
17q12 FOUNDATION

# NEWSLETTER

# Get to Know 17q12 Foundation

## Board Q&A

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.
**Where are you from?** The Colony, Tx originally but currently we are stationed in Fort Polk LA.
**If you could fly anywhere where would you fly to?** Rome, Italy.

Stefanie Turner
**Vice President**

**What does 17q12 Foundation mean to you?** I may not have a 17q-tie of my own, but I fell in love with all the 17q-ties and consider this community one big family. I feel honored to be in a position to hear all your stories and to work with you toward greater awareness and a better understanding of these rare disorders.
**Where are you from?** Livonia, MI
**If you could fly anywhere where would you fly to?** The Netherlands

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.
**Where are you from?** Cleveland area of Ohio
**If you could fly anywhere where would you fly to?** Germany to see where my Grandmother was from.

Liz Fourie
**Communications Director**

**What does 17q12 Foundation mean to you?** It means community and pushing forward for a better understanding of 17q12 disorders.
**Where are you from?** Raised in a small town in Illinois, but settled in Carpinteria, CA
**If you could fly anywhere where would you fly to?** Morrocco

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
**Where are you from?** Grimes, IA
**If you could fly anywhere where would you fly to?** Italy (or Europe in general)


## The 17q12 Family

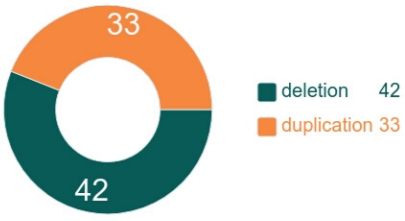
We represent a growing organization of individuals who share the same vision: to improve the quality of life of people with 17q12 deletions and duplications by increasing awareness, advancing research and providing family support. For many families and individuals, it can be daunting after the initial diagnosis of a chromosome 17q12 disorder. We hope to ease that feeling by providing a sense of community through our upcoming family match program, future family conferences, and in our private Facebook support group for parents and individuals directly affected. Since beginning in 2017, we have grown and have families connecting from various states and countries. We have also grown in support from the medical community, and we are grateful for the current and upcoming research opportunities for 17q12 deletions and duplications. We can't wait to see what the future holds for our expanding 17q12 family!

# 17q12 Foundation Membership

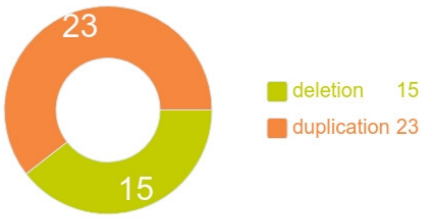
In preparation for our registry and to help our Research Network apply for grant funding, the Foundation would like to encourage all of our families to become a 17q12 Foundation member!

Becoming a member is as simple as filling out a few easy questions about you or your 17q-tie on our website (www.chromo17q12.org/join-us) and emailing our genetic counselor, Stefanie, a copy of your/their chromosomal microarray report. Joining the 17q12 Foundation will allow you to stay up-to-date on the latest information, research opportunities, Foundation events and more! Joining the Foundation also gives you access to the 17q12 Foundation Support Group on Facebook. Providing your mailing address will allow us to start a member match program to connect families that live near each other and may be interested in meeting in person or organizing local 17q events!

**75 Submitted Forms**




**38 completed forms with verified diagnosis**




**175 MEMBERS OF THE GROUP**

Having an organized way to keep track of how many individuals have the deletion and the duplication is an important first step in advocating for more research, and research is critical to a more complete understanding of these conditions. There are multiple deletions and duplications that can occur on chromosome 17q12 that are different from 17q12 deletion or duplication syndrome. This can be very confusing when families are comparing symptoms and features that they see in themselves or their children. We ask to verify the diagnosis to complete your membership and be added to the Facebook group to ensure that everyone in the group has the same condition. Having a copy of your/your child's chromosomal microarray report is important for many reasons: schools may use it to determine eligibility for long-term services, it may be helpful for specialists to familiarize themselves with the medical literature, and it may be requested to determine eligibility for Supplemental Security Income (SSI) when individuals turn 18. A copy of the chromosomal microarray will also be necessary to participate in our medical registry and many other research studies. If you do not have a copy of your/your child's test results you should be able to call the doctor that ordered the testing and request a copy of the chromosomal microarray report.

