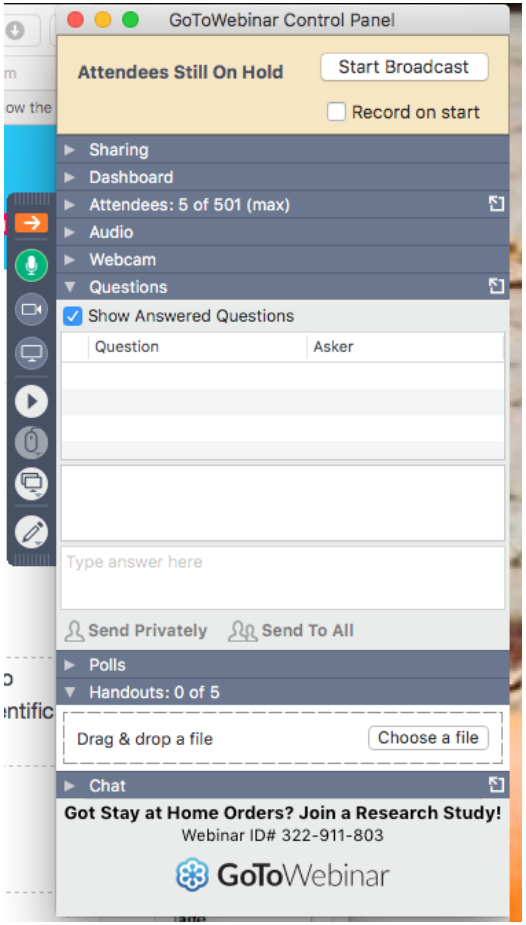
RESEARCH FAIR FOR FAMILIES

MAY 28, 2020

AUTISM SCIENCE FOUNDATION

Guidelines for today:

- We will be going in order of age of person with ASD or family member (one COVID related not specific to ASD at the end)
- They will all be listed on our ”Participate in Research” page
- Tell your friends!
- Questions in the question box



ahalladay@autismsciencefoundation.org

AUTISM SCIENCE FOUNDATION

Perceptions of unusual behaviors in children

Zoe Sargent, University of Virginia

- Young children understand unwritten rules for behavior
- Autism is characterized by unusual behaviors

What do kids think about unusual behaviors?

What do kids think about unusual behaviors?

- **WHO**: Any child 4-7 years old
- **WHAT**:
  - 6 short stories with characters behaving in different ways
  - Ask about behaviors and characters
- **WHEN**: 15-30 minutes, scheduled Zoom meeting
- **WHY**:
  - No direct benefits to families
  - But, it may help us learn more about how children understand behavior

What do kids think about unusual behaviors?

- **HOW**: Contact zsargent@virginia.edu for more details or to set up a time to participate!
  - Please include child's name, age (in years), and some times you would be available to meet

Food and Eating Behaviors

Anita Aalia Panjwani, Purdue University

PURDUE UNIVERSITY

Food and Eating Behaviors in Children with ASD in Response to COVID-19

Purdue Autism Research Center

Anita Panjwani, PhD

Purdue Autism Research Center

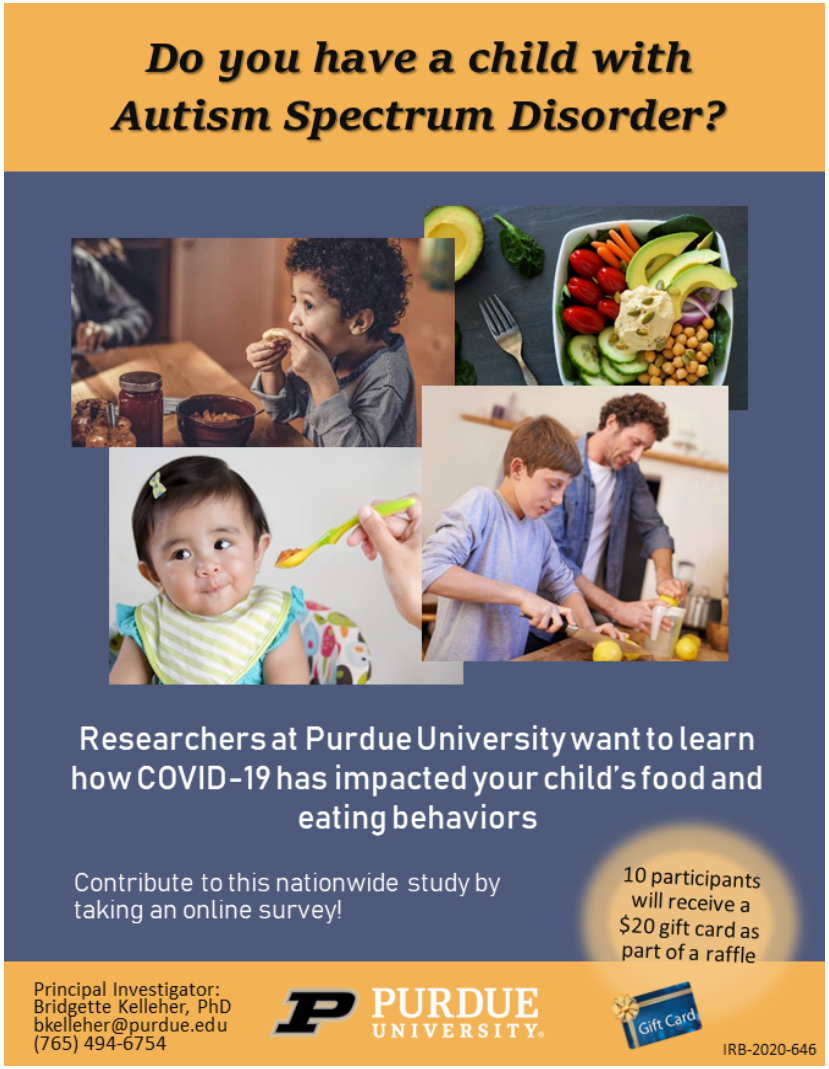
PURDUE UNIVERSITY

- Purpose: to understand how children with ASD are responding to the coronavirus (COVID-19) outbreak as it relates to food and eating behaviors. This information will help us understand how to tailor support strategies to address the dietary needs of children with ASD during high-stress periods of time.

