17q12 Review

The 17q12 Interest Group is a partnership between families and researchers interested in 17q12 deletions and duplications

First 17q12 Family Meeting is a Success!

On August 8, 2014, genetics and medical specialists from Geisinger's Autism & Developmental Medicine Institute (ADMI) hosted the first-ever family meeting on 17q12 deletions and duplications. The meeting was held at the Renaissance Airport Hotel in Philadelphia, PA.

Eleven families from 9 states were attendance. Parents and caregivers listened to talks about a range of topics, including genetics, understanding diagnoses, common medical problems, kidney issues, research opportunities, management of developmental disorders, augmentative and alternative communication strategies, and behavioral interventions.

Many families brought their children, who enjoyed a day of games and movies in the childcare room next door to the presentation hall. In addition to educational talks and a Q & A session, families also had the opportunity to participate in research. Research studies included eye-tracking, measurements of balance and sway, and a

fine motor task. Both children and their parents were eligible to participate in these short research studies with the goal of better understanding the effects of 17q12 deletions and duplications.

A big thank you to all those families who traveled from near and far to participate!

Presentation Slides Available on Website

Slides from the 17q12 Family Meeting in Philadelphia are now available to download in PDF format on the 17q12 page of the ADMI website: www.geisingeradmi.org/17q12. Presentations include:

- Genetics of the 17q12 Region
- Overview of Clinical Symptoms of 17q12 Deletions & Duplications
- Renal Symptoms and Medical Follow-up
- Making Sense of Genetic, Educational, and Psychiatric Diagnoses
- 17q12 Research Opportunities
- Treatment of Children with Developmental Disorders
- Resources and Strategies for Children with Complex Communication Needs
- Understanding the Basics of Behavioral Intervention

In this issue:

Families Invited to Participate in Online Research

As part of the ongoing 17q12 research study at Geisinger, participating families are asked to complete a series of online surveys for each family member. These measures examine several different types of behaviors and experiences and will allow researchers to better describe and quantify the effects of 17q12 deletions and duplications.

In addition to the surveys, researchers are also requesting genetic testing results for all family members participating in the study. This allows them to determine with certainty who has a 17q12 genetic change and who does not.

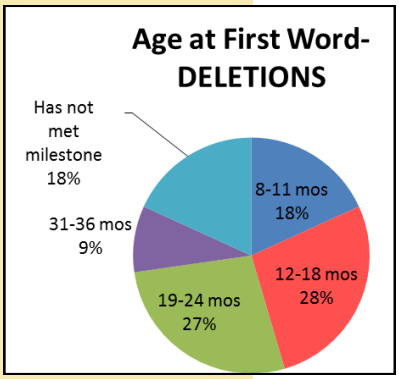
Geisinger ADMI researchers recognize that the surveys are lengthy and that genetic lab reports can be difficult to obtain, but these two parts of the study are crucial for our understanding of 17q12 deletions and duplications. As compensation for families' time and effort, $50 Amazon gift cards will be offered for completing the surveys and providing genetic testing results.

The hope is that the valuable knowledge gleaned from these surveys will help to improve future treatments and care for individuals affected by 17q12 deletions and duplications. Thank you to all the families who have already completed surveys!

If you are interested in participating, please email Marissa Mitchel (mwmitchel@geisinger.edu).

Insights from the Patient Crossroads 17q12 Registry

38 individuals have registered on the Patient Crossroads 17q12 interest group registry page (connect.patientcrossroads.org/?org=17q12), and a subset of these individuals have completed all or part of several surveys. Information from these surveys is based on self-report and so should be interpreted cautiously, as its accuracy has not been independently verified. Still, families may benefit from the information provided by other families. Some insights are summarized below.



Deletions

-58% report taking first steps by 18 months; 33% at 24 months, and 8% at 36 months

-92% report currently or previously receiving Occupational Therapy

-73% report currently or previously receiving Physical Therapy

-100% report currently or previously receiving Speech Therapy

-70% report anxiety disorder

-33% report short stature

-1 person reports heart defects

-1 person reports seizures

-33% report eye problems (e.g., strabismus, nystagmus, nearsighted)

-38% report cystic kidney; 50% report hydronephrosis; 33% report recurrent UTIs

-8% report constipation; 8% report diarrhea; 8% report GERD; 17% report feeding disorder

Duplications

-25% report taking first steps by 18 mos; 25% at 24 mos; 25% at 30 mos;

