

Autism Research and the Bernier Lab

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Who are we and what do we do?

Located at the University of Washington Center on Human Development and Disability and the Seattle Children's Autism Center

Research and science to improve the lives of individuals with neurodevelopmental disorders and their families



Why participate in autism research?

Autism is a spectrum, so we need all types of people to participate in research if we want to learn more about autism!

Help us understand and discover:

- How autism develops and how genes and the brain are involved
- Earlier diagnosis and treatment
- New, safe, and effective treatments (medications and therapies)
- Personalized interventions and precision medicine
- Treatments for challenging behaviors and medical conditions related to autism
- How to support adults with autism, girls with autism, and other groups within the autism spectrum
- How to support parents, caregivers, and siblings of people with autism

Why our families enjoy participating:

Give back to help others with autism and their families

Help find answers to your questions about autism

Interact with expert researchers and clinicians

Learn more about your child

Visit Seattle!

Earn some money!

Have fun!

Why is research important?

Ensure that new assessments and treatments are safe and effective

Help families and providers make decisions about treatments

Inform future assessments and treatments

“If you’ve met one person with autism...you’ve met one person with autism”

Bernier Lab Research: Genetics and Autism

Right now, we can identify a rare genetic mutation or difference that contributes to autism in up to 10% of people with autism

These genetic differences can be inherited or occur spontaneously (de novo)

Can occur in a single gene or across multiple genes on a specific chromosome (CNV)

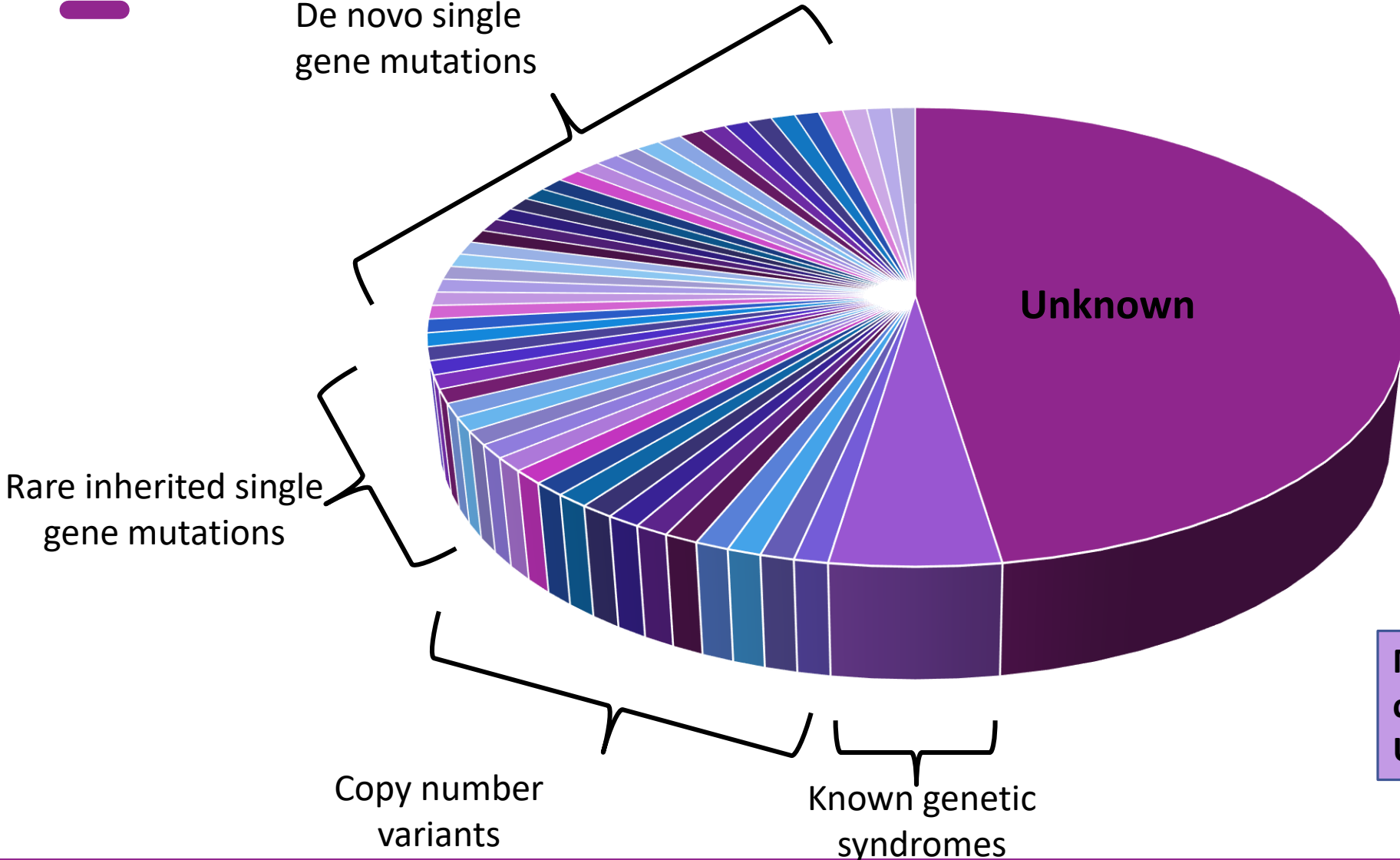
- Single gene: Fragile X syndrome and Rett syndrome
- Multiple genes: 1q21.1 duplications, 16p11.2 deletions and duplications