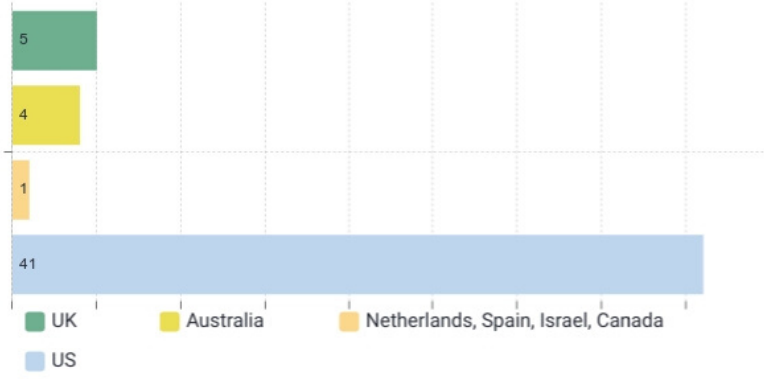
Even if you are already a part of the Facebook group we encourage you to fill out the membership form so we have your contact information and you are included in our total count when researchers ask us how many individuals have the deletion or duplication. You will not need to send a copy of the chromosomal microarray report if we've already verified it to add you to the Facebook group; please just mention that you are already in the group in the comments section of the membership form.

# COMING SOON!

The Foundation has been hard at work creating our Research Network, evaluating medical registry platforms, and working on a Membership Match program!

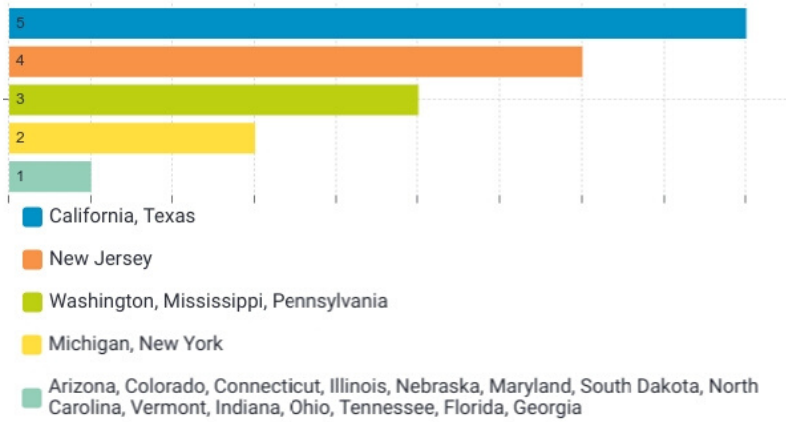
A medical registry is a large, secure database that we can use to store information like genetic data, symptoms and other features from many families all in one place. As we all know, there is A LOT to be learned about both the deletion and the duplication. There are so many features that you/your kids share, but they are not published in the medical literature for doctors to use when making medical management decisions. Being able to collect detailed information on every body system (eyes, heart, stomach, skin, etc.) in an organized way will allow us to publish a natural history study. This type of study is a "big picture" view of a condition from infancy to adulthood, describing all the associated features, how frequently they occur, and at what age they occur. The accuracy of this study depends on how many individuals participate; the more people that participate, the more confidently we can say which features are associated with the deletion/duplication and which features may be a coincidence. A study like this will also highlight the features that are not currently represented in the medical literature, which will make it clear what areas of research we need to advocate for the most.

## 17q12 Foundation Members (Worldwide)




There are many options to choose from when deciding which registry platform will work best to achieve our goals as a community and the 17q12 Foundation Board is working with our Research Network to evaluate our options carefully. We hope to be up and running to start enrolling families in 2021. Stay tuned for updates as we move forward with this exciting project!

## 17q12 Foundation Members (US)




We often get questions about where people are from and if there are other families nearby that could be close enough to meet in person. We understand how useful it can be to have a local support network to discuss specific doctors, school districts, laws, etc. We feel it would also be a great opportunity for families to work together for meet-ups, fundraisers, and other local events. So, we have started collecting geographical information through our membership form in order to offer a Member Match program. This program would allow interested families to sign up to be connected with other members that live near them. We are hoping to launch this program by the end of 2020, so, if you haven't already, be sure to become a member of the Foundation and keep an eye out for our announcement when the program is live!

## Jackson (6y)
### Deletion- US

**When was Jackson diagnosed with 17q12 deletion syndrome?**
He was 4 years old when he was diagnosed. He had been diagnosed with Autism Spectrum Disorder at 2 years 6 months. His developmental pediatrician wanted us to do genetic testing which is how we found out about the deletion. Jackson has an older brother with Autism but does not have the deletion.

**How does 17q12 affect his life?**
Jackson just turned 6 years old. He is nonverbal with global developmental delays. He also sees a nephrologist for cystic kidneys and has severe food allergies.

**What 5 words would you use to describe Jackson to the world?**
Jackson is happy, determined, fearless, tenacious, expressive.

## Taylor (14y)
### Duplication-US

**When was Taylor diagnosed with 17q12 duplication syndrome?**
By age 8 she was diagnosed with ADHD, and her IEP included supports in academics and speech. We saw a neurologist and she was diagnosed with 17q12 duplication. She was diagnosed with nighttime seizures shortly after that and started taking medication. By the age of 11 there weren't signs of seizures and we stopped the medication.

