17q12 Research Opportunity

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*Geisinger Health System*
*Lewisburg, Pennsylvania*
Goals

• To learn more about individuals with 17q12 deletions and duplications and their families.

• To better understand the developmental, behavioral, and medical features found in individuals with a 17q12 deletion or duplication and their family members.

• To improve medical and behavioral interventions and improve outcomes for individuals who have been diagnosed.
Update

- 23 families currently consented for research participation
  - 64 participants in total

Questionnaires completed by **48 participants** in total

In-person research testing completed by **26 participants** in total

**13 participants** have questionnaires *and* in-person testing complete
What is involved?

We will ask you to complete several different types of assessments:

a. A packet of surveys that will be provided to you

b. In-person assessments completed here at the conference
   • Direct Testing
   • Eye-Tracking
   • Motor Assessment

c. For those who are local to our center, we may follow-up regarding in-person assessments and other research testing at our clinic in Lewisburg, PA
Survey Packets

• 3 categories of questionnaires to review:
  • Questionnaires to be completed on your children
  • ….on yourselves as parents or caregivers
  • ….for children 18 year of age or older.
### Survey Packets

- Questionnaires for you to complete on your children

<table>
<thead>
<tr>
<th>Survey Type</th>
<th>Age Range</th>
</tr>
</thead>
<tbody>
<tr>
<td>Developmental Coordination DQ (DCDQ)</td>
<td>5-15 years of age</td>
</tr>
<tr>
<td>Children’s Communication Checklist- 2\textsuperscript{nd} edition (CCC-2)</td>
<td>4-16 years of age</td>
</tr>
<tr>
<td>Social Responsiveness Scale 2 (SRS 2)*</td>
<td>2.5-5 years of age</td>
</tr>
<tr>
<td></td>
<td>4-18 years of age</td>
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<tr>
<td></td>
<td>18+ years of age</td>
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<tr>
<td>Geisinger-Oxford-Liverpool Inventory of Feelings and Experiences- Child Version (GO-LIFE-C)</td>
<td></td>
</tr>
<tr>
<td>Child Behavior Checklist (CBCL)*</td>
<td>1.5-5 years of age</td>
</tr>
<tr>
<td></td>
<td>6-18 years of age</td>
</tr>
<tr>
<td>Childhood Routine Inventory- Revised (CRI-R)</td>
<td></td>
</tr>
<tr>
<td>Adaptive Behavior Assessment System 2 (ABAS 2)*</td>
<td>0-5 years of age</td>
</tr>
<tr>
<td></td>
<td>5-21 years of age</td>
</tr>
</tbody>
</table>

*Note:"

(5+ yo)  
(1-5 yo) 
(< 1 yo)
Survey Packets

- Questionnaires for you to complete *on yourselves*

<table>
<thead>
<tr>
<th>Survey Packet</th>
</tr>
</thead>
<tbody>
<tr>
<td>Adult Routines Inventory (ARI)</td>
</tr>
<tr>
<td>Geisinger-Oxford-Liverpool Inventory of Feelings and Experiences (GO-LIFE)</td>
</tr>
<tr>
<td>Social Responsiveness Scale- Adult Self Report 2 (SRS 2)</td>
</tr>
<tr>
<td>Adult Behavior Checklist (ABCL)</td>
</tr>
<tr>
<td>Adaptive Behavior Assessment System 2 (ABAS 2)</td>
</tr>
</tbody>
</table>
Survey Packets

**If your child is 18 years of age or older and is willing and able to self-report**

- Adult Routines Inventory (ARI)
- Geisinger-Oxford-Liverpool Inventory of Feelings and Experiences (GO-LIFE)
- Social Responsiveness Scale 2- Adult Self Report (SRS 2)
- Adult Behavior Checklist (ABCL)
- Adaptive Behavior Assessment System 2 (ABAS 2)
In-Person Assessments at the Conference

We are holding research sessions at this conference.

3 tasks:
1. Direct Testing
2. Eye tracking
3. Motor Assessment
   • Purdue Pegboard
   • Balance Test
Direct Testing

We will ask you and your family members to sit with our study staff to complete some measures face-to-face. This will include playing some games and answering some questions.
Eye Tracking

You and your family will be asked to sit in front of an eye tracker and to look at some pictures on a screen.

Your eye movements will be recorded, but the machine will not record your face

Your family will be asked to watch some pictures on the computer screen.

There are no instructions other than to look at the pictures as they are presented to you.

We will also record your eye movements using eye tracking.
Purdue Pegboard

You and your family will be asked to complete several motor tasks that involve placing pegs into holes.

You will be given instructions about how to place the pegs.

http://www.oandp.org/olc/course_extended_content.asp?frmCourseId=BC8264FC-BB7-437D-85FF-004A53C34723&frmTermId=purdue
Balance and Sway

You and your family will be asked to stand on a force plate (similar to a Wii fit board) and perform several tasks:

1. Eyes open, arms at side
2. Eyes closed, arms at side
3. Eyes open, arms out
4. Eyes closed, arms out

http://www.turbosquid.com/3d-models/man-silhouette-3d-model/640497
In-Person Assessments at ADMI

We are also inviting families who live within traveling distance of our center in Lewisburg, PA to come in and complete some additional assessments and participate in additional research opportunities.
Functional Magnetic Resonance Imaging (fMRI)

An MRI is a large magnet that is used to take 3D pictures of the brain.

You and your family will be asked to lay as still as possible while the machine takes pictures of your brains. The machine is not harmful and there is no radiation used.

The machine is a little noisy, but we will provide ear plugs!
Diffusion Tensor Imaging (DTI)

Functional MRI

Structural MRI
Our research would not be possible without the generous donation of our families’ time and effort!
Questions?