

17q12 Research Opportunity

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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

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What is involved?

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

- A group of surveys that will be sent to you online using our survey tool, Qualtrics®
- In-person assessments completed here at the conference
- For those who are local to our center, in-person assessments at our clinic in Lewisburg, PA

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Online Assessments

- We will email you links to several surveys to complete online
- The surveys are private and we will send you a password in a separate email
- The surveys will save automatically while you are completing them; you can close your browser and continue working at a later time
- We are asking that you complete the survey battery for each member of your family

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Survey for:

Welcome to the Geisinger 17q12 Interest Group!



What are chromosome 17q12 deletions and duplications?

Our chromosomes contain genetic material that tells our bodies how to grow and develop. They come in pairs, one set inherited from each parent. When a person is missing genetic material from their chromosomes, it is called a "deletion." If a person has extra genetic material on their chromosomes, it is called a "duplication." People with deletions or duplications may have an increased chance for health or developmental concerns, depending on the material that is missing or extra. People with missing or extra information on chromosome 17q12 are at increased risk for neurodevelopmental disorders (such as autism or learning problems) and medical issues (such as kidney problems).

What is the purpose of this study?

The purpose of this study is to identify and better understand the developmental, behavioral, and medical features found in individuals with a 17q12 deletion or duplication and their family members. Our hope is that such knowledge will drive medical and behavioral interventions and improve outcomes for individuals who have been diagnosed.

Who is conducting this study?

We are a group of researchers and clinical care providers from Geisinger Health System's Autism & Developmental Medicine Institute (ADMI), a center of clinical care, research, and education focusing on children with neurodevelopmental disorders.

Click the arrow in the bottom right corner to begin!

www.17q12.org



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GEISINGER IRB NUMBER: 2013-0438
IRB Approved: 06/26/2014

The study staff will explain this study to you. Ask questions about anything that is not clear at any time. You may take home an unsigned copy of this consent form to think about and discuss with family or friends.

- Being in a study is voluntary – your choice.
- If you join this study, you can still stop at any time.
- No one can promise that a study will help you.
- Do not join this study unless all of your questions are answered.

After reading and discussing the information in this consent form you should know:

- Why this research study is being done;
- What will happen during the study;
- Any possible benefits to you;
- The possible risks to you;
- Other options you could choose instead of being in this study;
- How your personal health information will be treated during the study and after the study is over;
- Whether being in this study could involve any cost to you; and
- What to do if you have problems or questions about this study.

Please read this consent form carefully.

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IRB NUMBER: 2012-0430
IRB Approval: 06/26/2014

CONSENT FORM
Review the online consent form, and choose the appropriate consent option.

ADULT PARTICIPANT CONSENT
By checking the appropriate boxes next to the six statements below, you certify that you have read the consent, certified with the information provided and agree to be in the study. By certifying that you have read the consent form, you will not give up any legal rights. You will be able to review, download and/or print a copy of the consent form upon completion.

I have read and understand the informed consent.
 I understand that I do not have to take part in this study unless I want to.
 I understand that I do not have to answer all of the questions.
 I understand that the information I give will be kept securely, and will only be seen by the researchers.
 I agree to take part in this study.

You must agree to all items listed in the checklist in order to proceed with the study. If you have questions about the study or would like to speak with a study member, please call the underlined number listed below.

WHOM DO I CALL IF I HAVE QUESTIONS OR PROBLEMS?
For questions about the research study, contact the study doctor (David L. LEBERTZ, MD # 370.226.2200).
For questions about your rights as a research participant, contact the Human Research Protection Program staff of the Geisinger Institutional Review Board (which is a group of people who review the research to protect your rights) at (717) 271-0865.

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Sorry, you cannot continue until you correct the following:
* Select the "I must check all statements in order to proceed with the study."

IRB NUMBER: 2012-0430
IRB Approval: 06/26/2014

CONSENT FORM
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ADULT PARTICIPANT CONSENT
By checking the appropriate boxes next to the six statements below, you certify that you have read the consent, certified with the information provided and agree to be in the study. By certifying that you have read the consent form, you will not give up any legal rights. You will be able to review, download and/or print a copy of the consent form upon completion.

You must check all statements in order to proceed with the study.

I have read and understand the informed consent.
 I understand that I do not have to take part in this study unless I want to.
 I understand that I do not have to answer all of the questions.
 I understand that the information I give will be kept securely, and will only be seen by the researchers.
 I agree to take part in this study.

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I agree to allow a member of the research team to contact me in the future as needed to gather updated health information.

I agree
 I do not agree

Please type your name below to indicate that you have reviewed the informed consent and agree to take part in this research study.

First Name	Middle Initial	Last Name	Date (mm/dd/yyyy)
<input type="text"/>	<input type="text"/>	<input type="text"/>	<input type="text"/>

Survey Completion: 0% / 100%

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IRB Approval: 06/26/2014

CONSENT FOR CHILDREN
If you are the parent or legal guardian of a child who is being asked to participate, the term "you" used in this consent refers to both you and your child.
By checking the appropriate boxes next to the six statements below, you certify that you have read the consent form certified with the information provided and agree to be in the study. By certifying that you have read the consent form, you will not give up any legal rights. You will be able to review, download and/or print a copy of the consent form upon completion.