**How does 17q12 affect your life?**
The duplication affects me with school. I have teachers that help me and I work really hard. Sometimes it takes me longer to learn things.

**What are your goals for the future?**
when I start High School in September, I want to maybe try swim team, take Spanish , do yearbook. In the future, I want to go to College And be a Kindergarten Teacher.

## Bill (65y)
### Deletion-US

**When was Bill diagnosed with 17q12 deletion syndrome?**
Bill became ill a few weeks after turning 45. Just as our other older friends with 17q12 deletion syndrome, Bill was given different diagnoses after visiting 100's of doctors along the way. After going to over 5 of the best hospital systems for almost two decades we finally had an answer after they performed three genetic tests in August 2017.

**How has 17q12 affected Bill's life?**
This syndrome and its magnesium wasting side of it made 17 years of raising our family and trying to treat many life threatening incidents in that time truly unbearable. However,daily IV infusions, open heart surgery and other medical inconveniences has never deterred Bill from giving his family a fairly normal life of fun and travel.

**What accomplishments are Bill most proud of?**
Bill is a former state Game Commission officer and always champions for all animals and our environment. He is proud of all of his family and especially his grandson, Landon, who he shares his love of the outdoors and traveling with. He just celebrated 39 years of marriage with his wife, JoAnn.

# 17q12 syndromes Q&A

## Davey (3y)
### Duplication-US

**When was he diagnosed with 17q12 duplication syndrome?**
Davey was diagnosed at 1.5 years old.

**How does 17q12 affect his life?**
He is affected in many areas. Davey has oropharyngeal dysphasia, hypotonia, global delays, autism, constipation, acid reflux, aspiration, he has a gtube, speech delay, motor skill delay, short stature, and congenital hip dysplasia. He has come so far in the year that he has been in therapy, and we have so far to go. We take one day at a time.

**What 5 words would you use to describe Davey to the world?**
Courageous, Determined, Silly, Kind, Relentless

## Jack (9y)
### Duplication-UK

**When was Jack diagnosed with 17q12 duplication syndrome?**
He was diagnosed with the duplication aged 8.

**How does 17q12 affect his life?**
It has affected his life massively. He has autism, sensory processing difficulties, learning difficulties, very challenging behaviour, anxiety and hypermobility. All this affects his ability to cope in public, as he is hypersensitive on all his senses so he melts down when overstimulated. On a positive note he loves to bounce, he bounces all day long. He can talk forever and loves to learn about the human body.

**What is Jack's greatest superpower?**
His greatest superpower... Love. He will cuddle me from dawn til dusk if I let him.

## Sherie (41y)
### Duplication- US

**When were you diagnosed with 17q12 duplication syndrome?**
I was diagnosed at age 36.

**How does 17q12 affect your life?**
I now know that my learning difficulties as a child were directly related to 17q12 duplication, as was my “tired legs” (low muscle tone). I still deal with getting physically tired quicker than my peers when I’m very active, like long walks hiking or playing a sport. I also have some gastro issues caused by some food sensitivities and I suffer from sinus headaches and ocular migraines.

**What accomplishment are you most proud of?**
My biggest accomplishment was being able to be a successful woman in my career and still be able to be a great single mother to my two boys.

# 17q12 Deletion Syndrome

**Overview**: 17q12 deletion syndrome is an autosomal dominant condition where individuals are missing genetic material from one copy of their 17th chromosome. There are many symptoms of this condition that are detailed below, but it is important to remember that this is a variable condition; affected individuals may have only some or all of the features listed below, and unfortunately we cannot predict which features and/or how significantly someone may be affected.

