



MSSNG

**Changing the future of autism through
open science**

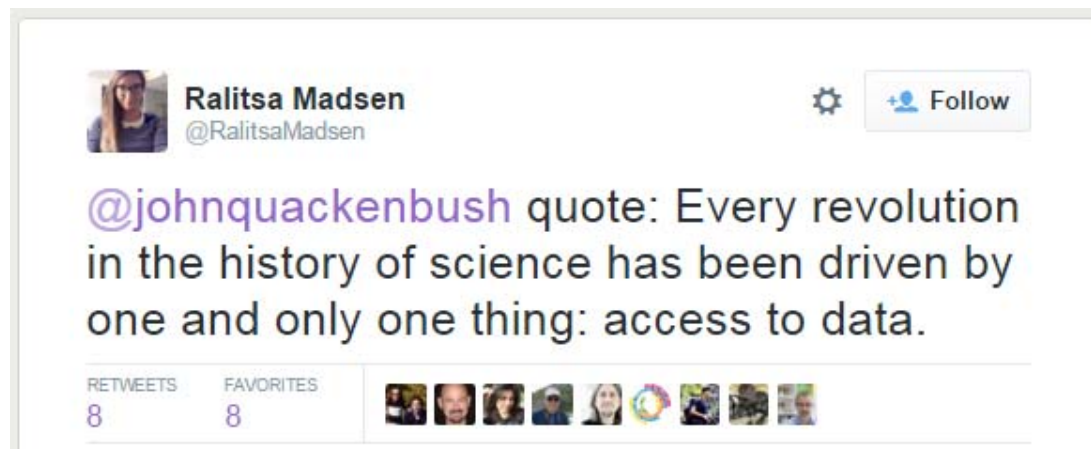
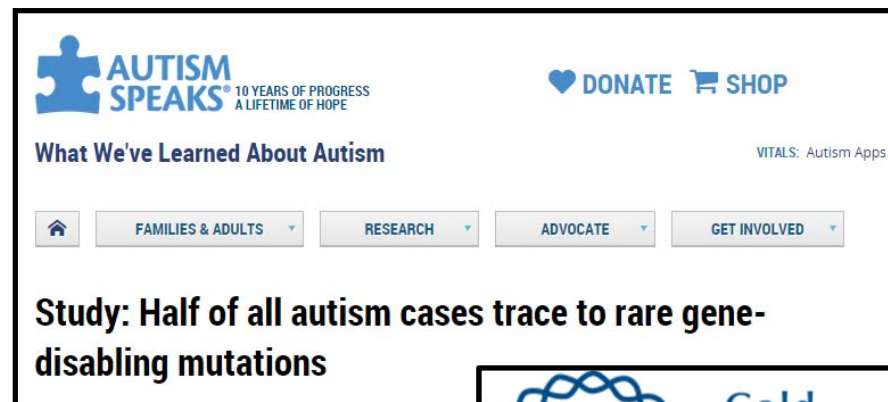
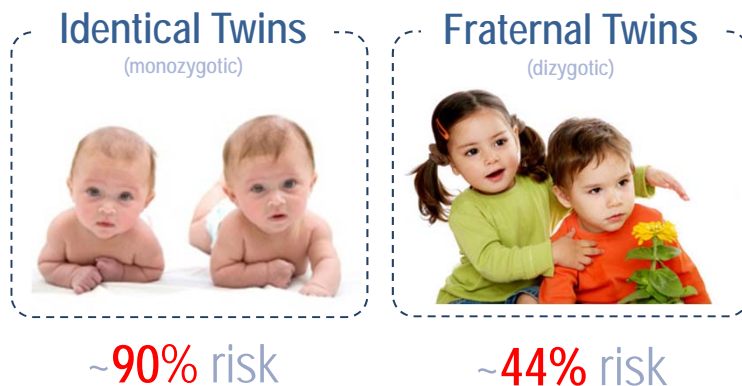
March 31th, 2016

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.

