MSSNG

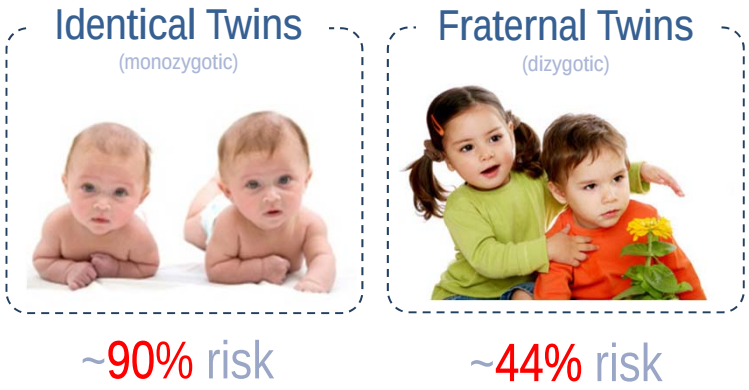
Changing the future of autism through open science

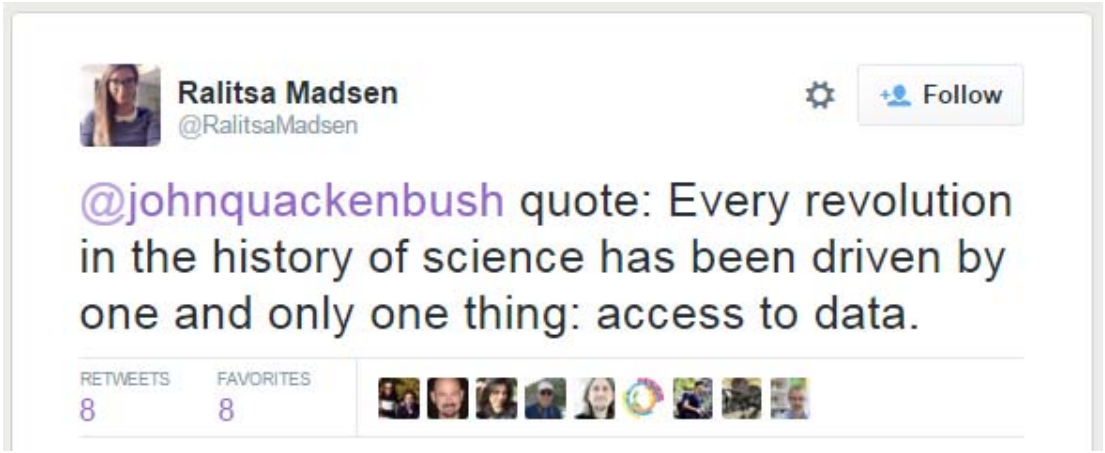
March 31th, 2016

Dr. Mathew T. Pletcher, Ph.D.
Chief Science Officer

Genes and their role in autism

When it comes to well risk factors and etiology of ASD, evidence overwhelmingly implicates the genome.





MSSNG – Using open science to deepen understanding of autism

AUTISM SPEAKS®

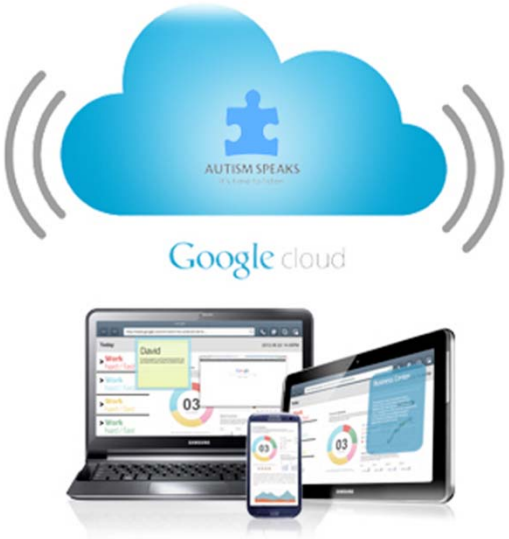
Google™

SickKids®
THE HOSPITAL FOR SICK CHILDREN

AUTISM SPEAKS™
It's time to listen.

