17q12 FOUNDATION

NEWSLETTER

q12

Learn more about chromosome 17q12 deletions and duplications

CLINICAL RESOURCE GUIDE

Doctors and clinicians who can talk about 17q12

FROM THE EDITOR

WELCOME!

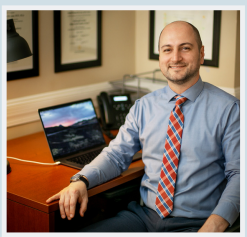
It is our pleasure to share with you to the first edition of the newsletter on this 17q12 CNV (copy number variant) awareness day!

We hope that the newsletter brings useful information in a fun and engaging way. This was only made possible by working closely together, so a special thanks to all of you! In particular, thanks to all the families and "qt's" who shared their stories and efforts, to the 17q12 board members for their support, to David Ledbetter, Christa Martin, Cora Taylor, and the ADMI (Autism & Developmental Medicine Institute) team, to Molly Goldman and the PRISMA (Precision Medicine in Autism) team, and to Ms. Silvana Guerrero for being the heart and mind behind putting together this wonderful newsletter.

In this edition, we'll get a chance to review some of the facts around 17q12 CNVs (including deletions and duplications, to learn about resources for the 17q12 community, and to read about the achievements of the 17q12 Foundation members. In addition, we'll explore some of the current research around 17q12 and learn all the details about the upcoming International 17q12 Family Meeting.

Although this newsletter does not provide individual medical advice, please feel free to take it with you to share this information and resources with your healthcare team or community as a way of letting them know more about 17q12 CNVs.

It has been very rewarding to see the 17q12 Foundation grow and expand to continue bringing awareness and resources to 17q12 families, and we are honored to be working with you in research, clinical care, and advocacy efforts. We hope that you enjoy the newsletter and that you continue sharing your stories, questions, and resources for the benefit of the 17q12 CNV community.

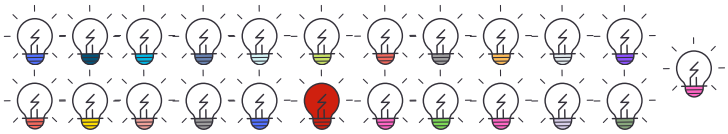


Daniel Moreno De Luca, MD MSc
Scientific and Clinical Advisor 17q12 Foundation

What are 17q12 CNVs?

The 17q12 Foundation represents two separate syndromes:

17q12 deletion syndrome



is caused by a missing piece of chromosome 17 (deletion) that is present from the moment the child is conceived

17q12 duplication syndrome

is caused by an extra piece of chromosome 17 (microduplication) that is present from the moment the child is conceived

1 in 14500 people

in the general population have this deletion syndrome. It's more common in populations with developmental disorders developmental delay, intellectual disability autism spectrum disorder) and schizophrenia

The deletion is most often a brand new (de novo), sporadic event in the person that is diagnosed

of the time a person with the deletion will have inherited it from a parent

chance that each of that parent's children will also have the deletion

chances that a sibling has if it was de novo (both parents tested negative)

1 in 2500 individuals

in the general population have this duplication syndrome. It's more common in populations with developmental disorders (developmental delay, intellectual disability autism spectrum disorder) and schizophrenia

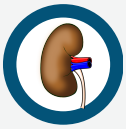
The duplication is most often inherited from a parent. Oftentimes, the duplication is only identified in parents after the child is diagnosed and could have similar, milder or apparently no features

chances that each of that parent's children will also have the duplication

of people with the duplication will have a brand new (de novo) duplication that wasn't inherited from either parent

chances that a sibling has the duplication if it was de novo (both parents tested negative)

A syndrome is defined as a recognizable group of signs and symptoms that consistently occur together.



Kidney and urinary tract abnormalities

Macrocephaly (Large head size)

Maturity onset diabetes of the young type 5 (MODY-5)

Neurodevelopmental/Psychiatric: developmental delay, autism spectrum disorder, learning disabi-lity, intellectual disability, anxiety, bipolar disorder, schizophrenia

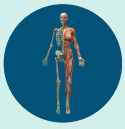
It's important to remember that no two people with the deletion will have the same combination and/or severity of symptoms, even people within the same family

The most common features of the duplication are related to neurodevelopment.

