



17q12 Research Opportunity

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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:

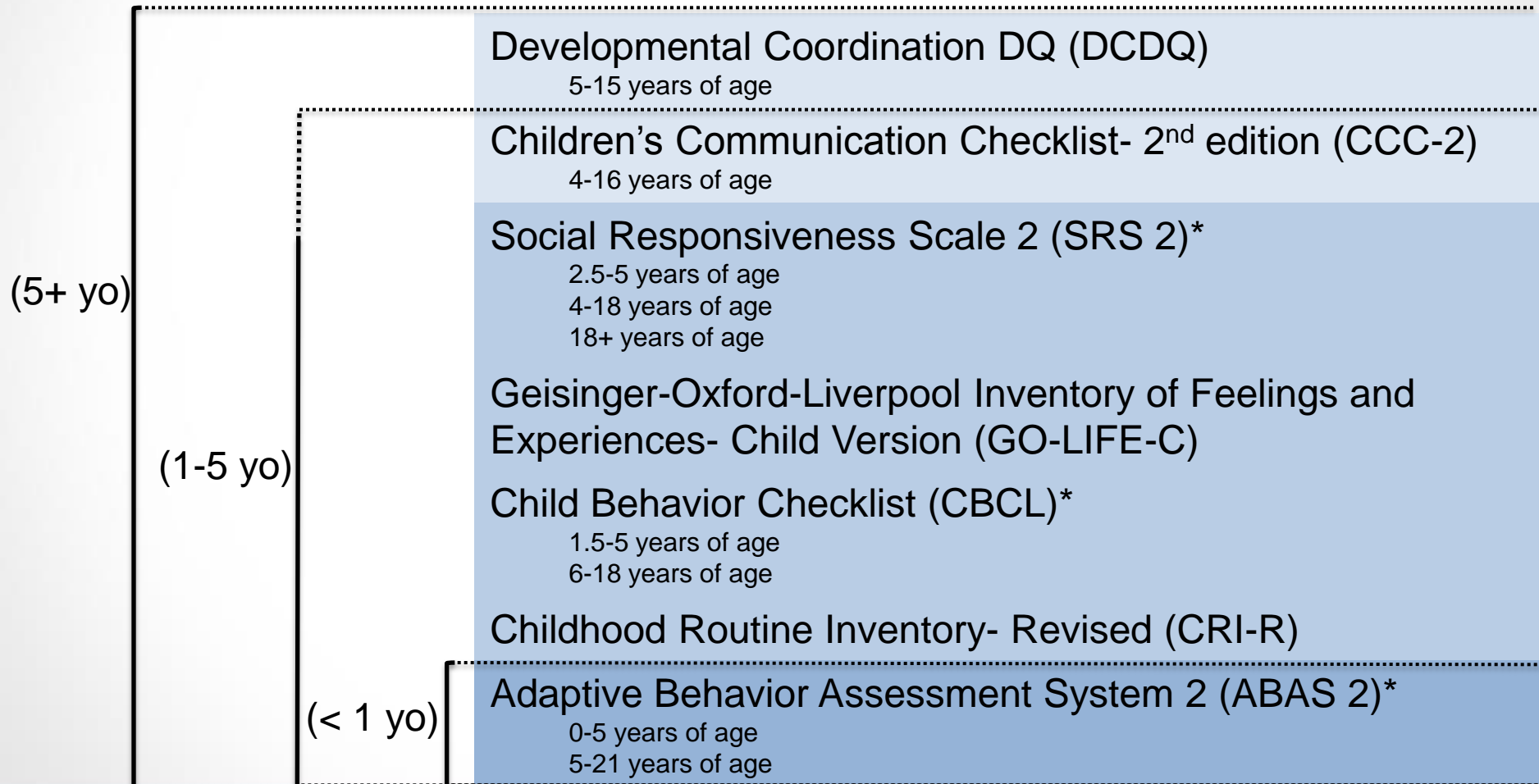
- a. A packet of surveys that will be provided to you
- b. In-person assessments completed here at the conference
 - Direct Testing
 - Eye-Tracking
 - Motor Assessment
- c. For those who are local to our center, we may follow-up regarding in-person assessments and other research testing at our clinic 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