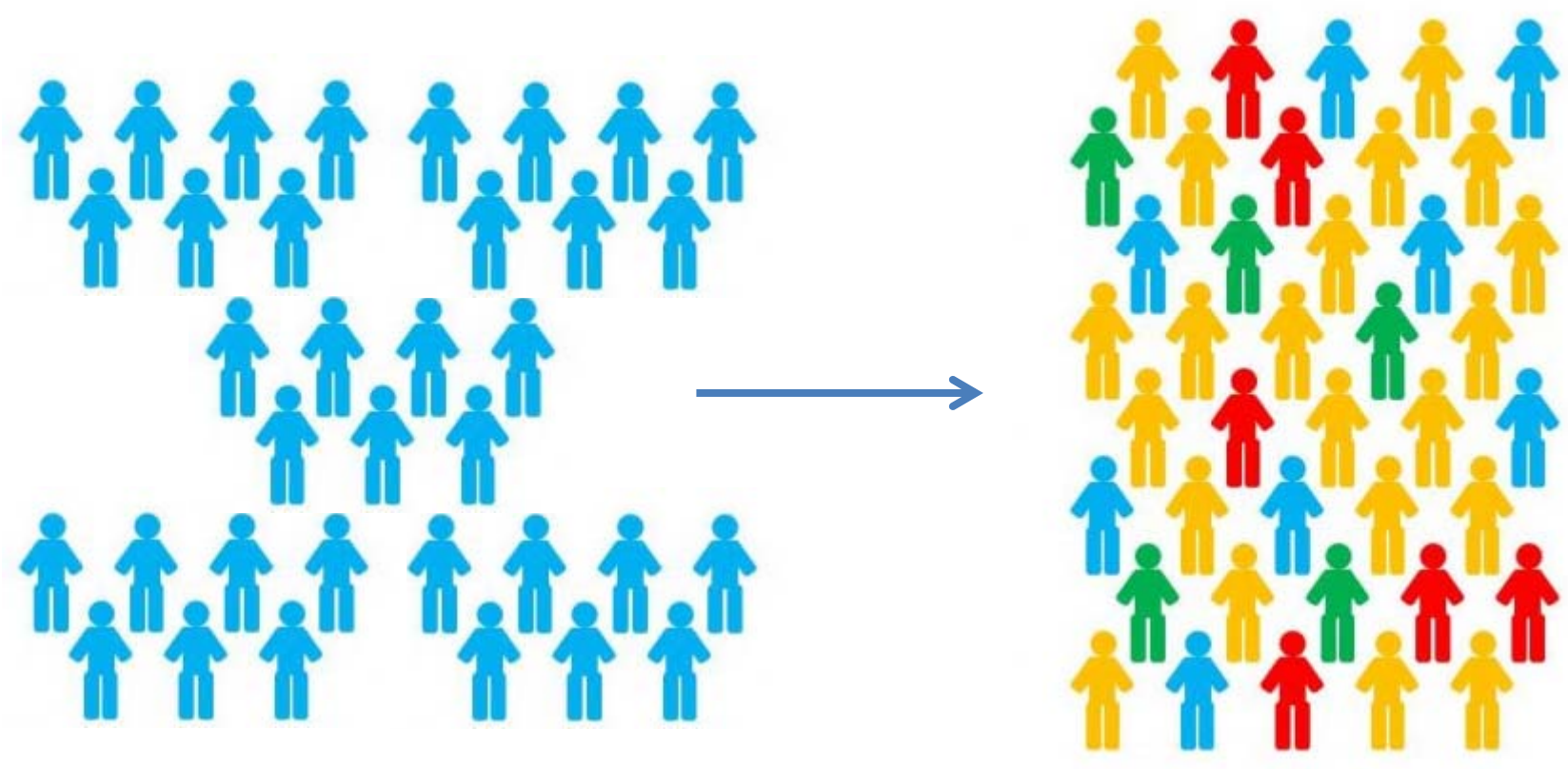
Private-Public partnership to improve understanding and treatment of autism