I have read and understand the informed consent.
 I understand that I do not have to take part in this study unless I want to.
 I understand that I do not have to answer all of the questions.
 I understand that the information I give will be kept securely, and will only be seen by the researchers.
 I agree to take part in this study.
 I agree to provide information about my child for this study.
 I agree to allow my child/dependent to take part in this study.

You must agree to all items listed in the checklist in order to proceed with the study. If you have questions about the study or would like to speak with a study member, please call the underlined number listed below.

WHOM DO I CALL IF I HAVE QUESTIONS OR PROBLEMS?
For questions about the research study, contact the study doctor (David L. LEBERTZ, MD # 370.226.2200).
For questions about your rights as a research participant, contact the Human Research Protection Program staff of the Geisinger Institutional Review Board (which is a group of people who review the research to protect your rights) at (717) 271-0865.

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Please type each of your participating children's names and dates of birth below to indicate that you agree to allow your child/dependent to take part in this research study.

	Name	Date of Birth
Child 1	<input type="text"/>	<input type="text"/>
Child 2	<input type="text"/>	<input type="text"/>
Child 3	<input type="text"/>	<input type="text"/>
Child 4	<input type="text"/>	<input type="text"/>
Child 5	<input type="text"/>	<input type="text"/>
Child 6	<input type="text"/>	<input type="text"/>
Child 7	<input type="text"/>	<input type="text"/>
Child 8	<input type="text"/>	<input type="text"/>
Child 9	<input type="text"/>	<input type="text"/>
Child 10	<input type="text"/>	<input type="text"/>

Parent/Guardian Typed Name:

First Name	Middle Initial	Last Name	Date (mm/dd/yyyy)
<input type="text"/>	<input type="text"/>	<input type="text"/>	<input type="text"/>

Survey Completion: 0% / 100%

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Please click the link below to save a copy of the informed consent form for your records:

[17a12_Study_Informed_Consent_Form](#)

Survey Completion: 0% / 100%

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Parent/ Guardian Information:

What is your full name?

First

Middle Initial

Last

What is your gender?

What is your age (in years)?

Today's Date (mm/dd/yyyy):

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Social Responsiveness Scale 2 (SRS 2)

Survey for: John T. Doe

Social Responsiveness Scale 2 (SRS2) Parent Report- School Age

Today's Date (mm/dd/yyyy):

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Survey Completion 100%

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Social Responsiveness Scale 2 (SRS 2)

Directions:
For each question, please check the box that best describes the child's behavior over the past 6 months.

Seems much more fidgety in social situations than when alone.

Not True Sometimes True Often True Almost Always True

Expressions on his or her face don't match what he or she is saying.

Not True Sometimes True Often True Almost Always True

Seems self-confident when interacting with others.

Not True Sometimes True Often True Almost Always True

When under stress, he or she shows rigid or inflexible patterns of behavior that seem odd.

Not True Sometimes True Often True Almost Always True

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Not True Sometimes True Often True Almost Always True

There are 20 unanswered questions on this page

Would you like to continue?

Answer the Questions Continue Without Answering

Shows unusual sensory interests (for example, mouthing or spinning objects) or strange ways of playing with toys.

Not True Sometimes True Often True Almost Always True

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Adaptive Behavior Assessment System 2 (ABAS 2)

Communication

	Is Never Sometimes Always	Check if you Guessed	Comments
	Not When Able Needed When When Needed When Needed		
Looks at others' faces when they are talking.	<input type="radio"/> <input type="radio"/> <input type="radio"/> <input type="radio"/>	<input type="checkbox"/>	<input type="text"/>
Laughs when a parent or other person laughs.	<input type="radio"/> <input type="radio"/> <input type="radio"/> <input type="radio"/>	<input type="checkbox"/>	<input type="text"/>
Raises and lowers voice to express different feelings or needs.	<input type="radio"/> <input type="radio"/> <input type="radio"/> <input type="radio"/>	<input type="checkbox"/>	<input type="text"/>

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This is the end of the survey. Once you click the next arrow, you will complete the survey, your responses will be submitted, and you will not be able to change your answers. Please take some time to review your responses to ensure that you have answered everything completely and accurately.

Survey Completion 100%

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Survey for: John T. Doe

We thank you for your time spent taking this survey.
Your response has been recorded.

Survey Complete 100%

Survey Powered By [Qualtrics](#)

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In-Person Assessments at the Conference

Will be holding research sessions this evening at the conclusion of the conference


3 tasks:

1. Eye tracking
2. Balance and sway
3. Purdue Pegboard

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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

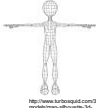

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Balance and Sway

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


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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



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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

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