Studying these genetic differences helps us learn more about how autism develops and identify new treatments for autism

We need more research participants to find more genes!

Current related studies: SPARK, TIGER, Arbaclofen, CONNECT-FX

Genetic Landscape of Autism in 2019



Many findings are still considered “Variants of Unknown Significance”

Bernier Lab Research: Clinical Trials

Studying the safety and effectiveness of new treatments or diagnostic tools

Follow a carefully designed protocol or study plan that is approved by an institutional review board

Frequent health and safety monitoring

Some participants may receive a placebo (inactive) treatment so we can understand the unique effects of the new treatment

Current clinical trials: JAKE-2, V1ADUCT, oRBiting (non-drug), Arbaclofen, CONNECT-FX

Bernier Lab Research: Autism and the Brain

Using non-invasive technologies like EEG, fMRI, and eye tracking to understand how the brain processes and responds to information

- EEG: Measures electrical activity in the brain
- fMRI: Measures blood flow in the brain
- Eye tracking: Measures the motion of eyes and what the eyes are looking at

Shows us how people with autism learn new information, process sensory information, and perceive social situations

Current related studies: GABA, TIGER, Arbaclofen

Participating in Bernier Lab research



Bernier Lab Members

Manager of Program Operations: Micah Pepper

Coordinators: Koko Hall, Wes Ganz, Christina Sargent, Daniel Cho, Theo Ho, Stacy Riffle, Curtis Eayrs

Clinicians: Rachel Earl, Eva Kurtz-Nelson, Jennifer Beighley, Katie Ahlers, Tara Rutter

Interns and Volunteers

Collaborators

First Steps: Screening and Informed Consent

Our coordinators will ask screening questions to make sure that you or child meet eligibility criteria for the study

- Some studies (especially clinical trials) have very specific eligibility criteria; others (like SPARK) are more broad

A coordinator will give you a consent form and discuss the form with you in person or over the phone

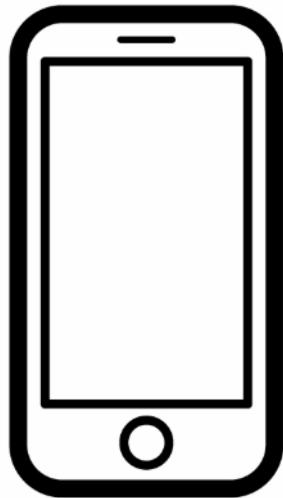
- Describes benefits and risks of participating in a study
- Ask as many questions as you want!

If you give your consent, you and your child will be enrolled in the study

Remote Participation

Complete surveys and interviews online or over the phone

- Schedule with coordinators at times that work for you



Study Visits

At CHDD or Seattle Children's

For your child:

- Social stories, visual schedules, and videos
- Appropriate toys and games
- Allergy-conscious snacks and drinks
- Breaks as needed
- Childcare for siblings



Travel arrangement and reimbursement may be available (depending on the study) as well as compensation for your time

Bernier Lab Studies



Current research studies

- SPARK
- oRBiting
- JAKE-2
- V1ADUCT
- GABA
- Arbaclofen
- TIGER
- CONNECT-FX

Background of SPARK



- Large sample sizes needed
- Families should know their genetic information
 - Targeted studies and treatments
 - Connection with other families

