17q12 Interest Group

17Q12 Review

Fall 2016

IN THIS ISSUE

Webinar Series

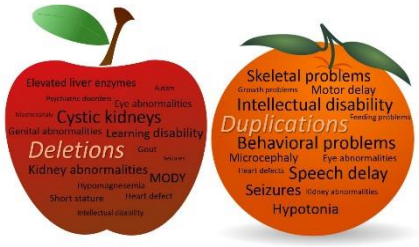
A diagnosis is often viewed as the end of the diagnostic odyssey. Once you have that, you finally have all the answers to your questions, right? Unfortunately, that is not the case with many genetic diagnoses and some families find themselves with even more questions after the diagnosis than before. We understand how frustrating it can be to have an "answer" but still not know where to go or to whom to turn to for medical care.

In an effort to answer some of those remaining questions, we will be launching a series of webinars focused on specific topics that your families deal with on a regular basis. We hope this will arm you with the most up-to-date information and strategies that will help you and your child's healthcare team provide the very best care for your child.

17q12 Webinar Series

- Features, Genetics, & Research, Oh My!
- Kidneys 101
- Evidence-based treatments
- Behavior strategies for home and school

Links to each webinar will be posted on the Facebook support group page as well as the Geisinger 17q12 webpage (http://www.geisingeradmi.org/care-innovation/studies/17q12/). Contact information will be provided at the end of each webinar to submit questions. Each presenter will compile the questions, and we will post an FAQ page with the answers.



Apples and Oranges: What's the difference between deletions and duplications?

17q12 deletions and duplications may share the same chromosome, but they are different conditions from a genetic standpoint and a symptom standpoint.

Page 2

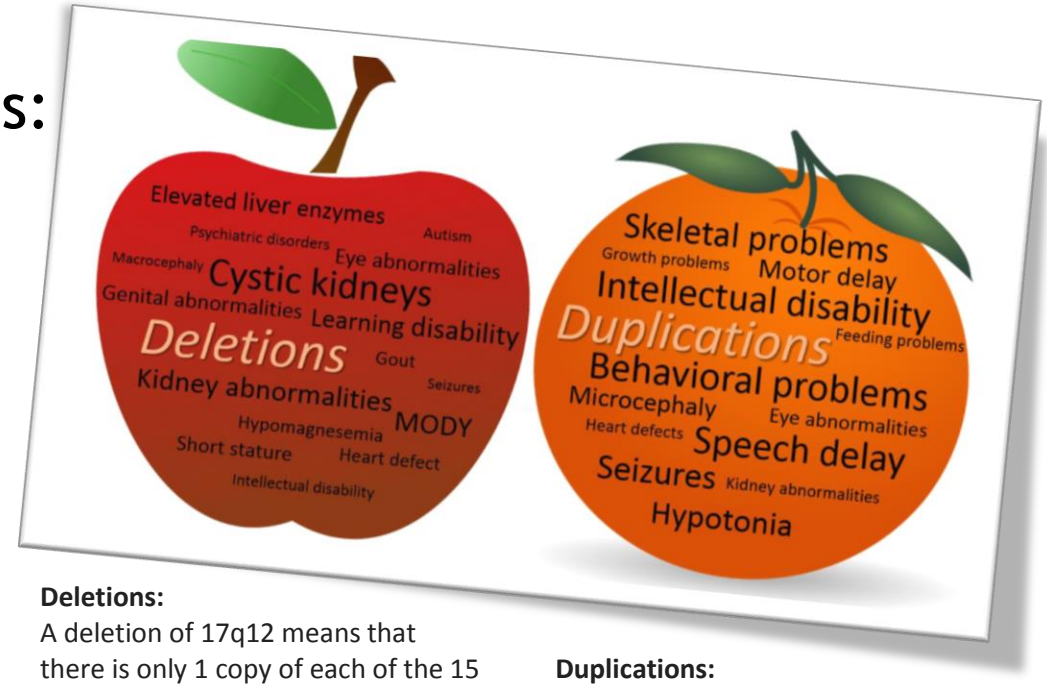
Let's go public!