Survey Packets

- Questionnaires for you to complete *on yourselves*

Adult Routines Inventory (ARI)

Geisinger-Oxford-Liverpool Inventory of Feelings and Experiences (GO-LIFE)

Social Responsiveness Scale- Adult Self Report 2 (SRS 2)

Adult Behavior Checklist (ABCL)

Adaptive Behavior Assessment System 2 (ABAS 2)

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)

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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

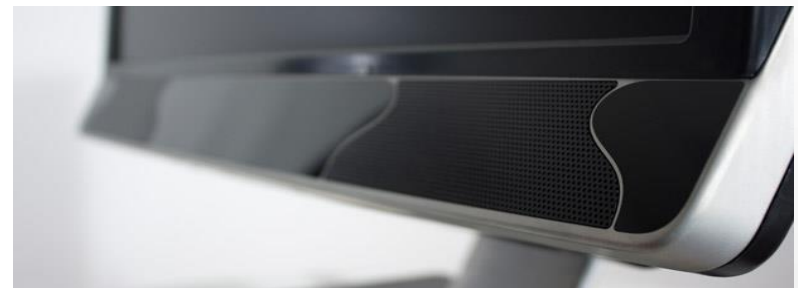


You and your family will be asked to sit in front of an eye tracker and to look at some pictures on a screen.

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



<http://www.tobii.com/en/eye-tracking-research/global/products/hardware/tobii-t60xl-eye-tracker/>



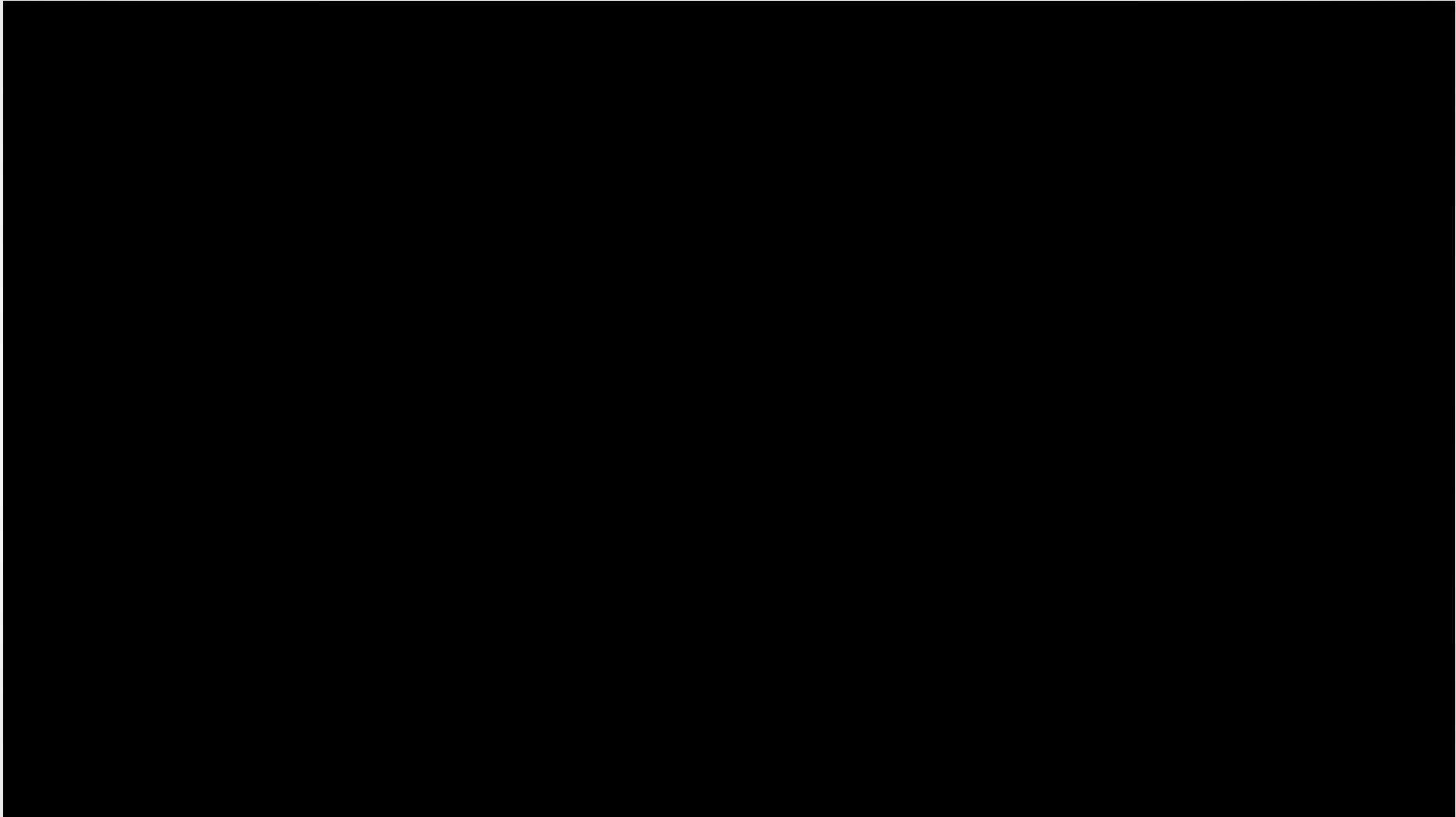
<http://www.tobii.com/en/eye-tracking-research/global/products/hardware/tobii-t60xl-eye-tracker/>