- By participating in this study, your family will be contributing to the body of knowledge about how large-scale crises affect the food environment and dietary needs of children with ASD. There is no personal benefit guaranteed by participating in this study.

- Eligibility: parent of a child under 18 years of age with a diagnosed Autism Spectrum Disorder (ASD) living in the U.S.

PURDUE UNIVERSITY



- One-time 15-20 minute anonymous online survey
- Chance to win one of ten $20 Amazon gift cards
- URL for online survey: https://tinyurl.com/y8q572vh
- Find us at our Facebook page: https://www.facebook.com/purdueautism/

Contacts:

Dr. Anita Panjwani
apanjwan@purdue.edu

PI: Bridgette Kelleher
bkelleher@purdue.edu

Parent preferences about technology based interventions for children and young people with autism spectrum disorders: The PATH study

Nancy Kouroupa

University of Hertfordshire UH

The PATH study

Our goal

- Explore parents' attitudes towards use of a range of different types of technologies to support interventions for young people with ASD.
  - What is parents' preferred technology device?
  - What are parents concerns about the use of technology for intervention with children with ASD?
  - What sources of information influence parental decisions towards technology based intervention?

The PATH study

We need

- Parents/carers of children with autism
- The child with autism (suspected/diagnosed) aged 0-18 years old
- Understanding of English

What does the study involve?

- Anonymous online questionnaire
- 10-20 minutes
- No more involvement

We are looking for parents of children with autism aged 0-18 years to participate in research.

WHAT ARE PARENT'S PREFERENCES ABOUT TECHNOLOGY BASED SUPPORT WHEN IT COMES TO CHOOSING THEIR CHILD'S SESSION?

There is limited research about the use of technology in autism research. Working together while learning about parent's preferences we could change this!

We are researchers at the University of Hertfordshire. We would like to invite parents/carers to complete a questionnaire and share your views about using phones, tablets, virtual reality goggles and robots with children and young people on the autism spectrum. Your child doesn't need to have used any of these technologies for you to take part in this research.

Here is the link to access a survey that lasts for about 15-20 minutes (https://herts.eu.qualtrics.com/jfe/form/SV_6s6AJo88ZoYkvch)

For more information, contact: Nancy Kouroupa - ak18adj@herts.ac.uk

University of Hertfordshire

Ethics number: LMS/PGR/UH/04164

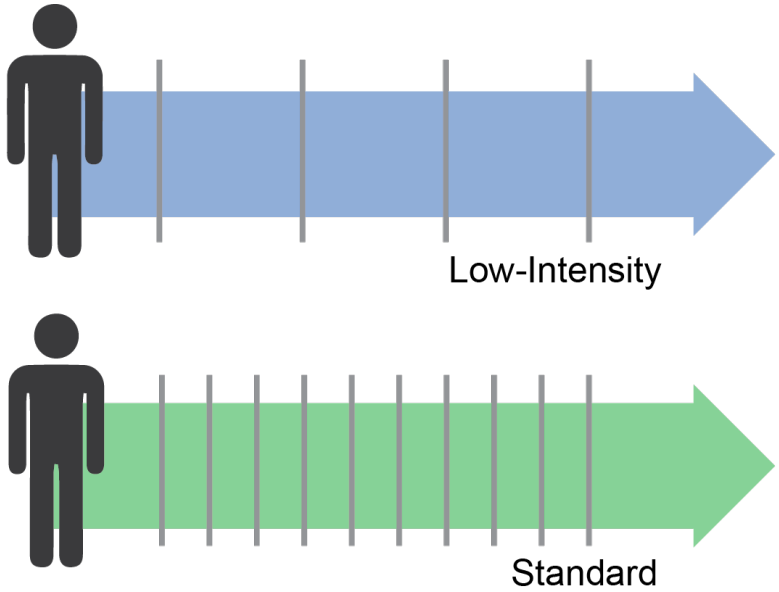
https://herts.eu.qualtrics.com/jfe/form/SV_6s6AJo88ZoYkvch

Telehealth for anxiety

Lynn Hana and Eric Storch, Baylor College of Medicine

Parent-Led Cognitive Behavioral Teletherapy for Children with Anxiety and ASD

- FREE treatment for anxiety designed to fit the needs of children with autism spectrum disorder (**ages 7-13 years old**)
- Sessions delivered via video to your home
- Families will be in one of two groups; low-intensity or standard
- Compensation for assessments completed (total $120)



