Identify risk factors whether genetic, environmental or epidemiological.

Use non-human organisms to understand how these risk factors alter brain function and animal behavior.

Promote preclinical and clinical investigations to improve autism diagnosis & therapy.
Simons Foundation Autism Research Initiative (SFARI)

- 2020 budget: **$92 million**
- Since launch in 2003: **>$525 million** in external research support
- **>550 investigators**
SFARI Cohorts

SSC
Simons Simplex Collection
~10,000 individuals

SEARCHLIGHT
~2,0000 individuals

SPARK
Simons Powering Autism Research
More than 300,000 individuals

AIC
Autism Inpatient Collection
~1,000 individuals

BRAIN NET
~250 individuals

sfari.org/resources
Mission: improve the understanding, diagnosis and treatment of autism spectrum disorders (ASD) by funding innovative research of the highest quality and relevance

• Current Funding Opportunities: sfari.org/funding-opportunities
• Policies and Procedure: https://www.sfari.org/funding-opportunities/policies-and-procedures/
• Resources: sfari.org/resources
• Data & biospecimen requests, recruitment: base.sfari.org
SFARI aims to support reproducible scientific research of the highest quality. This can be greatly facilitated by making all raw data, analysis methods, and, when applicable, computer code, available to the research community as quickly and transparently as possible.

Data generated from collections is considered “community resources” and, as such, all these data, including, but not limited to, alignment files and variant calls, will be made available to the entire research community, pre-publication, in real-time, as they are produced.

Besides being subject to any limitations by consent and IRB protocol, a publication embargo will also be enforced for 6 months after the defined project is complete while foundation-sponsored analyses are underway.
The Simons Foundation has no wish to participate in the commercialization of any intellectual property that may result from a grant. Notwithstanding this commercialization disclaimer, the Simons Foundation has the right to release a summary of findings of the research within a reasonable period of time, typically within six (6) months of the date of the grant expiration.
Some Current Funding Opportunities

To produce foundational knowledge about neurobehavioral differences associated with ASD, which will directly inform the development of refinement of tools needed for translational efforts, such as biomarkers and outcome measures.

Quantitative and objective measures of human behavior
- Psychophysics
- Computer vision and machine learning
- Wearables and app-based data collection

Human neuroscience
- EEG, fMRI, MEG

Encourage use of SPARK and Simons Searchlight Research Match
SFARI Base is a clearinghouse for autism and autism-related research data and biospecimens supported by the SFARI funding as well as an online portal for the submission of research recruitment requests.

The use of SFARI cohorts (SSC, Simons Searchlight and AIC) data and biospecimens is limited to projects related to advancing the field of autism and related neurodevelopmental disorder research. SPARK phenotypic and genomic data does not have this limitation. Applications will be reviewed to ensure the use of data and biospecimens is in compliance with the Institutional Review Board (IRB)-approved consent language.

All investigators using SFARI resources must adhere to the guidelines outlined in our Acknowledgement Policy.
Depositing Data to SFARI Base

Information for submission:

1. Project Title/Abstract/PI/Data manager
2. Data Manifest: List of all files, Each file should be accompanied by the Md5 checksum.
3. Samples IDs and all types of files that will be available per sample.
4. Size of the dataset.
5. Description of the methods/analysis used for generation of the data/analysis files including versions of the software and parameters applied. Github link.
6. Embargo information
7. Manuscripts other references.

SFARIBase ID will be issued to the deposited data set and it can be used as a reference in further publications.
Genomic Data Sharing

- **SEQUENCING LAB**
- **FLATIRON CLUSTER**
- **FNAL ARCHIVE**
- **AMAZON S3**

**RESEARCHER**

**SFARI Base Collections**
- Principal Investigator submits request for sets of genetic data (BAM, CRAM, SV, VCF, docs).

**SFARI Base Approval**
- Request goes through approval chain to confirm objective for research, adherence to HIPAA, key persons in lab.

**Access Granted**
- Access granted according to preferred method, chosen from Globus Online, AWS S3, or FermiLab.

**Access Confirm**
- Check in with lab manager to confirm access to datasets, and address technical issues with access.

**GLOBUS**

**OSG**

**AWS**
GPF: gpf.sfari.org

Ivan Iossifov Ph.D.
CSHL & NYGC
gnomAD-like SFARI Browser: genomes.sfari.org

CHD8 chromodomain helicase DNA binding protein 8

<table>
<thead>
<tr>
<th>Gene Constraint</th>
<th>Category</th>
<th>Exp. SNVs</th>
<th>Obs. SNVs</th>
<th>Constraint metrics</th>
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<td>Synonymous</td>
<td>502.4</td>
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<td>Z = 1.03 a/e = 0.94 (0.87 - 1.02) 0 1</td>
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<td>LoF</td>
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<td>pU &lt; 0.05 a/e = 0.94 (0.87 - 0.98) 0 1</td>
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</table>

1 denotes a consequence that is for a non-canonical transcript.

Monkol Lek Ph.D.
Yale University
Monthly Report
Aug 2020

New users: 33
Returning users: 108
New institutions: 5
Executed institution: 3
Manuscripts: 5
SF Grants: 0

Requests Submitted by Type
- 50: Biospecimen
- 50: Genetic Data
- 50: Imaging/EEG Data
- 50: Phenotypic Data
- 40: Research Match
- Total

Requests Submitted by Cohort
- AIC
- SPARK
- SSC
- Searchlight
- Biospecimen (SSC/Searchlight)
- Total

Month 2019
- Aug 2019
- Sep 2019
- Oct 2019
- Nov 2019
- Dec 2019
- Jan 2020
- Feb 2020
- Mar 2020
- Apr 2020
- May 2020
- Jun 2020
- Jul 2020
- Aug 2020
Conclusions

Functional Data Sharing Policy is important for facilitation of the reproducible biomedical research.

Major stakeholders - Researchers, Funders, Scientific Grant Managers, Informatics teams - should be included in the transparent communications and discussions.