can gather enough funds to achieve this target.

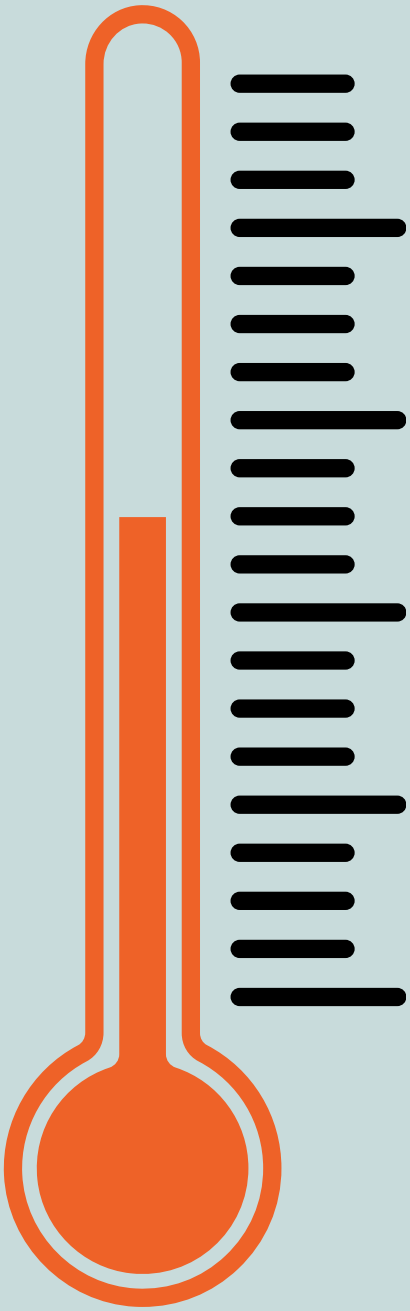
Do you have an idea for a fundraiser? No fundraiser is too small. The 17q12 Foundation is a registered 501(c)(3) organization and relies heavily on fundraising and community support. Any fundraising big or small makes an impact.

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!

A YEAR IN REVIEW



A LOOK INTO THE FUNDS WE RAISED AND SPENT IN 2023

>>> HOW WE RAISED

We recieved $4646 in donations through:

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

>>> HOW WE SPENT

We spent $2,963 on administration costs that included:

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

Gold Transparency 2024

Candid.

Emergency Info

NAME: .....

Age: _____

Date of birth: _____

HERES A PICTURE OF ME!

Emergency contact(s):

Name: _____

Relation: _____

Phone #: _____

Name: _____

Relation: _____

Phone #: _____

Parents/Caregivers:

Name: _____

Phone#: _____

Work place: _____

Name: _____

Phone#: _____

Work place: _____

Diagnosis(es):

_____

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Medication(s):

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Doctor(s):

_____ Phone:

_____ Phone:

_____ Phone:

_____ Phone:

_____ Phone:

Allergies:

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Things to Know, Communication Devices, Noise canceling headphones etc:

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QR CODE FOR MORE INFORMATION ABOUT 17Q12 :

SCAN ME