

17q12 Interest Group

17q12 Review

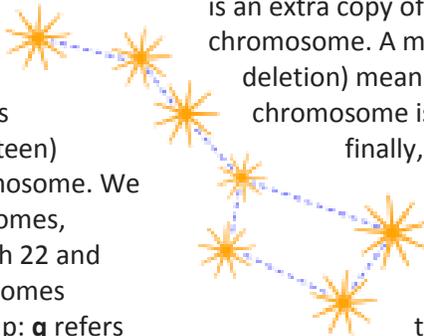
Spring 2017

IN THIS ISSUE

What's in a name?

When compared to an ear infection or the flu or heart disease, **17q12 microduplication or microdeletion syndrome** sounds like a mouthful! Genetic conditions can sometimes have complicated-sounding names, but they are really just specific descriptions of the genetic change that causes the condition. Let's break it down: **17** (seventeen) refers to the whole chromosome. We have 23 pairs of chromosomes, labelled number 1 through 22 and then the X and Y chromosomes determine gender. Next up: **q** refers to the bottom of chromosome number 17. The top is labeled "p" because it is smaller than the bottom, or petite. The bottom is labeled "q" because q comes after p in the alphabet. Then we have **12** (pronounced one-two). This helps us narrow in on the exact location or band on the bottom of chromosome 17. If we think of it like a map, 17 is

the state, q is the city, and 12 is the house number. **Microduplication or microdeletion** refers to what happened at that location. A microduplication (it's also okay to just call it a duplication) means there is an extra copy of that part of the chromosome. A microdeletion (or deletion) means that part of the chromosome is missing. And finally, a **syndrome** is a constellation or group of symptoms that consistently occur together. 17q12 deletions are commonly associated with kidney problems, diabetes, and learning difficulties while duplications are commonly associated with speech delay, learning difficulties and seizures (among other symptoms, of course). As always, a diagnosis does not define who a person is or can be.



Parental Testing: What, Why, & How

17q12 deletions and duplications may be inherited from parent to child. Oftentimes, it can be a challenge to get parental testing completed. This article will give some helpful pointers

for navigating your way through this confusing process.

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17q12 Interest Group
51q12 Family Conference
Chicago, IL
July 14-16, 2017

SAVE THE DATE!
July 14-16, 2017

We Need You!
We're looking for you to help us plan the conference. We are seeking anyone who has experience with genetic testing or experience that would be helpful to help fund the conference.

If you have any testing experience or links, please contact Melissa Turner at melissa@17q12.org or call 773-224-2340.

Thank you to all who participated in our survey!

Piktochart

Conference Updates

Learn about the tentative agenda, social event, childcare, registration information and hotel information.

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Parental Testing: What, Why, & How

Genetic conditions are unlike any other illness or disease because some of these conditions can be passed down through families. Because of this, genetic testing often does not end when one person is diagnosed. This is particularly true with 17q12 deletions and duplications, because we know that the range of symptoms can vary from nearly undetectable to severe. For this reason, **parental testing is recommended** when a child is found to have either a deletion or duplication. Even if it doesn't seem like a parent has any of the symptoms or features associated with the deletion or duplication, it's possible that he or she could have it, too.

It is important to keep in mind that not all deletions and duplications are inherited from a parent. It could be brand new in the child, or *de novo*. After all, even in families where it is passed down through generations, it had to start with someone! More importantly, nobody can choose which chromosomes they pass on to their children. **Passing on a deletion or duplication to a child is not anyone's fault.**

If a child is found to have a 17q12 duplication, **9 out of 10 times** it was inherited from a parent, whereas only **3 out of 10 deletions** turn out to be inherited from a parent.

For any individual known to have a deletion or duplication, there is a 50% chance that each child will also have the deletion or duplication.

Inheritance Statistics Breakdown		
	Inherited from a parent	De novo (brand new)
Duplications	90%	10%
Deletions	30%	70%

Once a person is known to have a deletion or duplication, there is a 50% chance that each of his or her children will also have it

WHAT: The test that is recommended for parents is usually a targeted version of the test the child had done. In most cases, it's probably called a targeted chromosomal microarray. This recommended test will almost always be indicated on the child's test report.

So, **WHY** is it recommended for parents to be tested? It is useful to understand if other children should be tested, and it's important to know for future children as well. If a duplication, for example, is identified in a child and his father, there's a 50% chance that any other children from that father would also have the duplication. Or, if a mother has some kidney problems, and had some difficulty in school, finding out about her own deletion because of her daughter's diagnosis could finally give her an explanation for her own experience.

HOW: The doctor who ordered the child's testing will sometimes order parental testing as well. Other times, the child's doctor will refer parents back to their primary care doctors to order testing. If the test was initially ordered through a geneticist/genetic counselor, they should be familiar with ordering parental testing. If the initial testing was ordered by a different doctor; it can sometimes be more of a challenge to get parental testing done. Regardless of who

ordered the initial testing, parents can take a copy of the child's report to their primary care doctor and request that the test be ordered. Another option is to go to the National Society of Genetic Counselors website (nsgc.org) and click on "Find a Genetic Counselor". Your local genetic counselor should be able to point you in the right direction to get the testing done. If insurance coverage is an issue, a letter of medical necessity provided by a doctor will often do the trick and get it covered. Any doctor can provide the letter, but some doctors turn to a genetics doctor or genetic counselor for assistance.