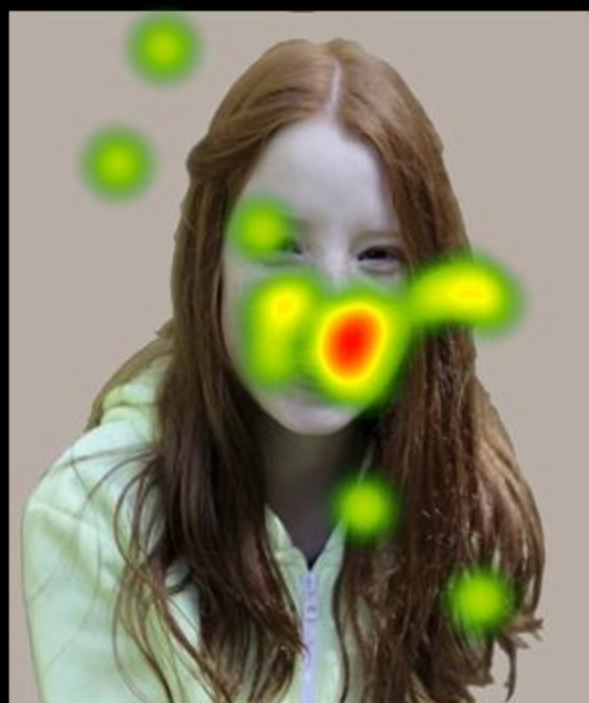
Your family will be asked to watch some pictures on the computer screen.



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

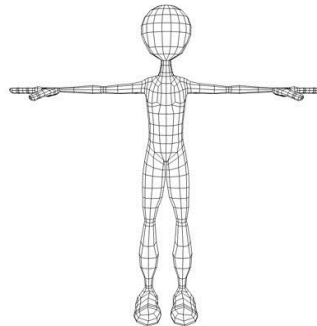


http://www.oandp.org/olc/course_extended_content.asp?frmCourseId=BCB264FC-8BB7-437D-85FF-004A53C34723&frmTermId=purdue

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>



<http://www.amti.biz/fps-guide.aspx>

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.

