Second Family Meeting Planned June 26-27

Geisinger’s Autism & Developmental Medicine Institute (ADMI) is excited to announce the second 17q12 family meeting in Philadelphia! Last year’s meeting included 11 families from 9 states, and we are expecting an even larger turnout this year.

The conference will be held over two days. A more detailed schedule of speakers and sessions will be sent out closer to the conference, but an overview of the two days follows:

**Friday, June 26**

Registration and breakfast will begin at 7:30. Professional childcare will also open at this time and will continue throughout the day’s events. Expect presentations about a variety of topics related to 17q12 deletions and duplications, including information about clinical symptoms, medical management, and special education. New this year will be a family panel, where parents will share their personal experiences with 17q12. The day will close with personalized “curbside consults” where families can ask questions and talk to healthcare professionals about a variety of topics, such as kidney issues, genetics, medication management, and speech/language disorders. Families will also have the opportunity to participate in research.

**Saturday, June 27**

Saturday will include a child-friendly trip to the Adventure Aquarium in Camden, just across the river from Philadelphia. A bus is available for those families who will not have a car. We expect to return to the hotel around 1:00 pm.

To register, email mwmitchel@geisinger.edu
Child Spotlight: Meet Alex

When Alex was a baby, he needed surgery to reconstruct his intestinal tract. At 9 months old, he was also diagnosed with Duane syndrome and blocked tear ducts. After trying to find a reason for his frequent migraines, Alex’s doctor recommended genetic testing and Alex was diagnosed with 17q12 a microduplication when he was almost 4 years old.

Now Alex is 6 and he is incredibly caring and considerate. He hates to disappoint anyone! He has few fears and is very independent. His favorite things to do are watching cartoons and movies, playing games, riding his bike, and taking swimming lessons. Alex’s mother describes him as “marching to the beat of his own drum.” He typically likes to do his own thing, but he has recently started following some friends.

Keep up the good work, Alex, and we look forward to meeting you at the family meeting in Philadelphia in June!

Research Update

Geisinger’s Autism & Developmental Medicine Institute continues to research the behavioral, developmental, and medical characteristics associated with 17q12 deletions and duplications. The current study has two parts. The first part involves filling out a series of online surveys about each family member, which can be done from home. The second part is optional for those who are able to travel to ADMI in Lewisburg, PA, and involves several short assessments, including eye-tracking, a fine motor task, a measure of balance, cognitive assessments, and fMRI (brain scans).

About 20 families have participated in some or all of these aspects of the study. Thank you so much to all our participating families! We are still looking for additional participants, however. As compensation for families’ time and effort, $50 gift cards are offered for completing surveys and providing genetic testing results. Financial assistance may also be available for travel expenses. For more information, please contact Marissa Mitchel. (mwmitchel@geisinger.edu)

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Genetics 101

By Jessica Goehringer, MS, LGC

The human body begins development as one cell at conception. That one cell has two copies of each of the 23 chromosomes, half from the mother, and half from the father, for 46 chromosomes in total. Cell division occurs and in the end; we have trillions of cells that make up our body and trillions of copies of the original 46 chromosomes.

Our genetic code (DNA) makes up each chromosome by forming a tightly coiled spring. Of the 23 pairs of chromosomes, 22 are the same in men and women. These are numbered in size from biggest (chromosome pair 1) to smallest (chromosome pair 22). The 23rd pair is called the sex chromosomes; women have two X chromosomes while men have an X and a Y chromosome.

Each chromosome contains hundreds to thousands of genes. Each gene contains a special DNA recipe that instructs our body to make something (like a protein) or perform a certain function.

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The genes are long stretches of DNA that lie back-to-back on each chromosome, and we have about 20,000 genes! The DNA code that makes up the 20,000 gene contains about 3 billion “DNA letters” that we call bases pairs. The base pairs are abbreviated as A, T, C, and G and are found in a different sequence and different length in each gene. Together, all the genes contain the information to build and maintain a person’s cells and pass genetic traits on to children. This is called our genome. If we took all the DNA from each cell in our body and stretched it out, it would span our solar system more than once! Errors sometimes occur in our DNA code that result in certain symptoms or a genetic condition. Errors can be as small as a one letter change in the DNA code to as large as a section of a chromosome that is missing or extra (deletion or duplication) or a whole chromosome that is missing or extra. 17q12 duplications and deletions are examples of chromosome errors where symptoms are related to missing or extra pieces of genetic material.

**Child Spotlight: Meet Valentino**

Valentino is 2 years old and lives with his mother in New York City. Before he was born, the doctors noticed that he had cysts in his kidney on ultrasound. Because of this, amniocentesis was recommended and Valentino was diagnosed with a 17q12 microdeletion.

Valentino has a great personality! He is extremely loveable and compassionate, and his mother describes him as a “warrior of life.” He is also polite and always remembers to say “please” and “thank you.”

Valentino loves learning new things. His favorite things to do are watching movies, riding his scooter, listening to story time, and playing with his trains and cars.

Valentino and his mother are looking forward to the family meeting in June!

**Getting the Most from Speech Therapy**

By Marissa Mitchel, MS, CCC-SLP

Most individuals with 17q12 deletions and duplications will receive speech therapy at some point during childhood. Many parents find the transition between home-based early intervention and school-based speech therapy to be difficult since they no longer observe or participate in therapy sessions. Help your child make the most out of speech therapy by implementing a few simple ideas at home:

1. **Practice makes perfect.** You are your child’s first and best teacher. His speech therapist is only with him for a short period of time, so it’s important to work on speech goals at home to promote carryover and generalization. Help your child with any speech homework he might have, and request home practice activities if his SLP is not already providing these.

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4. **Focus on reading.** Children with a history of speech/language disorders are at risk for future reading difficulties. Engage in shared reading activities, especially books that feature rhyming and word play. Ask questions about the characters and events in the story such as, "What do you think will happen next?" Ask your child to retell the story in her own words.

5. **Provide a good speech model.** Use a slow rate of speech, break down directions if comprehension is a problem, ask one question at a time, and use simple but grammatically correct language. Set aside talking time each day that is free of distractions.

6. **Celebrate each goal met, however small.** Learning to talk is an incredibly complex endeavor and progress can be slow at times. Kids grow and learn at different paces and it’s important to acknowledge the hard work your child is putting in to improve his speech. Do monitor his progress towards goals over time, however, and consult with his SLP if it seems as though progress has plateaued.