The Bottom Line: If you want parental testing; persist, persist, persist!

2017 Family Conference Updates

Planning for the family conference is well underway! We will have a jam-packed day of educational sessions on Friday, July 14th with an optional field trip to the Lincoln Park Zoo on Saturday.



Bring your child's lab report if you are interested in research opportunities or have genetics questions for the curbside consult!

Register Here:

<https://www.surveymonkey.com/r/gB8XVWV>



**REGISTRATION
DEADLINE:
APRIL 17, 2017**

Hotel Reservations:

<http://bit.ly/2lKHUHS>



**RESERVATION
DEADLINE: JUNE
22, 2017**

Childcare Info



A professional childcare company will be provided for children with and without 17q12 for Friday the 14th. If you are interested in this service, we must have your registration on file by April 17th!

[Tentative] Agenda

Friday, July 14, 2017

- Breakfast (provided)
- 17q12 Overview
- MODY-5
- Seizures
- Behavior strategies
- Lunch (provided)
- Kidneys
- Speech/Language
- Research updates
- Curbside Consults

Saturday, July 15, 2017

- Support group interest session
- Lincoln Park Zoo (bus to and from will be provided)

Child Spotlight



Isaac is 4 years old and lives near Denver, Colorado with his parents and three older brothers. He was diagnosed with 17q12 duplication after he was diagnosed with high functioning autism when he was 2 years old. His family considers his diagnosis a blessing because it led to the detection of a minor heart abnormality that is best managed when it is found before symptoms appear. He also benefits from speech therapy to help with his

stutter. His mom describes him as a fun, active boy who loves preschool and playing with friends. He enjoys raiding the costume box, too! On any given day, he can be a doctor, a superhero, a chef, or an astronaut. Isaac has recently started ski lessons and did a fantastic job. His instructor said he was fearless. He has started reading and expected to excel in kindergarten next fall. Look out NASA, here comes Isaac!

“Research is Creating New Knowledge”

- Neil Armstrong

Geisinger's Autism & Developmental Medicine Institute is conducting ongoing 17q12 research, studying how genetics and family background affect the clinical features in children and adults with a 17q12 deletion or duplication, through online surveys and using saliva samples for genetic studies. Children and adults with 17q12 and their family members can all participate -- **from home!**

As compensation for your time, families who submit genetic testing results and complete all surveys are eligible for an Amazon gift card.

Currently, we have **61** families who have consented to participate and **41** of those families have completed all or some of the surveys. Several families have come to visit us in Lewisburg, PA for direct assessments, as well.

If you're interested in getting involved in our research study, please contact Stefanie Turner (sturner5@geisinger.edu).

If you've started and haven't finished, or aren't sure where you left off, we are happy to help you get back on track! **A huge thank you to all our participants - we cannot do this important work without you!**

It's Easy As...

Our geneticist reviews the genetic lab report for the person with the deletion or duplication in order to verify that it's the region we are studying. If that is confirmed, we ask for genetic test results for any other family members who have been tested. This lets us know whether or not the deletion or duplication was inherited.

1

When genetic results are confirmed, we send out links for **3 online surveys** for each family member. The surveys ask for information about **personal traits, daily habits, and lifestyle**. We also send saliva sample kits to all family members. Together, the information about family genetics, environment, and lifestyle help us to describe how a range of symptoms can occur in different people with 17q12 deletions or duplications.

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Families can also participate in some face-to-face research to assess different types of skills in each family member. This is an **optional** portion of our study and can be done in **Lewisburg** or at the Family Conference in **Chicago** – look for our booth on July 14th to learn more!

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Child Spotlight

Tyler is almost 8 years old and lives in Cambridgeshire, UK with his parents and younger sister. He was finally diagnosed with 17q12 deletion syndrome at 6 years old, after multiple trips to several different doctors with no answers for Tyler's medical and developmental concerns. Tyler had a scary start to life when a diaphragmatic hernia (a hole in the diaphragm that allows abdominal organs to push into the chest) was

detected during a routine ultrasound, but he had a successful surgery after he was born to correct it. He still has some medical concerns in addition to some developmental challenges, but his parents are looking forward to getting Tyler started in a new school for kids with special needs this fall! Tyler loves all sorts of trucks and tractors. His mom says he is brilliant with an iPad and has an excellent memory.



Contact Us

We want to hear from you! Tell us what you like, dislike and want to see in the next newsletter. Our goal is to make this as informative as possible for our readers, so we need your feedback. We are always on the lookout for family stories to share. Also, if you're interested in writing for our newsletter or getting more involved with the group, please reach out to us!