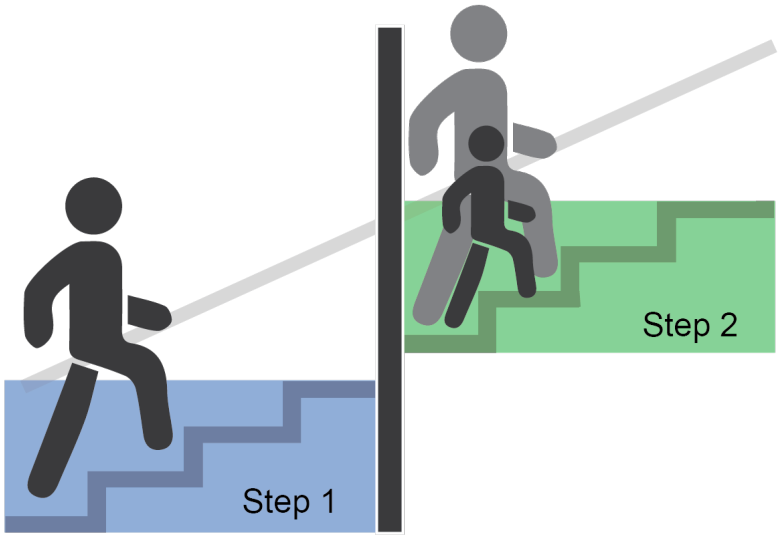
Texas Children's Hospital

*Residents of Texas

Baylor College of Medicine

Parent-Led, Stepped-Care CBT for Children with Anxiety and ASD

- FREE treatment for anxiety designed to fit the needs of children with autism spectrum disorder (**ages 4-14 years old**)
- Treatment will be provided in "steps" that vary by intensity beginning with Step 1
- If needed, families will “step-up” to receive a higher intensity in Step 2
- Compensation for assessments completed (total $160)



Texas Children's Hospital

*Residents of Texas

Baylor College of Medicine

Baylor College of Medicine

Texas Children's Hospital®

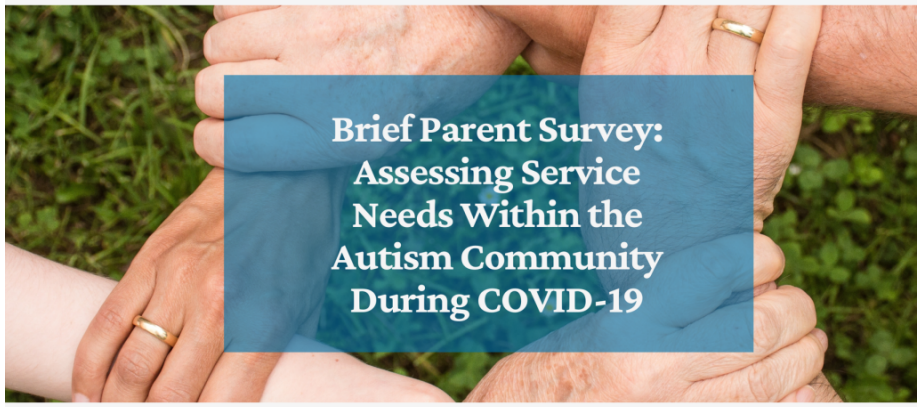
For more information, please email **anxietyandASD@bcm.edu**

Study staff will schedule a brief screening over the phone to determine initial eligibility.

Service needs during COVID

Emily Ferguson, UCSB

AN ONLINE PARENT/CAREGIVER SURVEY



UCSB Koegel Autism Center

Who?

Parents/ primary caregivers of an individual (aged 2 or older) diagnosed with autism spectrum disorder can participate in our study by completing this brief survey.

Why?

Your answers will provide helpful information to autism researchers and service providers to better support you and your family during the COVID-19 crisis and its aftermath.

A comprehensive list of nationwide resources will be offered alongside the survey.

To participate and gain access to our resource page, please follow this link: COVID-19 Experiences Survey

For more information, please email us at autism@ucsb.edu

We hope to better understand the specific needs and experiences of families nationwide with autistic children (aged 2 or older), adolescents, or adults during COVID-19. If we have a better understanding of your family's needs, we can develop better supports for the autism community.

Nuestra meta es comprender mejor las necesidades y experiencias específicas de las familias en todo el país con niños autistas (de 2 años o más), adolescentes o adultos durante el COVID-19. Al comprender mejor las necesidades de su familia, podremos desarrollar mejores apoyos para la comunidad autista.

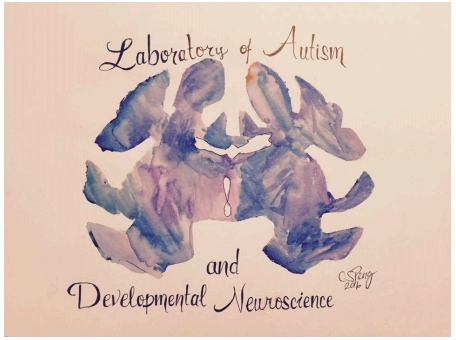
UC SANTA BARBARA

ASSESSING SERVICE NEEDS WITHIN THE AUTISM COMMUNITY DURING COVID-19

Brief Parent/ Caregiver Survey (15-20 minutes)

- Thank you for your consideration! Sharing your family's experience will help us develop better supports for the autism community during COVID-19 and its aftermath.
- Contact us: mariajimenezmunoz@ucsb.edu for assistance in English or Spanish.
- https://ucsb.co1.qualtrics.com/jfe/form/SV_06Qui2j7mDT1WqF