We need a public presence in order to raise awareness of 17q12 deletions and duplications. Starting an official, public website will be one step toward establishing a non-profit and ensuring your voices are heard.

Page 3

Apples & Oranges:

What's the difference between deletions and duplications?



Let's start with a quick flashback to high school biology. Our bodies are made up of trillions of tiny little structures called cells. Inside these cells are smaller structures, including chromosomes (our instruction manual), mitochondria (our energy factories), ribosomes (our protein factories), etc. Every cell has a full set of chromosomes (46 total) that come in pairs and contain all of our genes. We get one set from our mother and one set from our father. Each gene contains instructions, or a "recipe," to make a protein that serves a specific function within our body. Genes are turned on and off at different times in development and in different types of cells. Scientists don't yet understand all the details of these processes, nor do we understand the function of every gene.

Deletions and duplications can happen anywhere on any of our chromosomes. Certain areas of some chromosomes, however, are more prone to deletions and duplications. The long part of chromosome 17 ("q") at band 12 is one of those areas, so it is often referred to as a *recurrent* microdeletion or microduplication. Many individuals with a change at 17q12 have a deletion or duplication of about 1.4Mb (megabases = 1 million bases or "letters"), however some people have smaller or larger changes. The 1.4 million bases of DNA in this region contain about 15 genes, but science has not figured out the role most of them have in our body, yet.

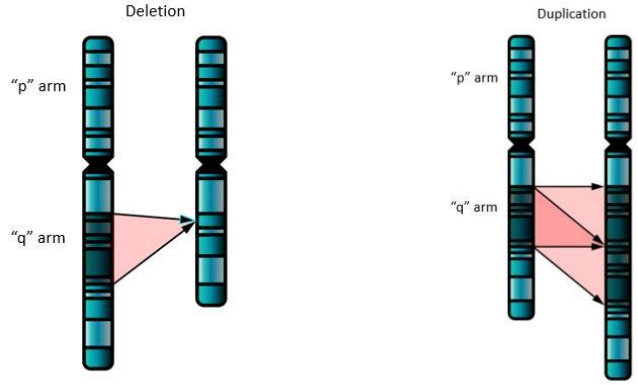
Deletions:

A deletion of 17q12 means that there is only 1 copy of each of the 15 genes in this region instead of the 2 copies that most people have. In theory, that means that people with the deletion have about half as much protein that works the way it's supposed to. The clinical features and symptoms (phenotype) of people with 17q12 deletions is not fully understood yet, but we know that it must have something to do with not having enough protein from 1 or more of the 15 genes that are deleted. The challenge lies in trying to determine which genes are responsible for which features and why those features are present in some people, but not everyone with the deletion.

Duplications:

A duplication of 17q12 means that there are 3 copies of each of the 15 genes in this region instead of the 2 copies that most people have. Instead of having too little of the protein, these individuals have too much of the protein. The clinical features and symptoms of these individuals are mostly different than the features of individuals with the deletion. Why? Well, the body compensates for too much protein differently than it does when there isn't enough. The details of these processes are, again, not well understood.

The Bottom Line: Even though we're talking about the same 15 genes, we're talking about 2 different conditions!



Let's Go Public!

As we live and breathe in the age of the Internet, it is time for the 17q12 group to have its own official website! Many, many successful non-profits related to genetic conditions have started with a simple website that has gone on to blossom into a fantastic resource serving multiple purposes. **A website serves as a "home base" for the many families around the world affected by 17q12 deletions and duplications.** While the Facebook group is a wonderful place to find support, it is difficult to locate unless you know the exact search term. **Having a website that pops up on a Google search makes it much more accessible to the many families seeking information and support.** Furthermore, a website creates a public presence so that medical professionals and researchers with an interest in this group can get involved more easily. There is still much to be learned about both 17q12 deletions and duplications! It is essential to increase awareness and interest within the research community so that we can learn more about these conditions and, in turn, provide better care for you and your families.