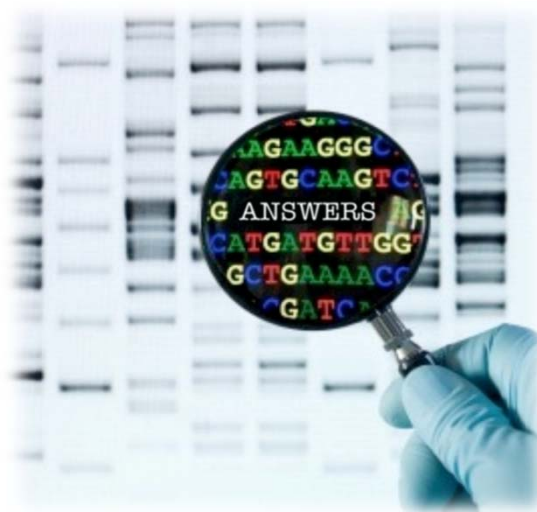


MSSNG – Using open science to deepen understanding of autism



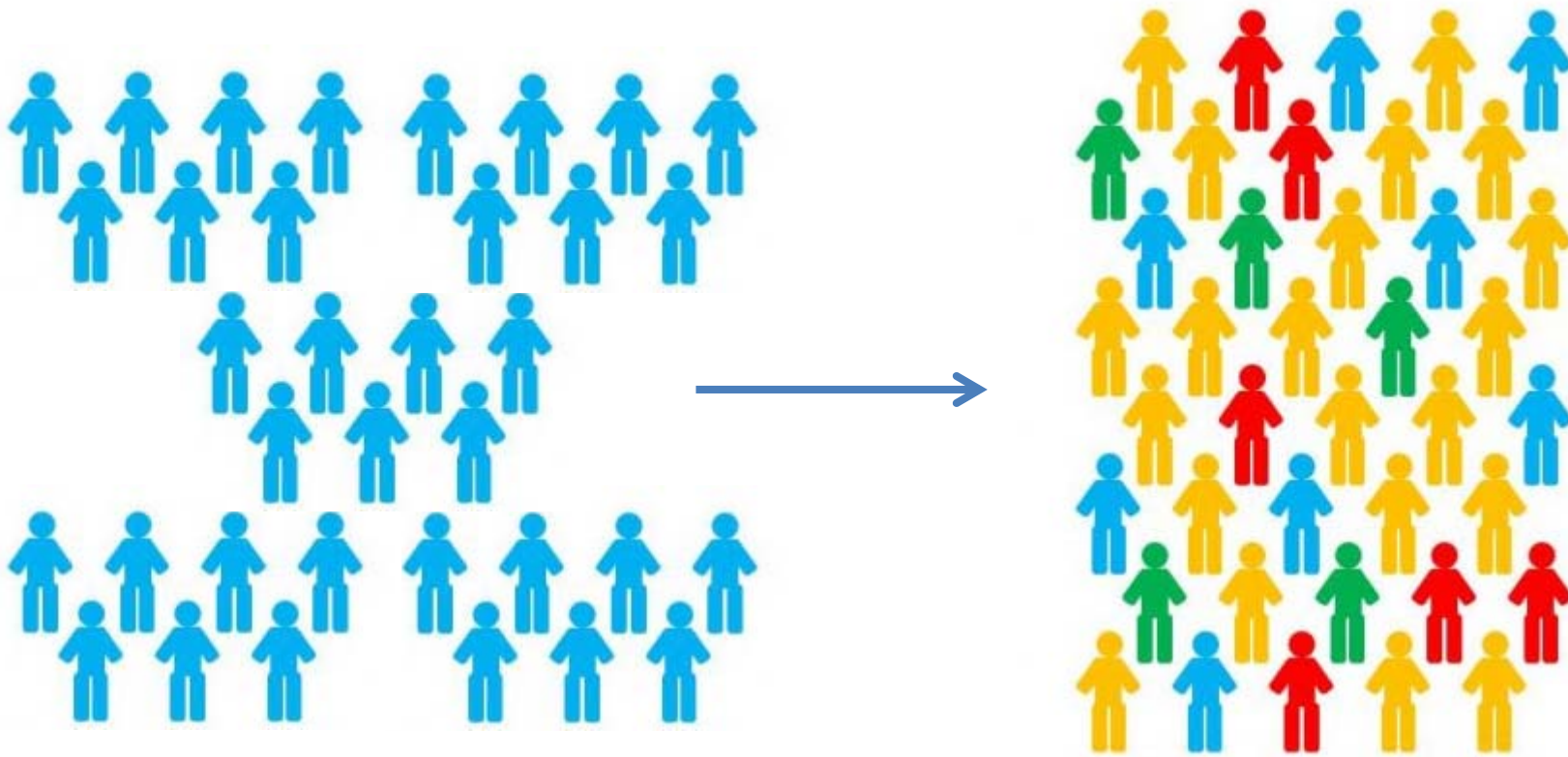
**Private-Public
partnership to improve
understanding and
treatment of autism**