THE GEORGE WASHINGTON UNIVERSITY
WASHINGTON, DC



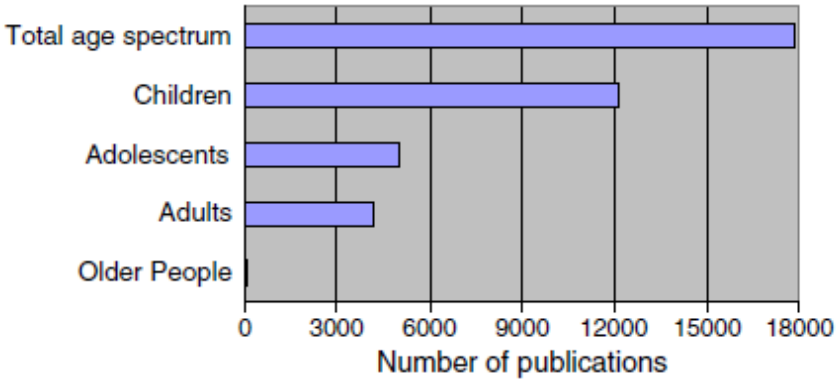
Adult Development and Aging in Autism

Greg Wallace, PhD

Associate Professor
Speech, Language, and Hearing Sciences
The George Washington University
https://blogs.gwu.edu/autismlab/

Aging and Autism

- The first individuals diagnosed with autism in the 1940s are only now reaching old age meaning that opportunities for studying aging in autism to date have been limited
- Thus, the knowledge base on aging in autism is woefully small
- Particularly important since we now have CDC-based estimates of autism rates among adults: ~2.2% or ~5,400,000 adults in the USA
- We want to hear about research priorities from central stakeholders, adults with autism



Mukaetova-Ladinska et al. (2012) *Int J Geriatr Psychiatry*

Participate in Research On Adult Development and Aging in Autism

Who can participate?

- Adults on the autism spectrum

What do you do?

- Complete an open-ended interview over the phone or a web-based calling system (e.g., Zoom or WebEx) and answer some basic demographic questions

How long does it take?

- 30-60 minutes, scheduled at a mutually convenient time.

Is there payment?

- No, this is not a paid study.

If interested, contact:

Colin Weiss or Alex Job Said at:

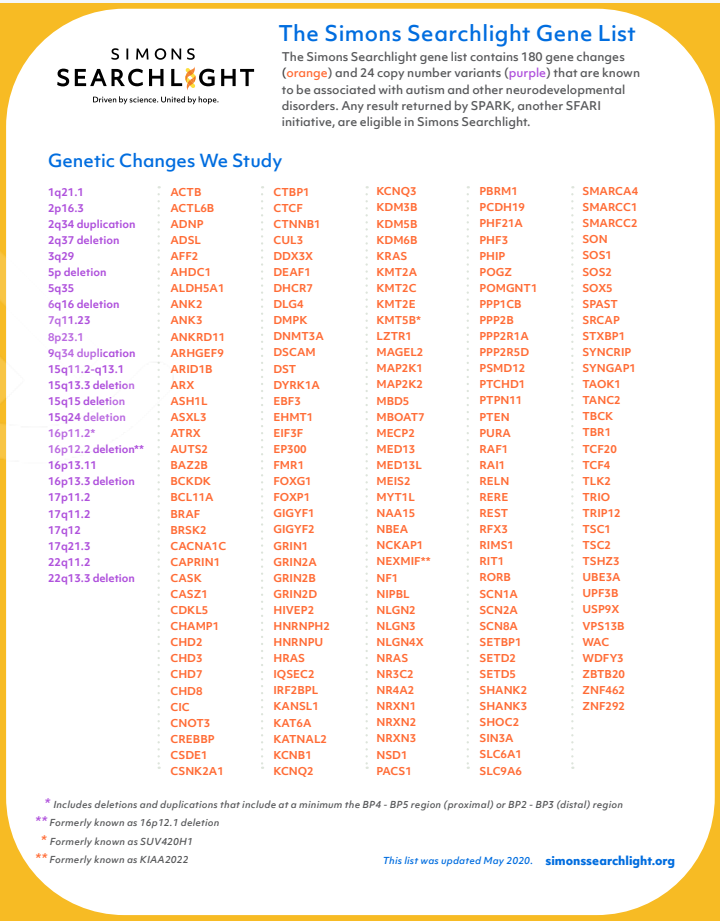
ladn@gwu.edu

Simons Searchlight

Misia Kowanda, Simons Foundation

What is Simons Searchlight?

Simons Searchlight is an **international research registry** for individuals who have a diagnosis with a **genetic change** associated with ASD or other neurodevelopmental conditions. The list of eligible genes is the same as **SPARK**, our affiliate study at the Simons Foundation Autism Research Initiative (SFARI).



SIMONS SEARCHLIGHT
Driven by science. United by hope.

The Simons Searchlight Gene List

The Simons Searchlight gene list contains 180 gene changes (orange) and 24 copy number variants (purple) that are known to be associated with autism and other neurodevelopmental disorders. Any result returned by SPARK, another SFARI initiative, are eligible in Simons Searchlight.