At least 10,000 whole genome sequences and deep phenotype data from families with autism



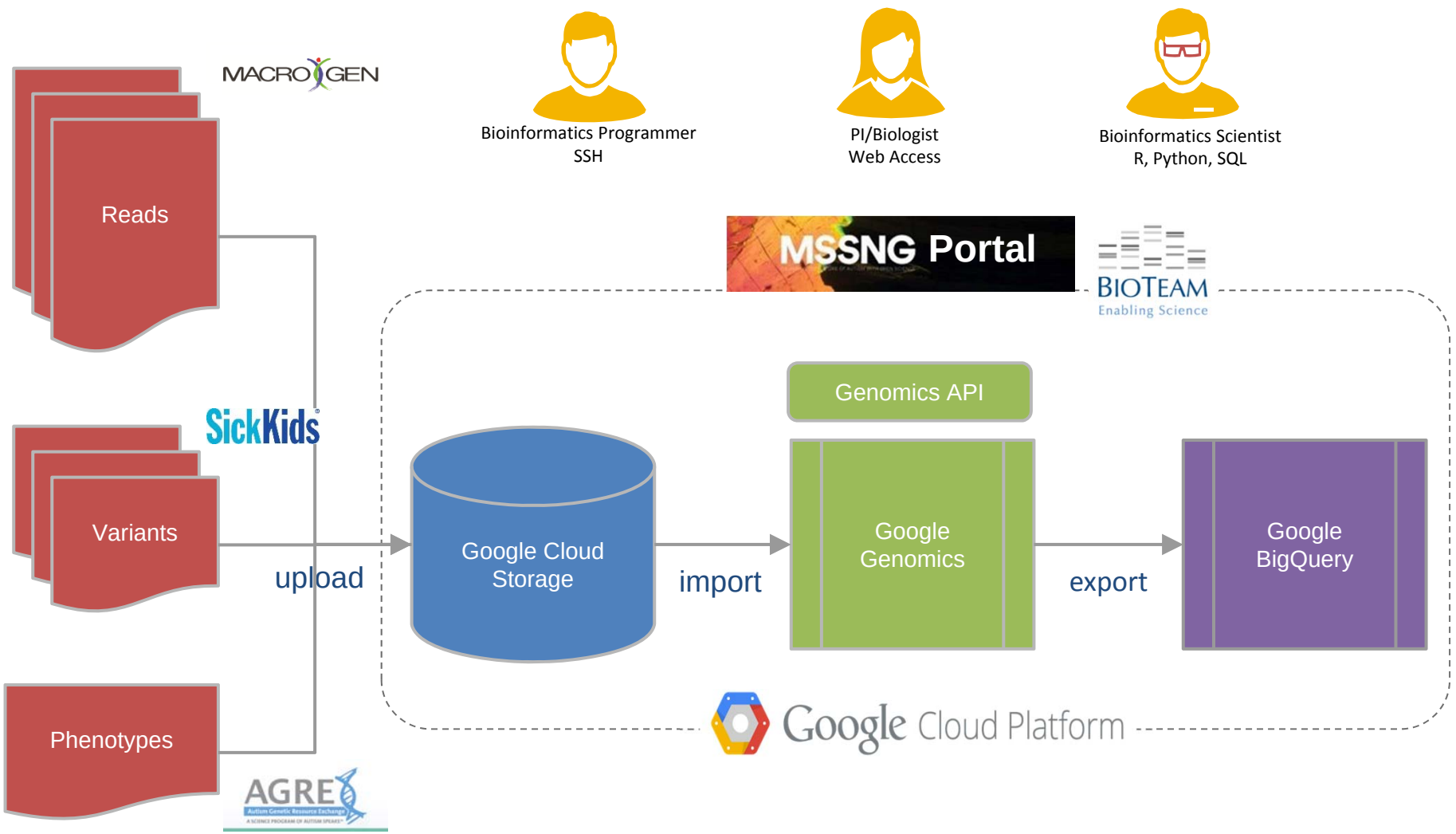
Data made freely available through open-access web-based portal