SOURCES

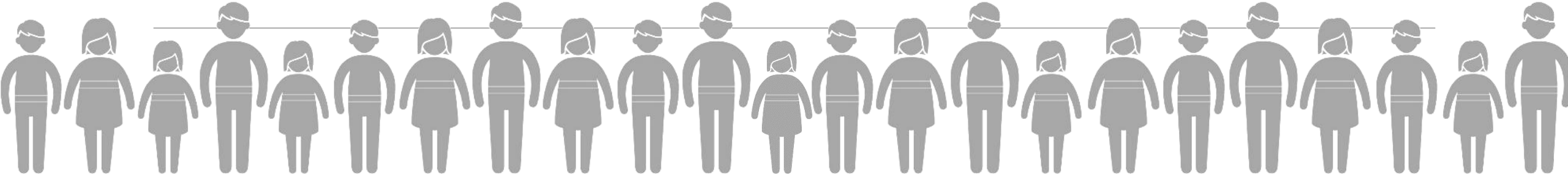
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SPARK's Mission

- An online research partnership involving 50,000 individuals with autism and their families to **speed up research and advance understanding of autism**
 - Sponsored by the Simons Foundation Autism Research Initiative (SFARI)
- SPARK will collect genetic information from individuals affected by autism via saliva samples

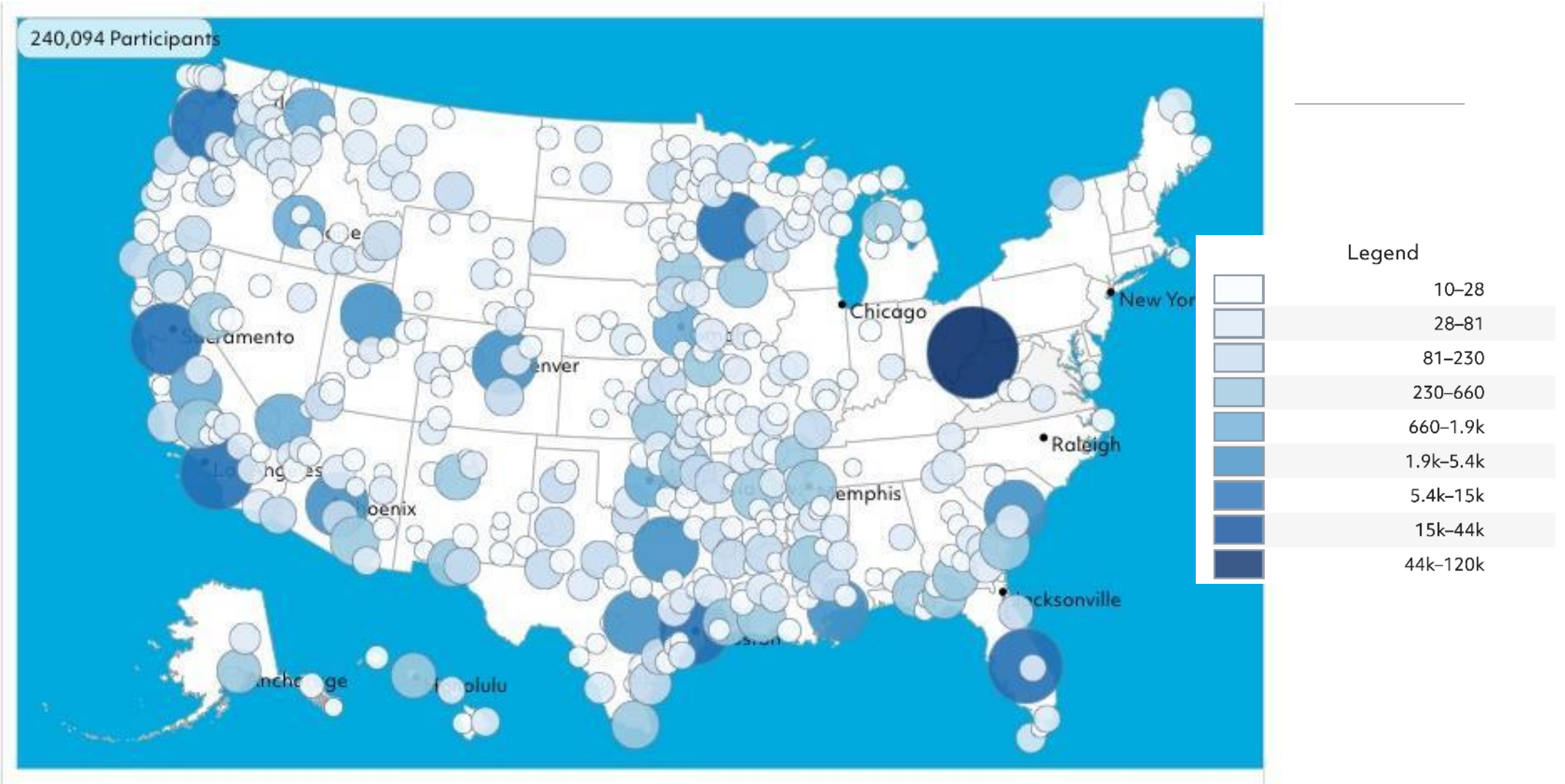
Who is eligible to join SPARK?



