Great Turnout for 2nd Family Meeting!

The second 17q12 family meeting was held in Philadelphia on June 26-27 and attendance more than doubled from last year! This year’s meeting included 20 families from 14 states as well as the UK and Australia. Speakers included medical and genetics professionals from Geisinger’s Autism & Developmental Medicine Institute as well as Dr. Ali Gharavi of Columbia University and Dr. Sara Pinney from Children’s Hospital of Philadelphia. Presentations touched on genetics, kidney issues, diabetes, medication management, and improving social skills. A parent panel shared families’ experiences and perspectives, and “curbside consults” enabled parents to ask experts questions about a variety of topics.

Many families chose to participate in research testing during the conference as well. Children and their families completed cognitive testing, eye-tracking, and measurements of balance and sway and fine motor skills.

On Saturday, families traveled by bus to the Adventure Aquarium in Camden, NJ. Despite rainy weather, children had a blast seeing penguins, watching hippos being fed, and walking through the famous “shark tunnel”! Thanks to everyone who helped make this year’s meeting a success!

Presentation Slides Posted to Website

Slides from the second 17q12 family meeting are available to download in PDF format from the ADMI website: www.geisingeradmi.org/17q12. Available presentations include:

- Overview of Genetics and Clinical Characteristics
- Psychotropic Medication Management
- Monogenic Diabetes in 17q12
- Effective Strategies for Social Skills Development
- 17q12 Research Update
**Child Spotlight: Justin**

Justin is 14 years old and recently began high school in Ontario, Canada. He loves to get up and go to school every morning, especially when he knows his daily routine. He is caring and sensitive and has a love for sports and cooking. He plays for a youth soccer club as well as a minor ball hockey league.

Justin’s mother reports that he has had a “tough go of things” in the past. Dilated, enlarged kidneys (hydronephrosis) were noted on prenatal ultrasound, and Justin had to spend time in the NICU after birth because of kidney and blood sugar issues. He required surgery to correct blockages in his kidneys, which has greatly helped although he has started to develop some small cysts.

Justin received early intervention services for developmental delays early in life and was eventually diagnosed with autism and intellectual disability. Genetic testing revealed a 17q12 deletion at age 5.

Justin takes medication and sees a psychiatrist to help with anxiety and other mental health concerns. His family suspects that he might have occasional seizures as well. Despite these concerns, Justin continues to do well at school and his family is proud of the progress he has made!

**Starting an Official 17q12 Nonprofit Group**

By Jennifer Voss

In June at the family conference, we began a discussion about forming our group into an official organization so we may continue to support each other, grow and hold conferences in the future.

Many people expressed interest in helping get an organization up and running, including filing necessary paperwork, looking into grants and sitting as board members.

I had the opportunity to do a little research and I spoke to several people who have experience in forming and running similar organizations. I also have personal experience in starting a nonprofit in 2007 in my home state of New Hampshire. From the conversations I had with Robby Miller of The National Fragile X Foundation and Emily Fields with PRISMS, I confirmed that the first thing we need to do moving forward is to file for 501c3 status with the IRS. There will be state-specific paperwork as well. We also need to elect a board who will be responsible for a variety of very important tasks. This is going to be a fairly large time and resource commitment for those who wish to serve, and it is important that people who are elected are active participants within our community and plan to be heavily involved in events and conferences in the future.

My last conversation was with Lauren Rosato at NORD. She is a mentor who helps new organizations form. She has offered to help me find pro bono legal assistance in order to get our 501c3 taken care of and she has also expressed an interest in mentoring us as we get our feet off the ground. I think that’s an incredible opportunity for us to take if we want to continue to grow into a larger group.

If you’d like the full summary of the phone conversations I had, or if you have any thoughts or concerns to share, I’d love to hear from you. Please email me at jvoss04@gmail.com.

Valerie Ragusa and I are going to be contacting Lauren to begin the process of moving forward. While we do this, please be thinking about whether or not you’re able to serve as a board member and see Valerie’s post on the Facebook page on information to submit to be considered for a seat.
Child Spotlight: Isaac

Isaac is 14 months old and lives outside of Houston, TX. At the age of 4 months, Isaac’s family noticed strange movements that turned out to be seizures. Genetic testing was done and Isaac was found to have the 17q12 duplication at the age of 6 months. Because of his seizures, Isaac slept a lot during his first 10 months and is behind in motor skills, feeding, and speech. Luckily, seizure activity has stopped and Isaac is now starting to move around a lot more! Despite his hypotonia, he’s rolling, low crawling, and even trying to get up on all fours. Isaac loves his brothers and will try to follow them and make noises to get their attention. He also loves playing with his toys. Great job, Isaac, and we hope you continue to stay seizure-free!

Research Update

To date, 39 families have consented to participate in the ongoing 17q12 research study through Geisinger’s Autism & Developmental Medicine Institute. This includes 11 families affected by 17q12 duplications, and 28 families affected by 17q12 deletions. Some families have completed all aspects of the study, including submitting genetic testing results for all family members, completing online surveys for all family members, and participating in direct assessments (cognitive testing, brain imaging, etc.). Many families have completed some, but not all, parts of the study and may be contacted in the future to provide additional information. Families who submit genetic testing results and complete all surveys are eligible for a $50 gift card. A big thank you to all participating families!

Childhood Anxiety

By Cora M. Taylor, Ph.D., Barbara Haas-Givler, BCBA, & Paul Kettlewell, Ph.D.

Anxiety disorders are a common concern for children and teens. While some level of anxiety can be normal in children, anxiety that is severe and persists should be treated. For example, children around one year of age commonly experience a period of “stranger anxiety” when they are reluctant to leave the care of a parent or guardian, and many children entering middle school or high school fear getting lost in the building or not being able to operate a locker.

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Several common strategies that children may be taught during CBT include:

**Relaxation and deep breathing exercises:** These strategies are practiced during non-anxious situations so that children can use them effectively during anxiety-provoking situations. Parents can help by modeling breathing and calming strategies at home, and practicing with their child.

**Cognitive Restructuring:** Older children and teens may be taught strategies to help them change their anxious thoughts. This part of therapy focuses on positive thinking and challenging the child’s anxious thoughts about a certain situation.

**Facing The Fear:** While it may be difficult, it is important to help children “face their fears” by supporting them through an anxious situation instead of letting the child continually avoid anxiety. Avoiding situations that lead to anxiety often adds to more anxiety in the future. After a child has been taught strategies to manage his or her anxiety, parents and therapists should provide opportunities for the child to practice gradually facing situations that make them anxious.

Despite the important of treating anxiety disorders, it can be difficult to assess in children/adolescents and may manifest as defiance, task refusal, attention seeking or hyperactivity. If you are worried that your child might be experiencing an anxiety disorder, talk to your pediatrician about resources in your area. The Cognitive Behavior Therapy approaches described above are a first-line treatment for anxiety, but some children also benefit from medication. A professional such as a developmental pediatrician or psychologist may be your best first source to help diagnose and identify strategies to address and treat anxiety.