-75% report currently or previously receiving Occupational Therapy

-50% report currently or previously receiving Physical Therapy

-88% report currently or previously receiving Speech Therapy

-33% report anxiety disorder

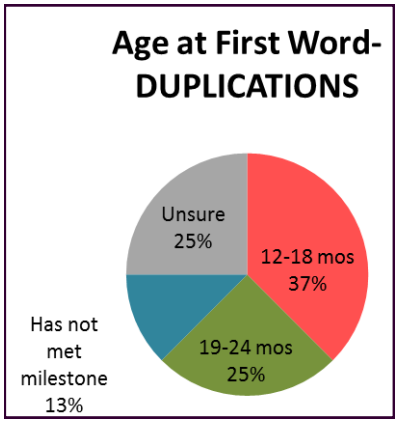
-40% report some type of disruptive behavior disorder

-25% report seizures

-29% report lacrimal (tear duct) blockage

-43% have feet turned in; 29% have flat feet

-1 reports kidney reflux; 33% report recurrent UTIs



-gastrointestinal and feeding problems are common (43% constipation, 29% dysphagia, 29% feeding disorder, 29% GERD)

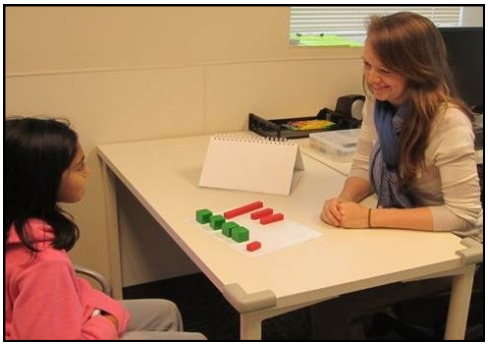
Plan Your Visit to the Autism & Developmental Medicine Institute

Local families and those who are willing to travel to ADMI in Lewisburg, PA may be eligible to participate in face-to-face assessments as part of the ongoing 17q12 research study at Geisinger.

This part of the research study involves several short assessments. These include eye-tracking, which involves looking at some pictures on a computer screen, as well as a fine motor task and a measure of balance. In addition, some families may be eligible to participate in cognitive assessments and fMRI (brain scans).

These measures may seem very different from each other, but each one will help researchers develop a more complete picture of your family and could provide valuable information about the effects of 17q12 deletions and duplications in affected participants in comparison to their unaffected family members.

Financial assistance may be available to offset travel costs for families traveling from a distance. For more information about the study, please contact Missy Slane (mmslane@geisinger.edu).



Professionals Team Up to Draft Medical Guidelines

"One of the major roadblocks to effective treatment and preventative care is a lack of information."

One of the major roadblocks to effective treatment and preventative care to individuals with 17q12 deletions and duplications is a lack of information available to primary care physicians and specialists about these genetic changes.

At the 17q12 family meeting in August, parents and caregivers voiced their concerns and suggested that it would be helpful to have a set of medical and treatment guidelines that could be shared easily with all those who care for their family members with a 17q12 deletion/duplication. Researchers and clinicians at Geisinger's ADMI are spearheading this effort and reaching out to a variety of specialists who are knowledgeable about the medical characteristics known to be associated with 17q12 deletions and duplications. The ultimate goal is to develop a peer-reviewed article, which will include a summary of what is known about the condition and provide information about disease characteristics, diagnosis/testing, medical management and genetic counseling considerations. This article could, in turn, be shared with the affected individual's doctors and medical specialists in order to provide a framework for the ongoing care of their patients.

Facebook Support Group Attracts Close to 200 Followers

The Chromosome 17q12 Support Group on Facebook currently has 181 members. It is a closed group, meaning you must request permission to become a member and view content, which is a safeguard to protect families' privacy.

Individuals with 17q12 deletions/ duplications, parents, grandparents, and other caregivers are welcome to join. Families often share information with each other, such as what to expect from doctor's visits, health issues experienced by those with 17q12 changes, and what types of services and treatments have been most beneficial. Valerie Ragusa, the parent of a child with a 17q12 deletion, is the administrator of the group.

Dear Families,

Thank you all for supporting our research and family outreach efforts. I hope you enjoy this first-ever 17q12 newsletter!

Marissa Mitchel
Clinical Research SLP,
Geisinger ADMI

Visit us on the web: GeisingerADMI.org/17q12

SAVE THE DATE! Second 17q12 Family Meeting in the Works

In light of the success of the first family meeting, the 17q12 Interest Group is planning a second 17q12 family meeting.

Dates: **Friday June 26 and Saturday June 27**

Location: Philadelphia

Agenda:

- Educational presentations on a range of topics
- Progress updates
- Consults with genetics professionals
- Research opportunities
- Family social event– stay tuned for more details!

Input and ideas from families are welcome!

Please contact Marissa Mitchel at mwmitchel@geisinger.edu