SPARK

Accelerating Research and Improving Outcomes in Autism Spectrum Disorder

Pamela Feliciano, Ph.D.
Scientific Director, SPARKforAutism.org

31 March 2016
Significant Challenges in Developing Effective Treatments for ASD

- Heterogeneity in **symptoms and etiology** (roughly 40% genetic/60% environmental)
  - Some progress in genetics, but much more to learn
  - Some progress in environmental factors, but largely unknown

- How are **symptoms linked to etiology**?

- Objective measures required for translational and clinical research

- Only treatments are behavioral and resource-intensive
How are symptoms linked to etiology?
Objective measures needed
More efficient treatments needed

• Only available treatments are behavioral and resource-intensive

• 10 years of intensive intervention = 20,000 hours of 1:1 therapy = $1m

• How much impact do such interventions have on long-term outcome?
Research efforts so far, but much more data needed

• Etiology
  • Genetics on 18K individuals with autism so far → 60 genes, but hundreds left to discover
  • Some progress in environmental factors, but largely unknown, perhaps up to 60% of cases
• Heterogeneity
  • Genetics-first approaches to study by genetic subtype
• What behaviors are proximal to etiology?
  • Genetic animal models of autism: what happens at a cellular and behavioral level?
• Objective measures required for translational and clinical research
  • EU-AIMS
  • Asdbiomarkers.org
  • Targeted grants in this area by SFARI
SPARK must scale

REGISTRATION PROCESS

1. Register at SPARKforAutism.org
   - Takes ~30-60 minutes and can be paused/resumed along the way
   - Invite family members to join

2. Consent to share your data
   - Known data: consent form to share the study uses of your personal information
   - Unknown data: contact for research studies

3. Share family history, behavioral and medical information
   - Data is stored securely and without any identifying information

4. Consent to provide a saliva sample
   - Receive genetic results from specific, non-invasive DNA saliva sample

5. Provide saliva DNA sample(s)
   - 2-3 weeks after registering, saliva collection kit will be mailed to your home
   - Kit includes a saliva collecting tube and an envelope to send back

6. DNA is analyzed and stored
   - Saliva samples are sent to a secure laboratory and DNA will be extracted for analysis
   - Diverse researchers analyze the saliva and determine if they have a sufficient sample - Once all necessary steps are completed, the individual with autism will receive a $50 gift card and an $50 per family
   - If you indicate you are interested in receiving genetic results and should there be any identified, they may be provided to the medical provider you indicate

7. Stay engaged with SPARK!
   - Check your Dashboard for the latest resources, new research opportunities, and updates
   - Sign up for the SPARK e-newsletter

SPARK
Igniting autism research
Improving lives
What SPARK will do for individuals and families affected with autism

- Participants are partners, not subjects
- Commitment to return individual genetic results related to autism to medical professional that participant designates
- Return individual results on standardized behavioral questionnaires
- Inform community of aggregate results
- Opportunities to interact with autism experts online
What SPARK will do for the research community

- Full understanding of the genetic architecture of autism
- Enable more efficient recruitment (genotype-based if desired) to entire research community at no cost
- All behavioral and genetic data will be made accessible to any qualified researcher
- Some embargoes on entire genetic dataset but data will be released quickly after it is generated
Genetic analysis in SPARK

- Exome sequencing of trios to accelerate gene discovery
- Sequencing of unaffected siblings when possible
- State of the art genome-wide genotyping for GWAS
- SPARK genomics consortium will analyze first 10,000 families
What’s next for SPARKforAutism.org

- Wrapping up pilot phase (recruitment of 500 trios)
- National launch on April 21
- Release data in scheduled releases beginning in Q3, 2016
- Open cohort to recruitment by research community in 2016
- Recruit and collect specimens from at least 10,000 trios through April 2017
- Report on pilot and genetic analysis of first 10,000 families
The SPARK Team: From Exomes to Twitter

Wendy Chung
Dir. Clinical Research

Pamela Feliciano
Scientific Director

Amy Daniels
Project Manager

Stephen Zukin
Dep. Dir. Clinical Research

LeeAnne Green Snyder
Clinical Research Scientist

Jennifer Tjernagel
Project Manager, VIP

Casey White-Lehman
Project Manager, SSC

Elizabeth Brooks
Aas. Project Manager, SSC

Karen Walton-Bowen
Clinical Operations

Rick Remington
Outreach Manager

Julie Manoharan
Project Coordinator

Vincent Myers
Research Assistant

Hana Zaydens
Administrative Assistant

Alpha Amatya
Sr. Software Engineer

Richard Marini
Sr. Software Engineer

Martin Butler
Software Engineer

Andrei Salomatoav
Bioinformatics Engineer

Natalia Volfovsky
Analytics Manager

SFARI
SIMONS FOUNDATION
AUTISM RESEARCH INITIATIVE

SIMONS FOUNDATION
Advancing Research in Basic Science and Mathematics