We are looking for individuals with a background in IT and/or website design who would be interested in getting a website up-and-running. The folks here at Geisinger are more than willing to help with the content, but we hope the website will ultimately be maintained by your group. After all, this is your party; we're just trying to help get it started!

WE NEED YOU!

If you are interested, please contact:

STEFANIE TURNER
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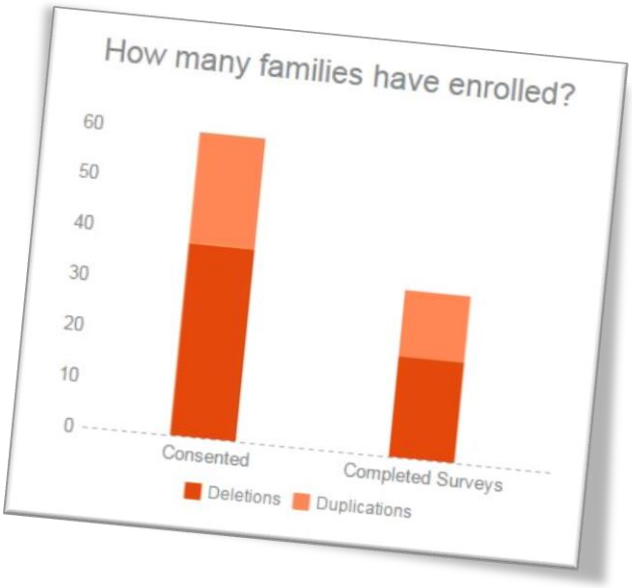
Child Spotlight

Sophia is 5 years old and lives in upstate NY. She was recently diagnosed with 17q12 duplication after her parents sought an evaluation for speech and developmental delays, sleep issues, and ADHD. She is the youngest of 5, and her older siblings adore her and love to give her attention. She enjoys looking at books and being read to- she is rarely without a book in her hand! She also loves her many pets, including 2 Saint Bernards and 4 cats. Sophia has recently started playing with baby dolls and engaging in pretend play. Her mother describes her as loving, empathetic, and having a wonderful memory. Although she struggles with social anxiety and tantrums that can be difficult to manage, she has been a blessing to her family. Sophia's mother states, "She is going to be the child that slows us down and makes us look at life differently, and that's a good thing." Good luck in kindergarten this year, Sophia!

Research is the Gateway to Better Healthcare

Geisinger's Autism and Developmental Medicine Institute has an ongoing 17q12 research study, the goal of which is to better understanding the behavioral profile, also known as a phenotype, of 17q12 deletions and duplications (i.e. what do they look like from a behavioral standpoint) as well as finding out how a person's genetic background (all of the other genes) affect how a 17q12 deletion or duplication is expressed in a particular person.



Our geneticist reviews genetic testing results 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 if the deletion or duplication was inherited.

Once genetic test results are confirmed, we send out links for a series of online surveys for each family member (whether they have the deletion/duplication or not). This gives us information about the behavioral characteristics for each family member as well as information about how family background may influence the expression of the deletion or duplication.

For families willing and able to travel to our center in Lewisburg, PA, we also have some face-to-face assessments, such as cognitive testing, fMRI (brain scans), and motor tasks. This is an **optional** portion of our study and is useful for getting more in-depth information about the behavioral phenotype.

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

Currently, we have 52 families who have consented to participate and 34 of those families have completed all of the surveys. Additionally, we have had several families come visit us in Lewisburg for direct assessments! **If you're interested in getting involved in our research study, please contact Stefanie Turner.** 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 of our participants- we cannot do this without you!**

On the Horizon

The next 17q12 Family Conference will be held in Chicago, Illinois from July 14-16, 2017! We are hoping to secure some grant funding to help offset the registration fees, however we need your help! If anyone has any connections to private foundations and/or corporations that may be willing to sponsor our event, please let us know.

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!