Genetic Changes We Study

1q21.1	ACTB	CTBP1	KCNQ3	PBRM1	SMARCA4
2p16.3	ACTL6B	CTCF	KDM3B	PCDH19	SMARCC1
2q34 duplication	ADNP	CTNNB1	KDM5B	PHF21A	SMARCC2
2q37 deletion	ADSL	CUL3	KDM6B	PHF3	SON
3q29	AFF2	DDX3X	KRAS	PHIP	SOS1
5p deletion	AHDC1	DEAF1	KMT2A	POGZ	SOS2
5q35	ALDH5A1	DHCR7	KMT2C	POMGNT1	SOX5
6q16 deletion	ANK2	DLG4	KMT2E	PPP1CB	SPAST
7q11.23	ANK3	DMPK	KMT5B*	PPP2B	SRCAP
8p23.1	ANKRD11	DNMT3A	LZTR1	PPP2R1A	STXBP1
9q34 duplication	ARHGEF9	DSCAM	MAGEL2	PPP2R5D	SYNCRIP
15q11.2-q13.1	ARID1B	DST	MAP2K1	PSMD12	SYNGAP1
15q13.3 deletion	ARX	DYRK1A	MAP2K2	PTCHD1	TAOK1
15q15 deletion	ASH1L	EBF3	MBD5	PTPN11	TANC2
15q24 deletion	ASXL3	EHMT1	MBOAT7	PTEN	TBCK
16p11.2*	ATRX	EIF3F	MECP2	PURA	TBR1
16p12.2 deletion**	AUTS2	EP300	MED13	RAF1	TCF20
16p13.11	BAZ2B	FMR1	MED13L	RAI1	TCF4
16p13.3 deletion	BCKDK	FOXG1	MEIS2	RELN	TLK2
17p11.2	BCL11A	FOXP1	MYT1L	RERE	TRIO
17q11.2	BRAF	GIGYF1	NAA15	REST	TRIP12
17q12	BRSK2	GIGYF2	NBEA	RFX3	TSC1
17q21.3	CACNA1C	GRIN1	NCKAP1	RIMS1	TSC2
22q11.2	CAPRIN1	GRIN2A	NEXMIF**	RIT1	TSHZ3
22q13.3 deletion	CASK	GRIN2B	NF1	RORB	UBE3A
	CASZ1	GRIN2D	NIPBL	SCN1A	UPF3B
	CDKL5	HIVEP2	NLGN2	SCN2A	USP9X
	CHAMP1	HNRNPH2	NLGN3	SCN8A	VPS13B
	CHD2	HNRNPU	NLGN4X	SETBP1	WAC
	CHD3	HRAS	NRAS	SETD2	WDFY3
	CHD7	IQSEC2	NR3C2	SETD5	ZBTB20
	CHD8	IRF2BPL	NR4A2	SHANK2	ZNF462
	CIC	KANSL1	NRXN1	SHANK3	ZNF292
	CNOT3	KAT6A	NRXN2	SHOC2	
	CREBBP	KATNAL2	NRXN3	SIN3A	
	CSDE1	KCNB1	NSD1	SLC6A1	
	CSNK2A1	KCNQ2	PACS1	SLC9A6	

* *Includes deletions and duplications that include at a minimum the BP4 - BP5 region (proximal) or BP2 - BP3 (distal) region*
** *Formerly known as 16p12.1 deletion*
* *Formerly known as SUV420H1*
** *Formerly known as KIAA2022*

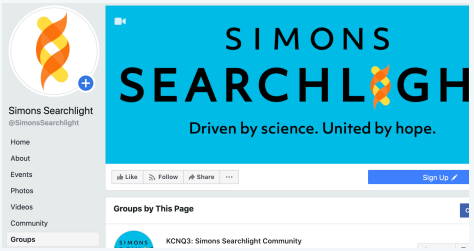
This list was updated May 2020. **simonssearchlight.org**