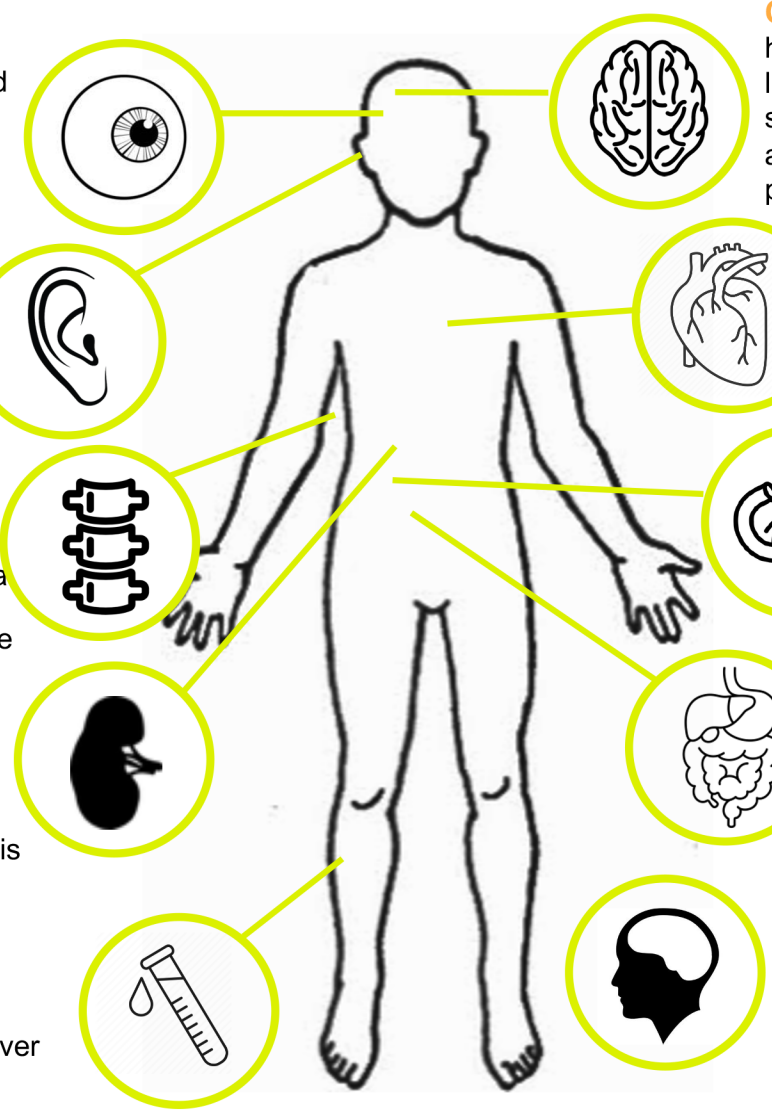
**Optic Features:**
Present in about 35% and include strabismus, nystagmus, cataracts, coloboma, and more.

**Hearing:**
Sensorineural hearing loss has been reported, but is not considered a common feature.

**Muscles/Bones:** Short stature, macrocephaly, hypotonia, joint laxity, long fingers, hip dysplasia

**Kidneys:** Over 80% have structural or functional problems with their kidneys. Kidney cysts are the most common structural change. Tubulointerstitial disease is the most common functional problem.

**Laboratory Features:**
Hypomagnesemia, hyperuricemia, elevated liver function enzymes



**Cognitive Differences:** >50% have developmental delay and/or learning difficulties. Seizures and structural differences of the brain are also more common among people with 17q12 deletions

**Heart Defects:**
Around 20% of babies born with 17q12 deletion have a heart defect.

**Pancreas Problems:**
About 25-50% have maturity onset diabetes of the young, type 5. Others may have a small or underdeveloped pancreas

**GI and GU Problems:**
Diaphragmatic hernia and structural anomalies affecting the GI tract and the reproductive organs.

**Psychiatric Illness:** Autism, schizophrenia, attention deficit disorders, anxiety, are more common among people with 17q

**Management**: The treatment, screenings, evaluations and general management of an individual with17q12 changes with age, and depends on what symptoms the person is presenting with. In general management includes routine bloodwork, annual ophthalmology and audiology evaluations, initial cardiac and renal screenings, and appropriate developmental interventions if the individual has developmental delay, speech and language delay or is presenting with difficulties in school. Additionally, parents must be vigilant in monitoring for changes in their child's behavior, emotional state and thinking; they should see a psychologist if there are any concerns for mental health disorders.