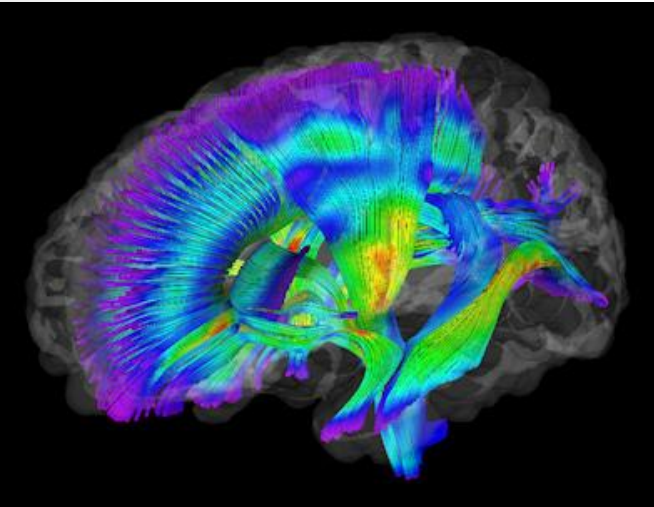
You and your family will be asked to lay as still as possible while the machine takes pictures of your brains. The machine is not harmful and there is no radiation used.

The machine is a little noisy, but we will provide ear plugs!



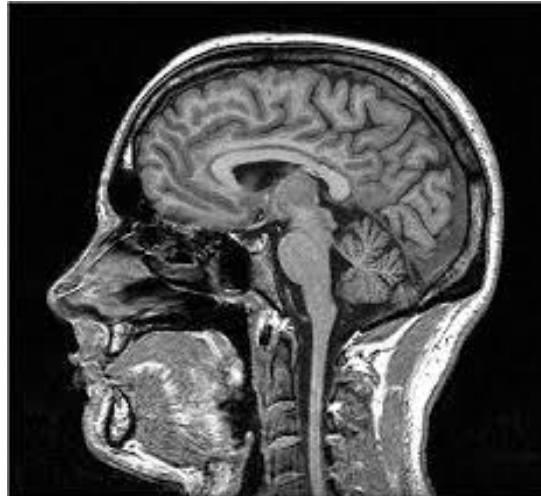


Diffusion Tensor Imaging (DTI)



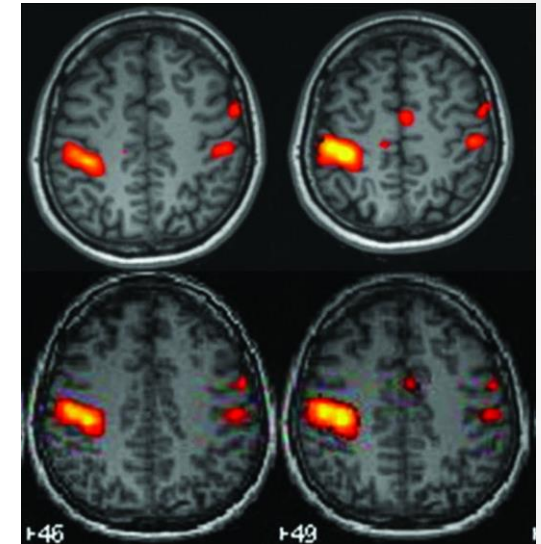
<http://www.auntminnie.com/index.aspx?sec=ser&sub=def&pag=dis&ItemID=98363>

Structural MRI



<http://www.mylifewithabrokenbrain.co.za/if-the-brain-breaks/>

Functional MRI



<http://www.ajnr.org/content/34/6/1188/F1.expansion.html>

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Autism & Developmental Medicine Institute

A whole new perspective on the evaluation & treatment of children with special developmental needs



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Care & Innovation

Advanced research for better results



Patients & Caregivers

Answers to your questions



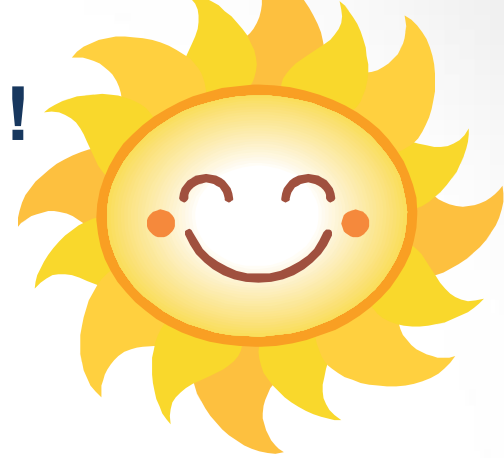
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HEALTH SYSTEM

THANK YOU!!



Our research would not be possible without
the generous donation of our families' time
and effort!

Questions?