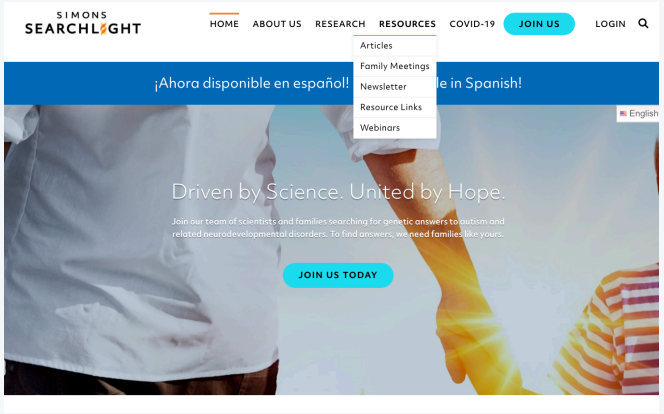
SIMONS SEARCHLIGHT

SIMONS SEARCHLIGHT

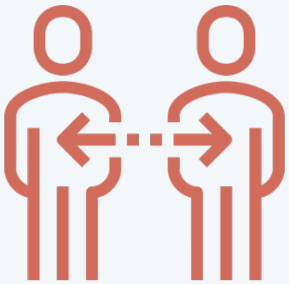
Enrollment and participation

Simons Searchlight Community





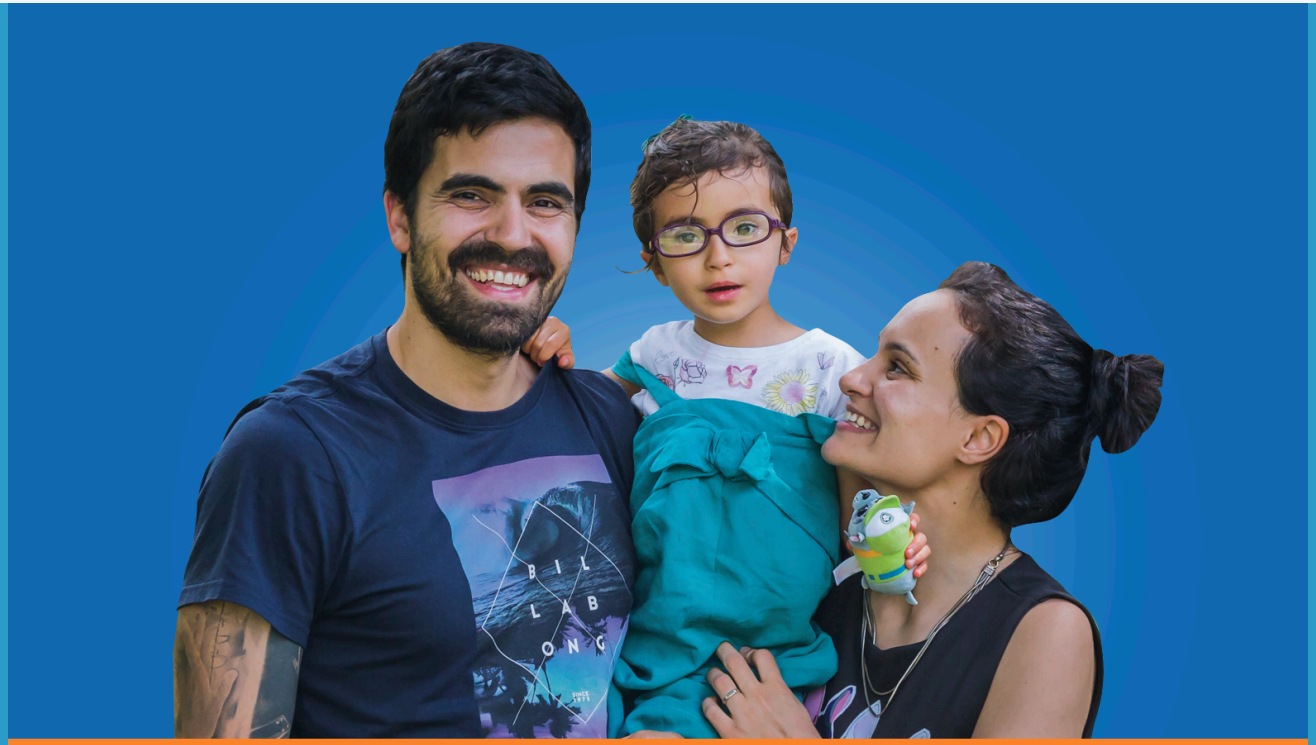
Collect **high quality data** in a standardized way for these rare genetic conditions.



Share this **de-identified data to qualified scientists** to help move research forward on these conditions.

Contribute to **advancements** that will also help future families.

SIMONS SEARCHLIGHT



www.SimonsSearchlight.org
coordinator@simonssearchlight.org | 1-855-329-5638

Facebook: **facebook.com/SimonsSearchlight**
Twitter: **@s_searchlight**
Youtube: **youtube.com/SimonsSearchlight**

SIMONS
SEARCHLIGHT

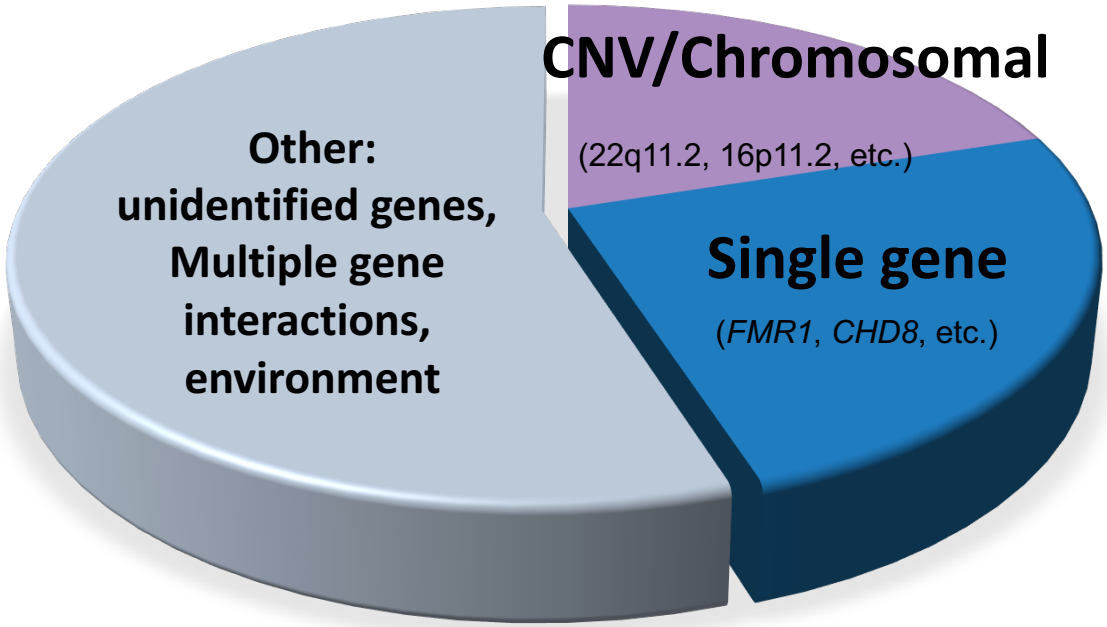
The Genes to Mental Health Network (G2MH)

Dr. Christa Martin
Associate Chief Scientific Officer, Geisinger
Professor & Director, Autism & Developmental Medicine Institute