# 17q12 Duplication Syndrome

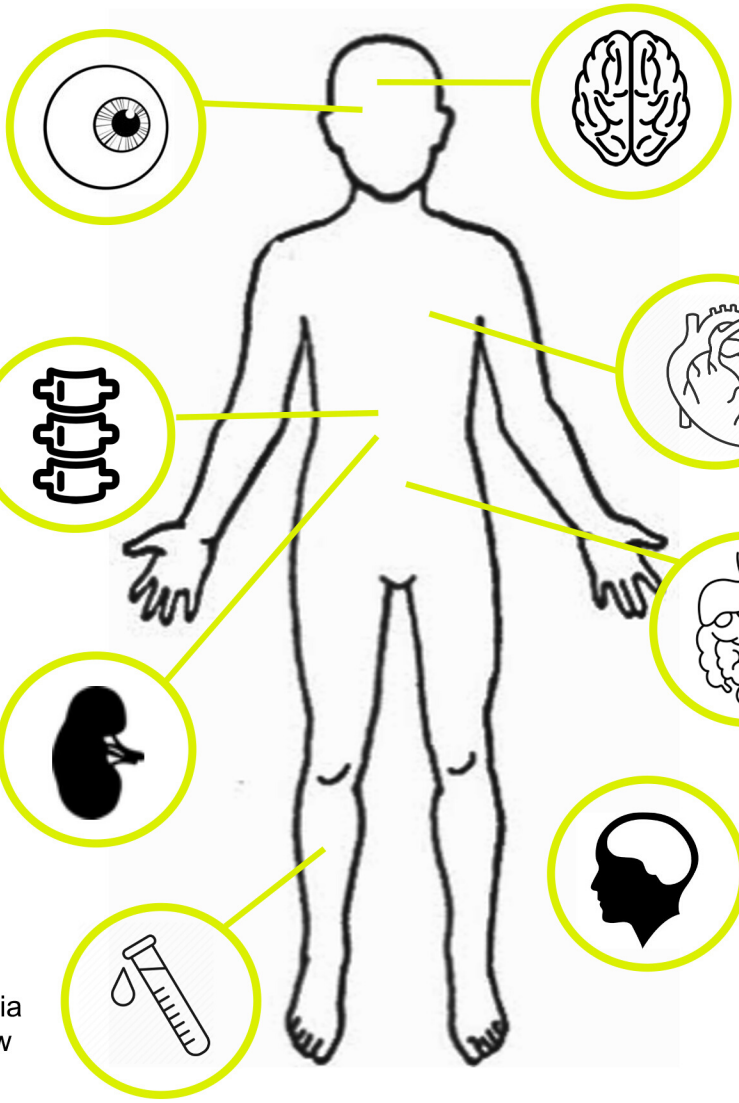
**Overview**: 17q12 duplication syndrome is an autosomal dominant condition where individuals have extra genetic material on one copy of their 17th chromosome. There are many symptoms of this condition that are detailed below, but it is important to remember that this is a variable condition; affected individuals may have only some or all of the features listed below, and unfortunately we cannot predict which features and/or how significantly someone may be affected.

**Optic Features:**
Present in about 45% and include strabismus, astigmatism, microphthalmia, coloboma, and more.

**Muscles/Bones:** Short stature, microcephaly, hypotonia, joint laxity, small hands/feet, and/or chest abnormalities

**Kidneys:** Structural differences of the kidneys including solitary kidney, horseshoe kidney and kidney cysts have been reported in about 25% of individuals.

**Laboratory Features:**
Hyperkalemia, hyponatremia, hypoglycemia have been reported in a few individuals



**Cognitive Differences:**
Cognitive abilities range from typical to severe developmental delays. Seizures and structural differences of the brain are also more common among people with 17q12 duplications.

**Heart Defects:** Some individuals with 17q12 duplications have been reported with various structural heart differences.

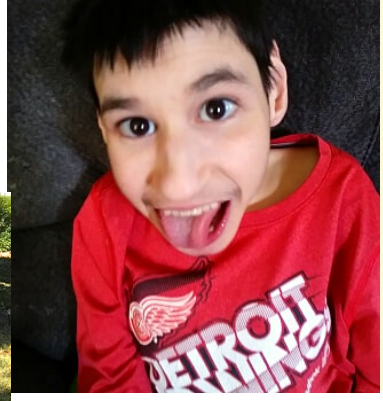
**GI Problems:**
Tracheoesophageal fistula and other structural anomalies affecting the GI tract. Many individuals also have various feeding difficulties.

**Psychiatric Illness:** Autism, schizophrenia, aggression, self-injury, and compulsive disorders are more common among people with 17q duplications

**Management**: The treatment, screenings, evaluations and general management of an individual with17q12 changes with age, and depends on what symptoms the person is presenting with. Initial screenings following a diagnosis include an ophthalmology evaluation, echocardiogram, kidney ultrasound and screening for GI problems. Routine screening is focused on appropriate developmental interventions if the individual has developmental delay, speech and language delay or is presenting with difficulties in school. Additionally, parents must be vigilant in monitoring for changes in their child's behavior, emotional state and thinking; they should see a psychologist if there are any concerns for mental health disorders.

dear world, this is

# #HowWeFly



## Samuel (18y)

### MI, USA

*Samuel said Mama on Mother's day for the first time ever. Now he says it a million time a day!*

## Liam(6y)

### CA, USA

*Liam learned how to ride a bike this year!*

## Greia Lliana (9mo)

### AUSTRALIA

*Greia Llana Is sitting on her own after a 2nd trans-abdominal surgery to correct Duodenal and jejunal atresia, bowel perforation and Malrotation.*

## Gunnar(3y)

### CA, USA

*Has recently become so independent and social at preschool. He says "all done" and "down" and knows many signs. He loves the ocean, and reading!*

## Kyle (10y)

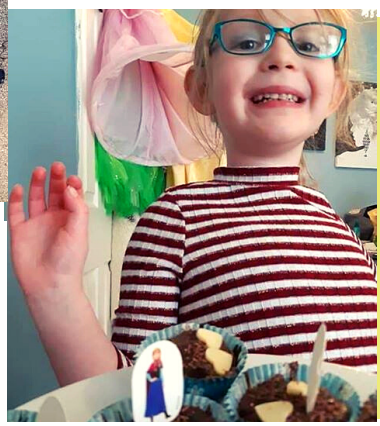
### UK

*Kyle is trying new foods and is really trying to overcome his anxieties. He brightens his families darkest days!*