Changing the way autism is diagnosed and treated



Autism Spectrum Disorder is an umbrella diagnostic term that does not provide meaningful guidance towards prognosis or treatment

MSSNG lives on the cloud



Enabling research around the world

- **93 Investigators**
- **40 Institutions**
- **9 Countries**

Balancing openness with protection of donors

Global Alliance for Genomics & Health

Collaborate. Innovate. Accelerate.

PUBLIC POPULATION PROJECT IN GENOMICS AND SOCIETY

A policy of controlled access

Autism Speaks controls all data access:

1. Researcher applies for access
2. Autism Speaks reviews application
3. Approval of applications by DACO
4. Autism Speaks adds the researcher to the mssng-research-read-access group

Limitations on access

1. Institutional co-signatures
2. IRB-approved protocol
3. Research must focus on autism and related diseases

Current status of MSSNG program

- 6,345 genomes sequenced and being uploaded to the Google cloud
- Over 1,204 additional samples in the queue
- 10,000 genomes completed by summer of 2016
- Two additional scientific papers currently in review

ARTICLE PREVIEW

view full access options ▸

NATURE MEDICINE | RESOURCE

日本語要約

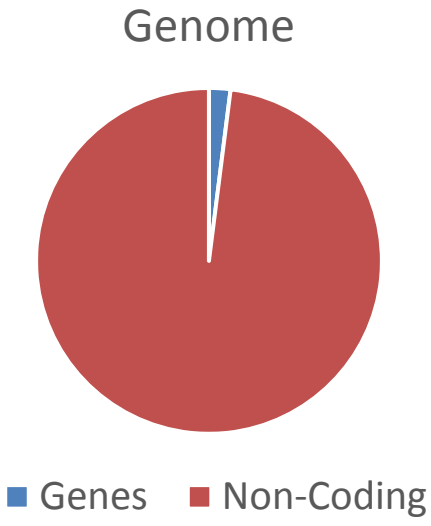
Whole-genome sequencing of quartet families with autism spectrum disorder

Ryan K C Yuen, Bhooma Thiruvahindrapuram, Daniele Merico, Susan Walker, Kristiina Tammimies, Ny Hoang, Christina Chrysler, Thomas Nalpathamkalam, Giovanna Pellecchia, Yi Liu, Matthew J Gazzellone, Lia D'Abate, Eric Deneault, Jennifer L Howe, Richard S C Liu, Ann Thompson, Mehdi Zarrei, Mohammed Uddin, Christian R Marshall, Robert H Ring, Lonnie Zwaigenbaum, Peter N Ray, Rosanna Weksberg, Melissa T Carter, Bridget A Fernandez + *et al.*