The entire autism community!

Recruiting individuals with a diagnosis of autism and their families to join SPARK

SPARK across the United States



Registration Process



Benefits of Participating in SPARK



Participating families will receive up to \$50 gift card!



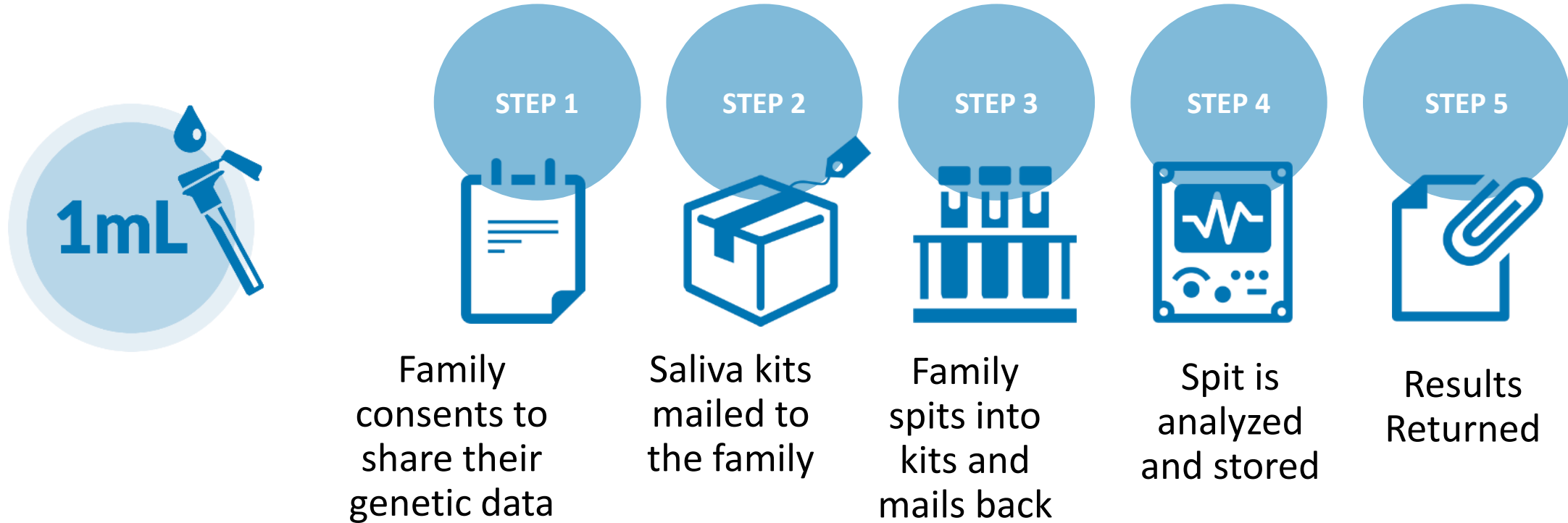
SPARK will provide families with tips and strategies for navigating life with autism



SPARK will conduct genetic testing in a CLIA-certified lab with results returned*

* This research study is not responsible for the cost of provider appointments.

Saliva Sample Collection and DNA Analysis



Progress as of Fall 2019



186,822
participants
enrolled

18,612
completed trios



71,436 participants with
ASD

To learn more about SPARK:

- www.SPARKforAutism.org/UW
- SCACstudies@seattlechildrens.org
- 206-987-7917
- Coordinator: Theo Ho

oRBiting

- The study: a non-drug clinical study aiming to characterize different scales to measure repetitive and restricted behaviors in different ASD sub-populations over time and also to explore the use of digital biomarkers
 - The study includes behavioral assessments completed with the caregiver and/or the participant by the clinician
 - The study is expected to last approximately 15 weeks, including screening
 - During clinic visits, we'll ask you to complete a range of questionnaires and we'll collect data from wearable devices (a smartphone and a wrist gadget) that we'll provide you with at screening
- Who is eligible?
 - Individuals with autism ages 5-45 years
- Coordinator: Daniel Cho, (206) 987-7502, daniel.cho@seattlechildrens.org