## McKenzie(4y)

### NY, USA

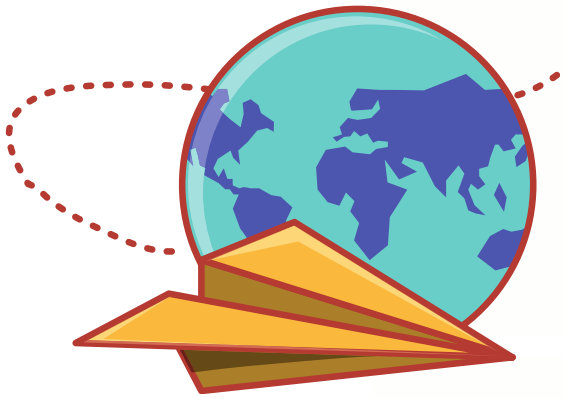
*Learned how to ride her tricycle all by herself. Her parents are proud of her for trying to improve her speech and language. She is learning to talk much clearer after her cleft palate repairal procedure.*

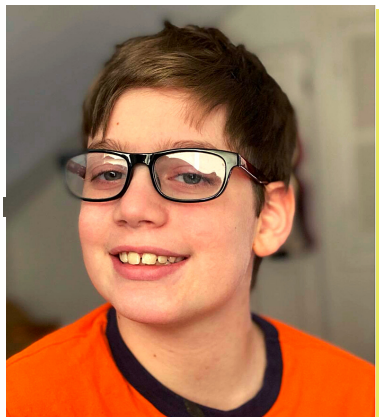


## Joyann (6y)

### UK

Has been really helpful during lockdown and her biggest achievement is helping make cakes with her daddy.





## Alex (11y)

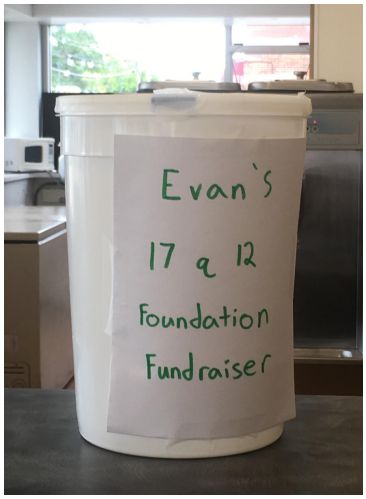
### OH, USA

Has been getting better with his music on his dj app and playing his percussion instrument. He has also become a better reader and is a quick learner when it comes to math!

chromo17q12.org


# A MESSAGE TO OUR SUPPORTERS

17q12 Foundation is a registered non profit organization that is fully funded by donations and run by five individuals volunteering their time; four parents and one genetic counselor.. We are grateful for all of the support we have received since beginning in 2017. Through donations big and small and a handful of fundraisers benefiting 17q12 Foundation; we have been able to reach a couple of our larger goals. Last year's family conference was made possible thanks to our sponsors: PRISMA, SPARK, Geisinger Caring, and Bradley Hospital. We are so grateful to the Artemenko family for matching the funds raised from the Casados family's successful art fundraiser. Along with a generous donation from the Palisades Foundation we surpassed our goal of raising $15,000. Looking into the next year, we hope to work towards more goals. Last year we met our fundraising goals in order to start a medical registry. Now that we have met that goal we will need to cover annual maintenance costs of the registry. We know that if we all work together we can get there. We have had a lot of new families join us in the last year, and we hope to work towards another family conference in the future. Every fundraiser will bring us closer to accomplishing these goals. Please consider putting on your own fundraiser benefiting 17q12 Foundation.. Please get in touch with **liz@chromo17q12.org** to get started. Thank you to everyone who has supported our community along the way!




# RESTAURANT FUNDRAISERS

There are many restaurants throughout the country that set up special fundraising events for organizations like ours. They donate a percentage of sales to a cause. You can find restaurants in your area by visiting: **groupraise.com**

**Here is a list of some restaurants:**

- Papa John's "Dollars for Dough" - Donates back 25% with flyer or mention
- Chipotle Donates back 50% with flyer or mention

# FUNDRAISER IDEAS AND WAYS TO GIVE BACK

We know that this has been a difficult time for many. We hope that this can be a useful guide for future fundraising