Skeletal differences

Microcephaly (small head size)

Seizures in up to 75% of individuals



Hypotonia (low muscle tone)

Neurodevelopmental/Psychiatric: intellectual abilities ranging from typical to severe disability, speech delay, motor delay, behavioral concerns (aggression, compulsive disorders), autism spectrum disorder

It is important to remember that no two people with the duplication will have the same combination and/or severity of symptoms, even people within the same family

For More Information
visit www.chromo17q12.org/

INFOGRAM MADE BY PRISMA RESEARCH

How are 17q12 CNVs diagnosed?

BY: MOLLY GOLDMAN & DANIEL MORENO DE LUCA

Now that we know more about 17q12 CNVs, how do people get a diagnosis?

Families arrive at a diagnosis in one of many ways: they may go to see their doctors because of developmental or psychiatric conditions, or because of issues with their kidneys or abnormal blood sugar levels. Doctors can then perform several tests, including genetic testing, where the 17q12 CNVs are revealed.

The main genetic test ordered by doctors in this setting is Chromosomal Microarray. This is a technique looks for small deletions and duplications throughout the genome, and can detect CNVs. In some locations, such as 17q12, these CNVs can be recurrent, meaning that they involve the same unique segment of DNA in unrelated people. The genetic region involved in 17q12 CNVs is about 1.4 Mb in size and includes 15 genes, among them *LHX1* and *HNF1B*. *LHX1* is expressed in the brain early in fetal development and *HNF1B* is important for the developing kidney, liver and pancreas. Children and adults with 17q12 CNVs may share some common features, while some other characteristics could be specific to people with the deletion, or those with the duplication. These features do not always affect everyone with 17q12 CNVs in the same way, and in any given person, they can be more or less obvious. Much remains to be discovered.

In some cases, people with 17q12 CNVs are the first in their families to have this genetic change. When these CNVs are not inherited from either of the biological parents, they are labeled as de novo, meaning that they happened spontaneously. Less often, 17q12 CNVs are inherited from a parent.

When this happens, the parents appear to be affected most of the time when they have the deletion, and sometimes when they have the duplication, although the degree can be very variable; in some cases these parents are just mildly affected and would go undiagnosed until they are tested to follow up the genetic findings in their children.

Whether the deletion is inherited or *de novo*, as a parent there is nothing you did to cause it and nothing you could have done would have prevented it from occurring in your child. No environmental, dietary, workplace or lifestyle factors are known to cause these changes in 17q12.

Liam's Story

WITH 17q12 DUPLICATION

Our son, Liam, is almost 4 and has the duplication. Liam was a totally normal pregnancy and the first few months were right on par. When Liam was about 6 months I started noticing he didn't "feel" right, I brought it up to his pediatrician, and they concluded I was clearly crazy and he was "fine".

However, I persisted and then Liam started missing milestones. Finally, the doctors took notice and sent us to a neurologist. Liam was diagnosed with severe hypotonia (low muscle tone). Then came trying to find out why, a blood test and a wait told us he had the 17q12 duplication.

Unfortunately having a diagnosis only meant more questions with no answers.

We scrounged online and through medical papers and still didn't really learn anything new. However, we did stumble on a conference and decided to attend. It was so wonderful meeting the other families. Liam' s hypotonia has been managed with physical therapy. His other symptoms are speech delay, mild cognitive delay, global development delay, obsessive compulsive disorder, anxiety, iron deficiency, and sensory seeking behaviors.

He sees his speech therapist every week, and an occupational therapist once a month. Liam started school last year and we saw so much improvement with his speech.

"Liam is the happiest kiddo and lights up our life. We are thankful we came across the conference and were really able to get involved"

17q12 CNV FACTS

Learn Something New Every Day

1.

Did you know that 17q12 is pronounced seventeen q one-two?

2.

Extra (duplicated, symbolized in blue) or missing (deleted, symbolized in red) chromosomal segments are called copy number variants (CNVs)

3.

de novo is a Latin expression used in English to mean "anew"

4.

2019 is the first year we are celebrating 17q12 awareness day, which is happening on July 12th

5.

17q12 Foundation is part of the Global Genes RARE Foundation Alliance, which is made up of over 500 disorder foundations. More information visit www.globalgenes.org

6.

Precision medicine is the ability to use precise individualized information, like genetics, for tailored clinical management

Greetings from the 17q12 Foundation

"Creating a community for those affected by 17q12 syndromes while raising awareness and encouraging research"

BY: 17q12 FOUNDATION

The 17q12 Foundation was born from the group that was initially organized through Geisinger Health System for their research study. At the last family conference in Chicago in 2017, a group of parents and a genetic counsellor decided it was time for us to embark on the journey toward becoming a free-standing non-profit organization dedicated wholly to increasing awareness and driving research forward for both the deletions and duplications.