JAKE-2

- The study: evaluates an investigational drug to determine the safety and usefulness for treating patients with ASD
 - Participants will randomly be assigned to receive either active medication or placebo
 - Eligible participants will meet with study doctors for up to 13 weeks.
 - 1 visit every 2 weeks for about 3 months with 1 additional follow up visit
- Who is eligible?
 - Anybody with a clinical ASD diagnosis between the ages of 13-30
- Benefits: You will receive study-related medical care, monitoring, and frequent visits with a study doctor. We will give you information about ASD and possible treatment options.
- Participants may be compensated for time and travel.
- Coordinator: Theo Ho, SCACstudies@seattlechildrens.org, 206-987-7917

V1ADUCT

- The study: A phase 3, double blind investigational study of Balovaptan, a drug that is being assessed to see if it can help adults with ASD better manage social and communication challenges
 - Participants will randomly be assigned to a placebo or active drug group
 - Participants will visit the study clinic for regular health checks and measures across 24 weeks
 - After 24 weeks, participants can also volunteer to be a part of a 2 year long open-label extension period.
- Who is eligible?
 - Adults with ASD who are 18 and older
- Participants will receive \$50 or \$75 per study visit, depending on the scope of the visit
- SCACstudies@seattlechildrens.org, 206-987-7917

GABA

- GABA And Behavior in Autism (GABA)
- The study: explores how the brain processes sensory information
 - The study will involve 1 visit to UW
 - Participants will complete neurocognitive assessments (4-5 hours) and a blood draw (30 mins)
- Who is eligible?
 - Participants 18-35 years old
 - Participants have normal or corrected to normal hearing and vision, and must be diagnosed with ASD
- Compensation: \$100
- [rablab@uw.edu](mailto:rabl@uw.edu), 206-616-2889

Arbaclofen

- The study: May help doctors and scientists learn more about 16p11.2 deletion. The study will try to find out if a medicine can help with speech, learning, and coordination.
 - You will come into the clinic 4 or 5 times – first visit determines eligibility for the study
 - The medicine is called arbaclofen.
 - Some people in the study will get tablets of arbaclofen. Other people will get a pill that looks and tastes the same but doesn't have any arbaclofen in it.
 - There are 2 blood draws in the study
- Who is eligible?
 - Children between the ages of 5-17 years
- You will be in the study for less than 28 weeks
- [rablab@uw.edu](mailto:rabl原因@uw.edu), 206-616-2889

TIGER

- The Investigation of Genetic Exome Research (TIGER) study
- The study: explores how specific genetic events may contribute to autism spectrum disorder and related developmental disorders.
 - Participation may include clinical and neurocognitive assessments, detailed interviews about developmental history, EEG, 2D and/or 3D photos, language testing, a medical examination, and a blood draw.
- Who is eligible?
 - Individuals ages 4 to adult with ASD or related neurodevelopmental disorders who have a known genetic mutation associated with ASD
- The study may be completed via remote participation or an in-person visit.
- Expenses related to the study visit will be covered by the study and families will be reimbursed \$100 for their participation upon study completion
- [rablab@uw.edu](mailto:rabl@uw.edu), 206-616-2889

CONNECT-FX

- The study: an investigational drug called ZYN002 is being studied to determine if it can help manage some of the most common behavioral symptoms associated with Fragile X Syndrome (FXS)
 - ZYN002 is a gel which is applied to the skin twice a day
- Who is eligible? ***other eligibility criteria may apply**
 - Boys and girls
 - Ages 3-17 years
 - Diagnosed with full mutation Fragile X
- Study related medical exams, study medication and reimbursement of reasonable study-related costs, such as travel and incidental expenses, will be supplied to qualified participants at no cost
- Patients who participate may receive reimbursement for reasonable study-related costs such as travel and incidental expenses
- Micah Pepper, 206-616-8770, peppem@uw.edu

Summary

Learn more about Bernier Lab studies by contacting:

- Bernier Lab: [rablab@uw.edu](mailto:rabl@uw.edu), 206-616-2889
- Seattle Children's Autism Center: SCACstudies@seattlechildrens.org, 206-987-7917

Our coordinators can talk to you about which studies might be a good fit for your child and your family

New studies starting all the time

We look forward to hearing from you!

Autism Research Resources

Bernier Lab website: <https://depts.washington.edu/rablab/>

Seattle Children's Autism Center: <https://www.seattlechildrens.org/clinics/autism-center/>

Spectrum: <https://www.spectrumnews.org/>

Organization for Autism Research: <https://researchautism.org/>

Thank you!