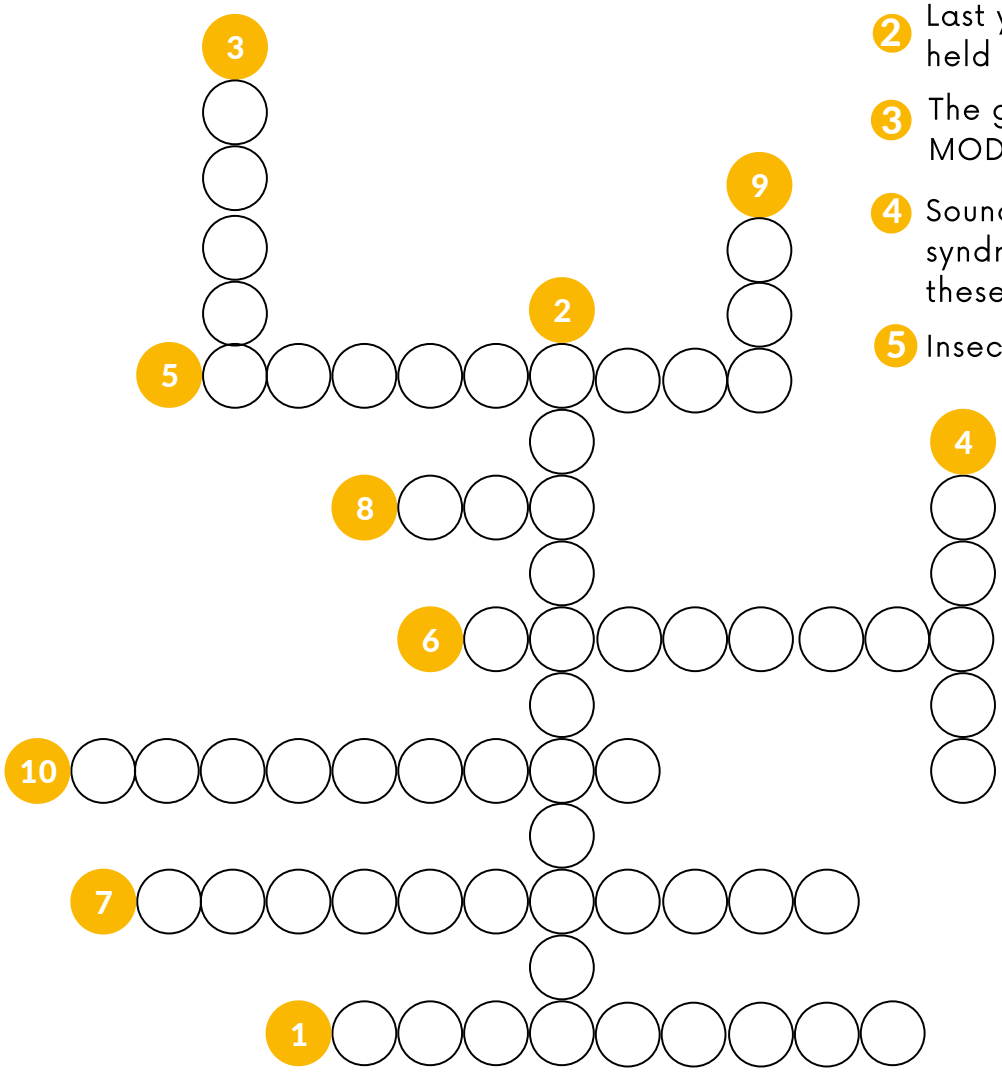
**Fundraising in your community:**

- Relaunch a past shirt campaign or start your own fundraiser through Bonfire to benefit 17q12 Foundation. Visit: **bonfire.com/store/17q12-foundation/**
- Plan a ride, hike, swim, or run for 17q12. Ask supporters to make a pledge per mile.
- Amazon will donate 0.5% of the price of your eligible AmazonSmile purchases toward 17q12 Foundation when you shop through Amazon smile. Visit: **smile.amazon.com**
- Organize a silent auction. Ask businesses, friends, and family if they can donate items to be auctioned off.
- Donations are welcome and appreciated throughout the year, and are tax deductible. **chromo17q12.org/donate**

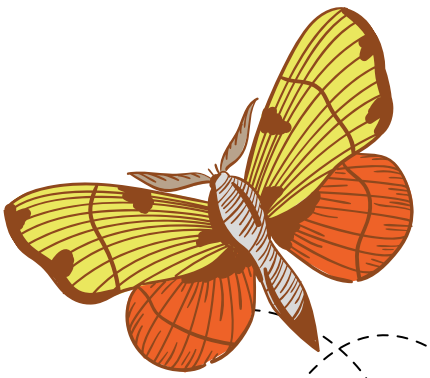

- Arby's -10% of sales for 3 hour period
- Blaze Pizza - Donates back 20%
- Chick-fil-A
- Noodles & Company
- Panda Express-Donates back 20% with flyer or mention
- Orange Leaf
- Panera Bread-Donates back up to 20% (depending)
- Potbelly-Donates back up to 25%
- Red Robin
- Coldstone Creamery
- Fazolis-Donates back 20% of net sales

# 17q12 Crossword

Can you find all of the answers and complete the crossword puzzle?