Since the summer of 2017, we have achieved 501(c)(3) status and have been working diligently to create our brand, grow our network of families around the world, and connect with researchers. We all know there is a lot to learn about both of these conditions, and it starts with us making our needs known to the scientific community. We felt that previous conferences have been instrumental in bringing families together and inspiring each other to keep moving forward in this (sometimes daunting) journey of discovery and advocacy. For this reason, we prioritized our first major project as a foundation to be hosting another family conference. We have more families registered for this year's conference than ever before! Our next big project is absolutely critical to answer all the questions that remain unanswered for you and your families: a medical registry.

A registry is basically a big, secure database that will hold all of the diagnoses, features, symptoms, etc. that you/your children have accumulated in one place. If we have all the data in one place, we can more easily convince researchers to design studies that will ultimately help medical professionals take care of you/your children more effectively. As you can imagine, it is not cheap to create and maintain a database that can securely store all of this information. Which means... we need to fundraise, fundraise, fundraise! As the old adage goes, it takes a village. As our village continues to grow we hope that everyone will feel a sense of community and continue to work together and encourage each other to achieve our common goal: setting our families up to reach their highest potential.

MEET THE BOARD

Allaina Wellman -President

My son, Liam (4), has the 17q12 duplication. We discovered it right before Liam turned 2. We knew fairly early something wasn't right with Liam in regard to his low muscle tone and his falling behind in reaching his milestones. Right after Liam's diagnosis we found the Geisinger Family Meeting that was in Chicago and decided to give that a try to find more information and meet others. Unfortunately, there wasn't a lot of information to discover, but we did meet some amazing families and together we came up with the 17q12 Foundation. I understand that this can be scary and lonely, but you aren't alone. The board and myself are working hard to make sure there are answers for all of us, maybe not tomorrow or the day after, but one day we'll have our answers.

Stefanie Turner, MS, CGC - Vice President

I'm a genetic counsellor by trade. I became closely involved with the 17q12 family while working for Geisinger as a study coordinator for their 17q12 project. I have since moved back home to Detroit, MI but a part of my job was to moderate the original Facebook support group and that's when I realized how much we still have to learn about these conditions. I would see posts about problems kids were having that I couldn't find anywhere in the medical literature. I'd get asked questions that I couldn't find the answers to. But most of all, I fell in love with the people, the stories, the challenges and the triumphs. None of us were trained to run a non-profit, but we are figuring it out as we go because we are all tremendously invested in the end goal.

Sherie Scott- Secretary

Both my son, Alex age 10, and myself have the 17q12 duplication. I found out that Alex had it when he was 3 while trying to figure out a cause for his migraines. I found out about myself almost four years ago. We both have low muscle tone, some digestive issues and some learning difficulties with reading and comprehension. I want to tell everyone that is worried like I was at first upon learning about Alex's diagnosis: don't stop dreaming for your child's future. I am here to prove that. I have a great career and am able to raise my two children. I know I was worried that Alex would not make it to adulthood, but that is no longer a fear of mine and I hope to help others work through that fear.

Liz Fourie - Communications Director

I grew up in Illinois but I call California home with my husband, Brian, and our two kids, Zander age 8, and Ruby age 6. During the week I homeschool my kids and do photography on the side. Our road to finding out our son, Zander, has 17q12 deletion syndrome was not an easy one, like so many other families with this diagnosis. Once we finally received a diagnosis we were still lacking answers regarding Zander's symptoms, information, resources, and a sense of community. This has motivated me to work alongside this awesome group of people in getting this organization off the ground. I run the website and social media for 17q12 Foundation, and I hope that future families find some sort of comfort and support when they find that there is a growing 17q12 community.

Mark Dempsey- Treasurer

Our family joined the 17q12 family when our middle child, Mason, was diagnosed with the deletion during his first year of life. He is now 4 years old and, despite the challenges, Mason loves all things sports! We have been actively involved in our local chapter of the National Kidney Foundation and are excited to take the next steps with the 17q12 Foundation!

RHODE ISLAND

*Venue for the International 17q12 Family Meeting-**August 10-11, 2019***

This year, the 17q12 Family conference is being held in beautiful Rhode Island, RI. Conveniently located between the west coast of the US and Europe, and well connected by air and land, RI is a fun summer destination with many things to do!

With just over a million residents living in 1,200 square miles, RI can be crossed in about an hour. The population is diverse, and the state has a single department of health, children's hospital and medical school. In addition, it is the home of Brown, a leading Ivy League research University, and to the Rhode Island School of Design, one of the best institutions in the world for art and design, as well as many other excellent colleges and universities.