Presenting on behalf of the G2MH Network

Geisinger

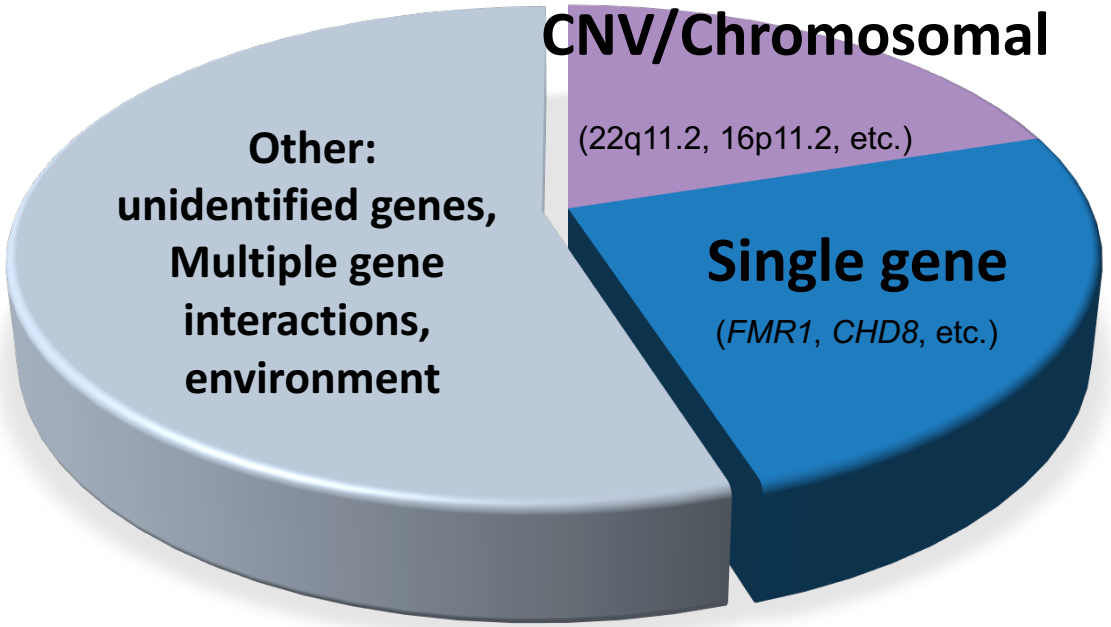
Genetic Causes of Developmental Brain Disorders



At least 40-45% of individuals with a DBD referred for clinical genetic testing have an identifiable genetic diagnosis

(CNV and single gene combined)

Genetic Causes of Developmental Brain Disorders



In Autism Spectrum Disorder – the diagnostic yield is ~25%
(CNV and single gene combined)

Behavioral vs. Etiological Diagnosis

Autism
Fragile X syndrome

Autism
22q11.2 del

Autism
Angelman syndrome

- Find a cause
- Provide information about recurrence risk
- Targeted medical monitoring
- Etiology-specific interventions

The Genes to Mental Health (G2MH) Network

- NIH-funded U01 project
- Initiated in June 2019
- International network
  - Currently includes:
    - 4 collaborative projects
    - 15 sites (11 North America; 3 Europe; 1 Africa)

Genes to Mental Health (G2MH) Network

4 Collaborative Projects
15 Sites
Funded by NIMH and NICHD

G2MH Principal and Co-Investigators

Organizing Institutes:

Geisinger
David Ledbetter
Christa Lese Martin
Christopher Chabris
Brenda Finucane
Scott Myers
Cora Taylor

Penn
Raquel Gur

Boston Children's Hospital
David Glahn

Broad Institute
Elise Robinson
Anne O'Donnell-Luria

Cardiff
Marianne Van Den Bree
Michael Owen
Nigel Williams

Children's Hospital of Pennsylvania
Donna McDonald-McGinn
Laura Almasy

Hospital for Sick Children, Toronto
Jacob Vorstman
Anne Bassett
Stephen Scherer

Katholieke Universiteit Leuven
Ann Swillen

Maastricht University
Therese Van Amelsvoort

Sainte-Justine University Hospital Center
Sebastien Jacquemont

UC San Diego
Jonathan Sebat

UCLA
Carrie Bearden

University of Cape Town
Kirsty Donald

University of Washington
Rachel Earl
Evan Eichler

Washington University St. Louis
John Constantino
Dustin Baldridge

Goals and Impact of Network

- Identify individuals and families with genetic causes of neurodevelopmental and psychiatric disorders, *including autism*
- Characterize the symptoms associated with these genetic causes
- Understand how family genetic background contributes to the risk for developmental and psychiatric symptoms
- Discover early interventions to prevent symptoms
- Develop targeted treatments
- Identify harmful or inadequate interventions

Recruitment

- Who is eligible?
  - Children and adults *with a known genetic cause of autism* and their family members (with and without genetic change)
  - Study is focused on:
    - 1q21.1 deletions
    - 15q13.3 deletions
    - 16p11.2 deletions and duplications
    - 22q11.2 deletions and duplications
    - *CHD8* single gene changes
    - But, also interested in others …

Who to Contact for More Information?