Affiliations | Contributions | Corresponding author

Backup

Looking under the lamppost



Important findings in the darkness

AJHG

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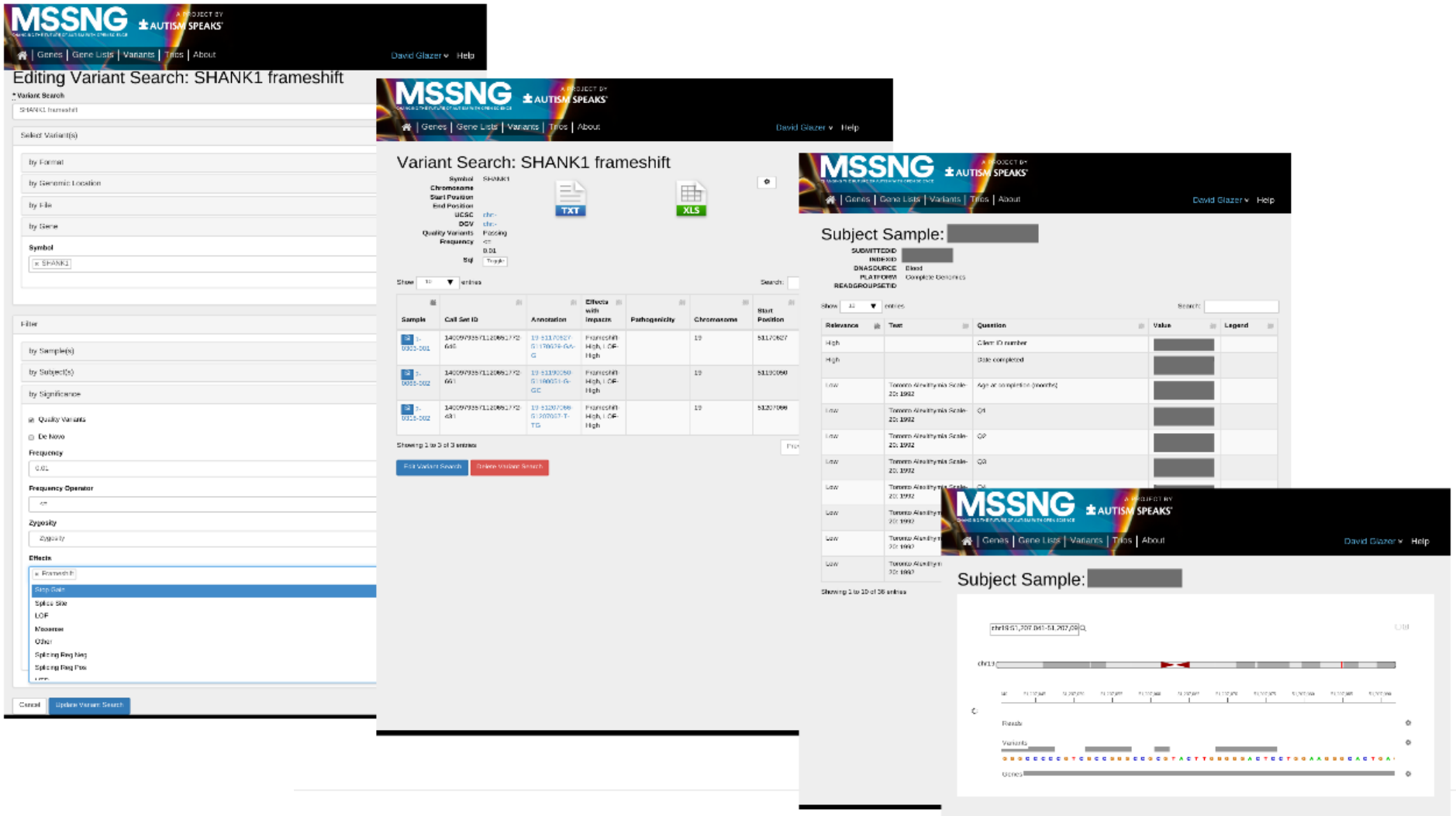
Article