Some of the best beaches in New England are located here; close to 400 miles of coastline and excellent surf and sand make it clear why RI has been adequately named the Ocean State. All of this makes RI an eclectic, unique place to visit.

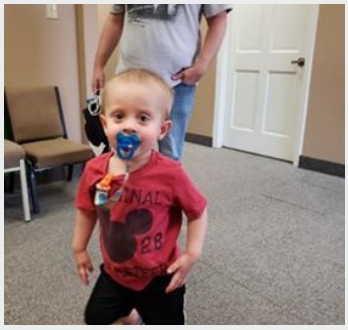
In the second day of the conference, we will be visiting ***Roger Williams Park Zoo*****,** one of the oldest in the country, where we can see animals from all over the globe. Naturalistic surroundings are home to more than 160 animals including a Komodo dragon, as well as zebras, red pandas, African elephants, Masai giraffes, snow leopards, bears, anteaters, flamingoes, sloths, alligators, and more!

We are very grateful for their sponsorship of this part of the meeting, where families will get a chance to bond and have fun, and to Brown University, Geinsinger, and the Simons Foundation for their support!

Finally, RI is also home to many iconic and staple foods and dishes. "Little Rhody" is known for its coffee milk, wich is so iconic here that it became the official state drink in 1993. There's also Del's frozen lemonade, which traces its origins back to 1840, as well Doughboys, the Cabinet, the famous Hot Wieners and Rhode Island Clam Chowder!

We hope to see all of you soon at the International 17q12 Family Meeting so that you can enjoy visiting this fun-sized state.

THIS IS HOW WE FLY...



Aksel (2y)
Calhoun, GA

Started walking a few months ago, is learning sign language and working on tag 300 calories by mouth a day.

Daisy (1.5y)
MN

Can climb up the stairs. Now we work on climbing down!

Holly (36y)
Boaz, AL

My wife is a wonderful mom of 2 boys. Our 7 year old also has the same duplication that Holly has. Holly does so much for our community. She is a substitute teacher, local program coordinator lo for Blessings in a Back-pack nonprofit, manager of the local Pirate Pantry and also the absolute best Dugout Mom. I met her 19 years ago and just last week we renewed our vows in Jamaica on our 15th wedding anniversary.

Max (2.5y)
Bethpage, NY

Starting to walk and is learning sign language with his big sister!

Victoria (7y)
NJ

She's been learning better coordination to tie her shoes. We are almost there!

Dawson (7y)
Boaz, AL

Dawson is the definition of determination, he does not let his speech stop him from making friends, ordering his own food, and reading a book to his entire class.

Peyton (4.5y)
Bethpage, NY

Learning sign language

Stephen (36y)
Bethpage, NY

He's a fantastic husband and awesome daddy to Peyton and Max and he has the 17q12 duplication like our kids. He is learning new skills everyday by watching our kids do sign language.

WORD SEARCH PUZZLE

C D R C I F R G I C R S N D M M D
E S E R L R F E D H F E E O I I I
T X E N O T G N D R G V U C C S A
E Y T I O Y H E T O H E R E R S B
A D M R Z V J S O M J N O H O I E
S I I R A U O R P O A T D J C N T
Y E C F R P R F Y S I E E I E G E
N C R D T M I E H O N E V N P N S
D I O H E M W E S M O N E H H P F
R S D O G L S I C E T q L E A I I
O I E W F F E F F E O O O R L E V
M E L W D G A T D E P N M I Y C E
E T E E C T R T I W Y E E T G E T
D E T F I E T E S O H T N E H Y Y
E R I L I W F L Y L N W T D J H V
N E O Y V K I D N E Y O A K N O J
O G N J F E A T U R E S L S R L L
V D U P L I C A T I O N L W E L E

- *de novo*
- CHROMOSOME
- DELETION
- INHERITED
- KIDNEY
- MISSING PIECE
- EXTRA PIECE
- FEATURES
- SYNDROME
- DIABETES FIVE
- DUPLICATION
- 17Q12 (WORDS)
- DIECISIETE
- MICROCEPHALY
- HYPOTONIA
- NEURODEVELOMENTAL
- SEIZURES

CERTIFICATE OF ACHIEVEMENT

THIS IS AWARDED TO

TO CELEBRATE HOW WE FLY...