Karahlyn Holdren

keholdren@geisinger.edu

570-522-6295

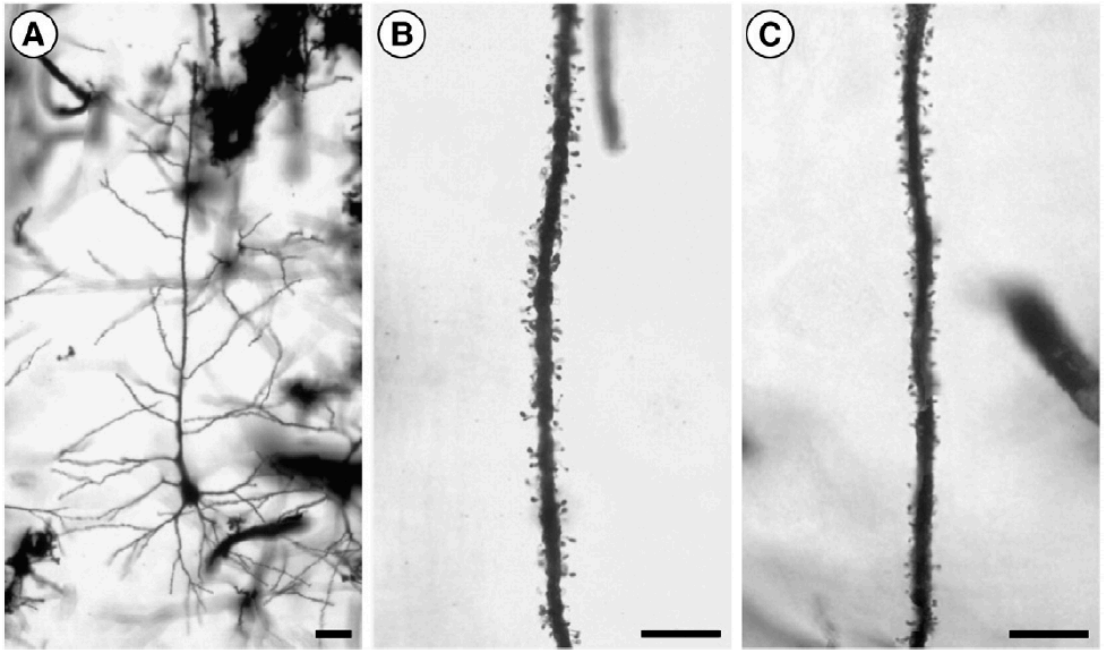
Thank You!

Autism BrainNet

Alycia Halladay

What have we learned from brain tissue studies?

Brain cells in people with autism have greater spine densities – too many connections



Autism Non-diagnosed

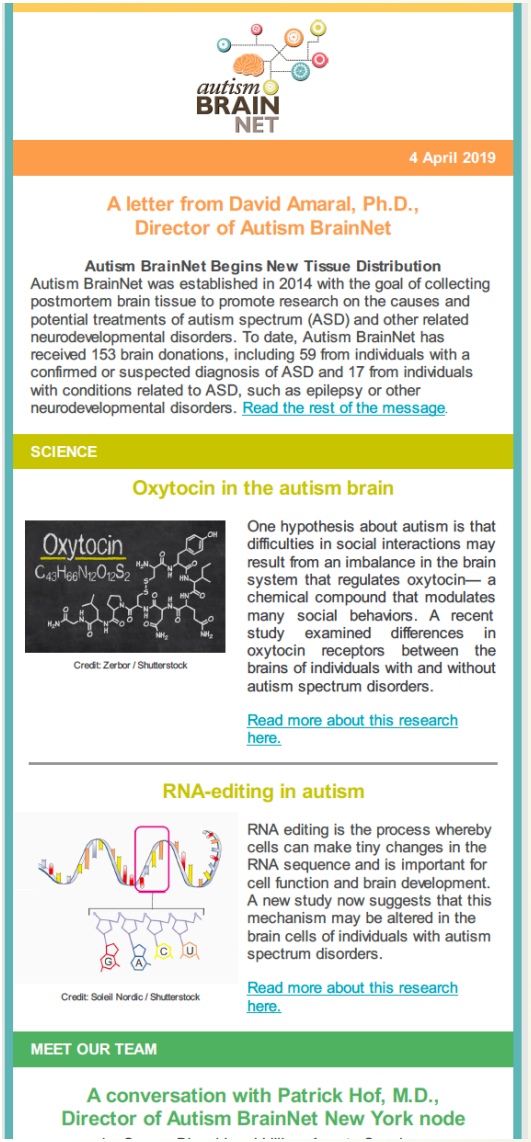
Hutsler and Zhang, 2010

How can you learn more?

Visit takesbrains.org and sign up to receive quarterly news and research updates.

Email us at info@AutismBrainNet.org

Follow us on **Facebook** and **Twitter**: @AutismBrainNet



autism BRAIN NET

4 April 2019

A letter from David Amaral, Ph.D., Director of Autism BrainNet

Autism BrainNet Begins New Tissue Distribution

Autism BrainNet was established in 2014 with the goal of collecting postmortem brain tissue to promote research on the causes and potential treatments of autism spectrum (ASD) and other related neurodevelopmental disorders. To date, Autism BrainNet has received 153 brain donations, including 59 from individuals with a confirmed or suspected diagnosis of ASD and 17 from individuals with conditions related to ASD, such as epilepsy or other neurodevelopmental disorders. Read the rest of the message.

SCIENCE

Oxytocin in the autism brain

Credit: Zerbor / Shutterstock

One hypothesis about autism is that difficulties in social interactions may result from an imbalance in the brain system that regulates oxytocin— a chemical compound that modulates many social behaviors. A recent study examined differences in oxytocin receptors between the brains of individuals with and without autism spectrum disorders.

Read more about this research here.

RNA-editing in autism

Credit: Soleil Nordic / Shutterstock

RNA editing is the process whereby cells can make tiny changes in the RNA sequence and is important for cell function and brain development. A new study now suggests that this mechanism may be altered in the brain cells of individuals with autism spectrum disorders.

Read more about this research here.

MEET OUR TEAM

A conversation with Patrick Hof, M.D., Director of Autism BrainNet New York node

Thank you for helping move research forward!

www.takesbrains.org

AutismBrainNet.org

Autism BrainNet is a program of SFARI SIMONS FOUNDATION AUTISM RESEARCH INITIATIVE