1. 17q12 Foundation represents two different _____.
2. Last year's family conference was held in this state.
3. The gene that causes kidney cysts, MODY5, and kidney abnormalities
4. Sounds like a type of pants. 17q12 syndromes are caused when 15 of these are missing or extra.
5. Insect that represents 17q12.



6. When a part of the chromosome is missing.
7. When there is an extra part of the chromosome.
8. seventeen-q-one-_____
9. How We ____
10. July 12 is 17q12 _____ day.

Awareness Day 2020

Name: Age: Date:

# Today you are you, that is truer than true. There is no one alive who is Youer Than You. –Dr. Seuss

I'M HAPPIEST WHEN I AM...

MY FAVORITE TOY OR GAME IS...

I'M REALLY GOOD AT.....

MY FAVORITE PLACE TO BE IS...

IF I HAD A SUPERPOWER IT WOULD BE...

# 17q12 Deletion syndrome Management checklist

## AT DIAGNOSIS

- [ ] Kidney Ultrasound
  - Date:_____
  - Results:_____
- [ ] Pelvic ultrasound in females
  - Date:_____
  - Results:_____
- [ ] Hearing evaluation
  - Date:_____
  - Results:_____
  - Follow-up: _____
- [ ] Ophthalmology exam
  - Date:_____
  - Results:_____
  - Follow-up:_____
- [ ] Endocrinology evaluation
  - Hemoglobin A1C level:_____
  - Follow-up:_____
- [ ] Genetics Evaluation
  - Date:_____
  - Follow-up:_____
- [ ] Cardiology Evaluation
- [ ] Echocardiogram:_____
  - Date:_____
  - Results:_____
- [ ] Nephrology Evaluation
- [ ] Labs
  - Serum BUN_____
  - Creatinine_____
  - Electrolytes_____
  - Calcium_____
  - Magnesium_____
  - Phosphorus_____
  - Uric Acid_____
  - Urine Magnesium (24 hr)_____
  - Urine Creatinine (24 hr)_____
- [ ] Liver Function Labs
  - AST_____
  - ALT_____
  - GGT_____
- [ ] Neurology (if concern)
  - Date:_____
  - Follow-up:_____
- [ ] Developmental Assessment

  (through school, neuropsychologist, developmental pediatrician, etc.)
  - Speech_____
  - Motor skills_____
  - Cognition_____
  - Behaviors_____
  - Follow-up:_____

# 17q12 Deletion syndrome Management checklist

## ANNUAL SURVEILLANCE

- [ ] Kidney Ultrasounds
- [ ] 12 months after diagnosis

Date:_____

Results:_____

- [ ] Every 2-3 years in childhood
- [ ] Every 3-5 years in adulthood

*If abnormalities are present, frequency of ultrasounds should be determined by nephrologist

- [ ] Audiology exams

Date:_____

Result:_____

Date:_____ _____

Results:_____

Date:____:_____

Results:_____:_____

- [ ] Developmental re-assessment

_____

_____

_____

_____

- [ ] Labs

Hemoglobin A1C_____

Serum magnesium_____

Potassium_____

Uric Acid_____

Urine Magnesium (24 hr)_____

Urine Creatinine (24 hr)_____

- [ ] Ophthalmology exam

Date:_____

Results:_____

Date:_____

Results:_____

Date:_____

Results:_____

NOTES

# 17q12 Duplication syndrome Management checklist

## AT DIAGNOSIS

- [ ] Renal Ultrasound
  - Date:_____
  - Results:_____
- [ ] Nephrology evaluation if abnormal
  - Date:_____
  - Results:_____
- [ ] Ophthalmology Exam
  - Date:_____
  - Results:_____
  - Follow-up: _____
- [ ] Cardiology evaluation
- [ ] Echo cardiogram
  - Date:_____
  - Results:_____
- [ ] Developmental Assessment

  (Neurodevelopmental pediatrician, psychologist/psychiatrist)
  - Date:_____
  - Diagnoses:_____
  - Follow-up:_____

NOTES

# 17q12 Duplication syndrome Management checklist

## ANNUAL SURVEILLANCE

- [ ] Ongoing follow-up with specialists if abnormalities present

- [ ] Specialist:_____
  Date:_____
  Results:_____
- [ ] Specialist:_____
  Date:_____
  Results:_____

- [ ] Neurology evaluation
  *if concerns for seizures
  Date:_____
  Results:_____
- [ ] GI evaluation
  *If feeding concerns
  Date:_____
  Results:_____

- [ ] Developmental Assessments
  (Neurodevelopmental pediatrician, psychologist/psychiatrist)
  Date:_____
  Diagnoses:_____
  Follow-up:_____

NOTES