Switch to Standard View

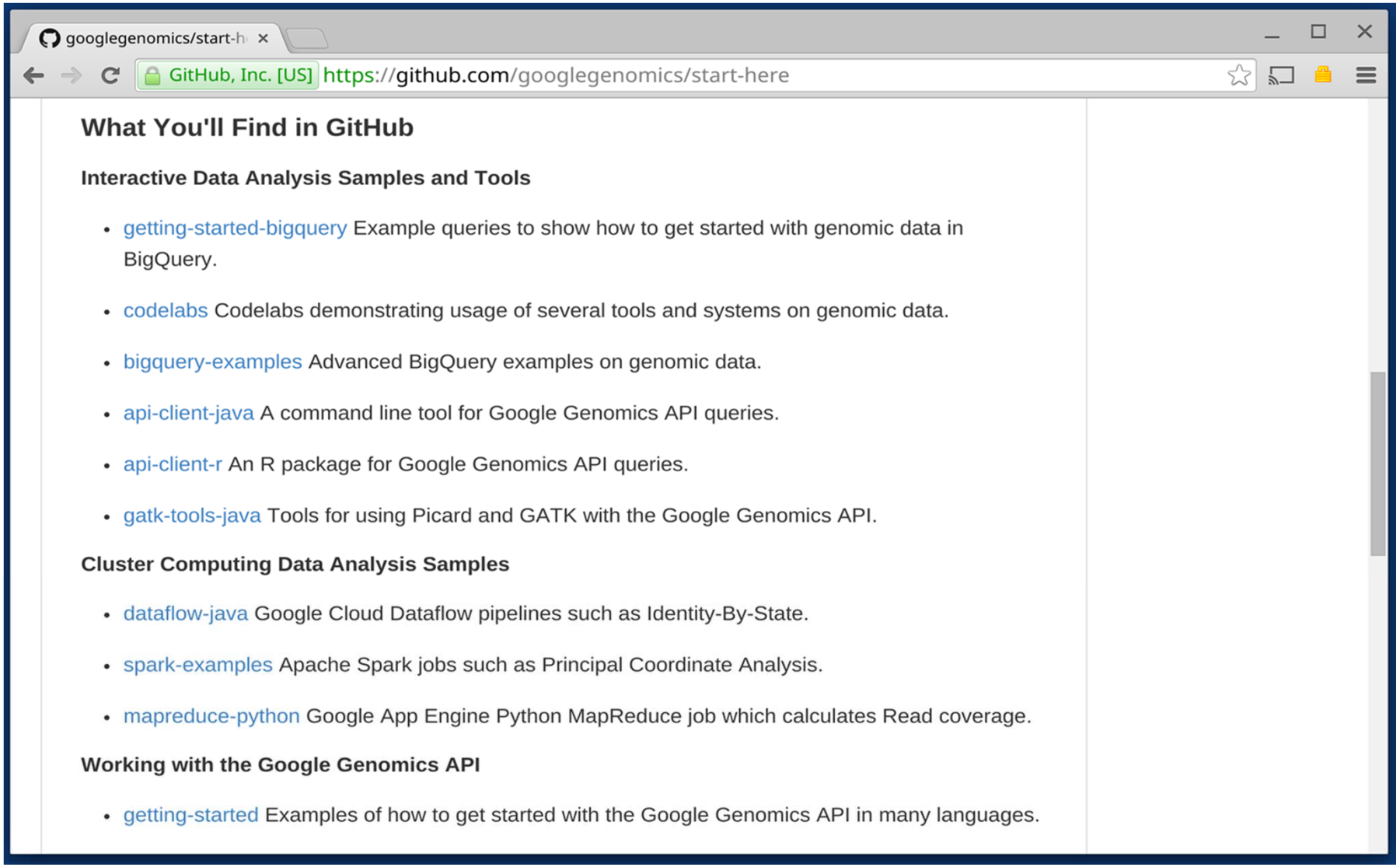
Genome Sequencing of Autism-Affected Families Reveals Disruption of Putative Noncoding Regulatory DNA

Tychele N. Turner, Fereydoun Hormozdiari, Michael H. Duyzend, Sarah A. McClymont, Paul W. Hook, Ivan Iossifov, Archana Raja, Carl Baker, Kendra Hoekzema, Holly A. Stessman, Michael C. Zody, Bradley J. Nelson, John Huddleston, Richard Sandstrom, Joshua D. Smith, David Hanna, James M. Swanson, Elaine M. Faustman, Michael J. Bamshad, John Stamatoyannopoulos, Deborah A. Nickerson, Andrew S. McCallion, Robert Darnell, Evan E. Eichler

MSSNG web portal



Google Genomics on GitHub



Genetic diagnosis provides certainty and enables action

AUTISM SPEAKS®

Why it matters