



SPARK

Accelerating Research and Improving Outcomes in Autism Spectrum Disorder

Pamela Feliciano, Ph.D.

Scientific Director, SPARKforAutism.org

31 March 2016

SPARK

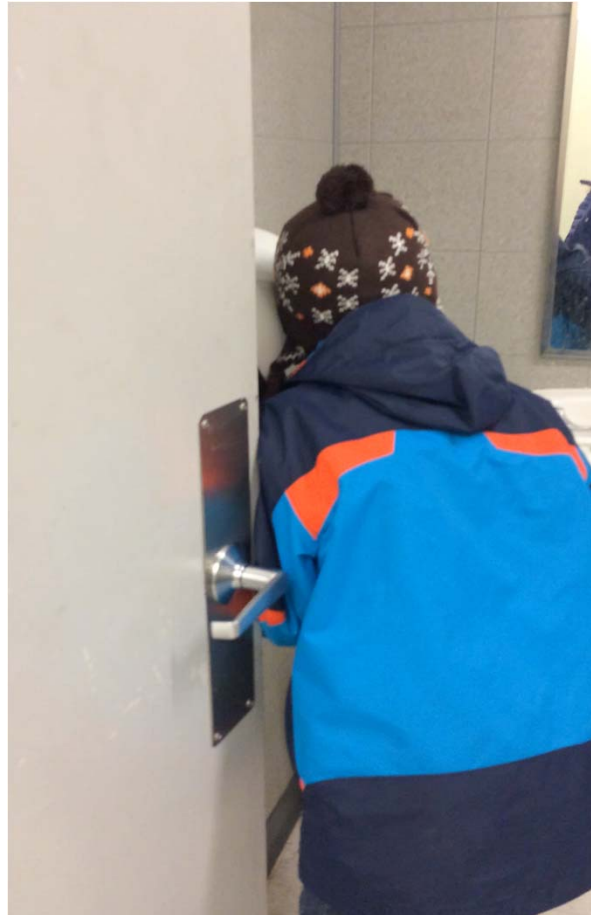
Igniting autism research
Improving lives



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 ~20-30 mins and can be paused/saved along the way
- Invite family members to join



2 Consent to share your data

- Review data consent form detailing the study, uses of your personal information and future contact for research studies



3 Share family history, behavioral and medical information

- Data will be stored securely and without any identifying information



4 Consent to provide a saliva sample

- Review genetic consent form specific to sharing DNA saliva samples



5 Provide saliva DNA sample(s)

- 2-3 weeks after registering, a saliva collection kit will be mailed to your home
- Kit includes a tube for spitting and an absorbent swab (for anyone unable to spit)



6 DNA is analyzed and stored

- Saliva sample(s) are stored in a secure laboratory and DNA will be extracted for analysis
- Once researchers analyze the saliva and determine they have a sufficient sample – and once all surveys in your dashboard are completed – the individual with autism will receive a \$50 gift card (maximum \$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 research updates
- Sign up for the SPARK e-newsletter

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
Asst. 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 Salomatov
Bioinformatics Engineer



Natalia Volfovsky
Analytics Manager