#HOWWEFLY

17q12 RESEARCH

ADMI 17q12 Project

As researchers and health care providers at Geisinger's Autism & Developmental Medicine Institute (ADMI), they're interested in better understanding the behavioral, developmental, and medical characteristics associated with extra or missing material in the 17q12 chromosomal region. Your participation helps increase our knowledge about 17q12 deletions and duplications, with the goal of improving the lives of affected individuals and their families.

Molecular Mechanisms of 17q12 deletion syndrome: Developing a novel mouse model of polygenic ASD

This study from Brown University will use an animal model to determine which genes on this area of chromosome 17 are responsible for autism vs. other physical issues including kidney problems and diabetes. Further knowledge of the function of multiple genes on this region will help develop better targeted therapies for autism and developmental disorders based on genes on chromosome 17.

A genomic approach to Precision Medicine for Autism (PRISMA)

This project at Bradley Hospital, Hasbro Children's Hospital, and Brown University is looking to understand how genetic information, specifically rare genetic changes, of people with autism and other neurodevelopmental and psychiatric conditions can inform healthcare. We want to see how genetic results could be used in the future to inform medication choice, behavioral interventions, educational interventions, and identification and treatment of other medical conditions that may impact mental health. We will be enrolling individuals with autism or other neurodevelopmental or psychiatric conditions for whom a genetic variant has been identified as the underlying cause, such as 17q12 CNVS.

PRISMA is directed by Dr. Daniel Moreno De Luca who is a psychiatrist and researcher specialized in autism and genetics.

GET INVOLVED

FUNDRAISING

Do you have an idea for your own fundraiser to help support 17q12? Please e-mail us at chromosome17q12@gmail.com, and we can help you get started.

CONNECT

Connect online by following our page on Facebook and stay informed about updates from the 17q12 Foundation.

EDUCATE

There is still much to learn about 17q12 syndromes, and it can be hard when it comes to school, doctor appointments, and every-day life. Please help us spread the word about 17q12.

GIVE A DONATION

17q12 Foundation is run solely by volunteers and is funded 100% by donations. It is recognized by the IRS as a 501(c)3 non-profit for tax purposes. Please consider donating by going to our website: www.chromo17q12.org

Let's show the world

#HOWWEFLY FOR 17q12

This year, we celebrates our first-ever Awareness Day on July 12th. For this momentous occasion, we're doing something special and new! We are launching a #HowWeFly t-shirt campaign, PLUS giving you an easy way to make your own t-shirt fundraising campaign so that your friends and family can support your 17q12 warrior. We've created a design template for you to use, or you can create a custom design (for free!). Visit this page (https://try.bonfire.com/17q12) to get started.

CLINICAL RESOURCES

17q12 CNV SYNDROMES

GENETIC COUNSELORS

Stefanie Turner, MS, CGC
Genetic Counselor

Molly Goldman, MS, CGC
Alberta University - Genetic Psychiatry Consultation Service - Tel: (780) 492 4467

Brenda Finucane, MS, LGC
Geisinger Autism & Developmental Medicine Institute - Tel: (570) 522-9432

ADULT AND CHILD PSYCHIATRIST

Daniel Moreno De Luca, MD MSc
Certified to provide clinical care in English, Spanish, French, and Italian
Alberta University - Genetic Psychiatry Consultation Service - Tel: (780) 492 4467

SPEECH-LANGUAGE PATHOLOGIST

Marissa W. Mitchel MS, CCC-SLP
Geisinger Autism & Developmental Medicine Institute - Tel: (570) 522-9420

NEURODEVELOPMENTAL PEDIATRICIANS

Scott M Myers, MD
Geisinger Autism & Developmental Medicine Institute - Tel: (570) 522-9430

Thomas D Challman, MD
Geisinger Autism & Developmental Medicine Institute - Tel: (570) 522-9430

MEDICAL GENETICIST

Heather C. Mefford, MD, PhD
University of Washington - Tel: (206) 543-9572

SPECIALTY HEALTHCARE SERVICES

Autism & Developmental Medicine Institute (ADMI)
Geisinger's - Tel: (800) 275-6401

Genetic Psychiatry Consultation Service
Alberta University - Tel: (780) 492 4467

SUPPORT AND ADVOCACY GROUPS

17q12 Foundation
www.chromo17q12.org

Chromosome Disorder Outreach (CDO)
https://chromodisorder.org/

Unique – Rare Chromosome Disorder Support Group
www.rarechromo.org/

Do you know of other resources? Send us an email at chromosome17q12@gmail.com and we'll add them.

www.chromo17q12.org