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



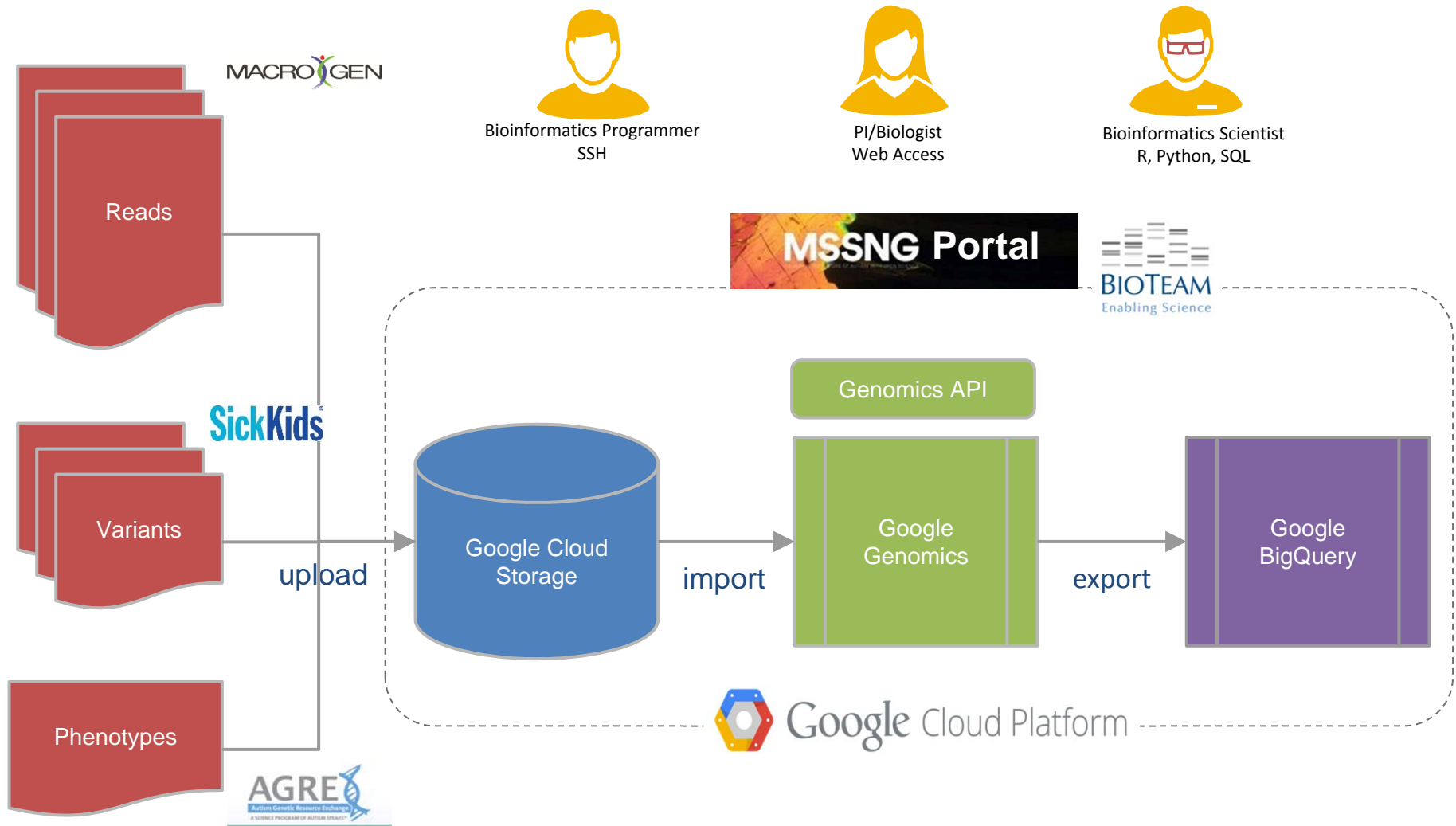
**Data made freely
available through
open-access web-
based portal**

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



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

A policy of controlled access

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](#)

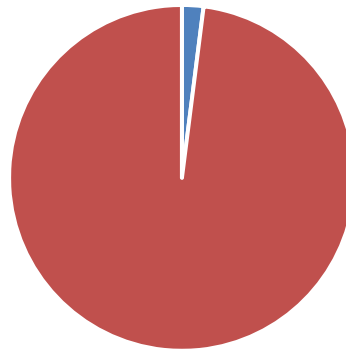
Backup



Looking under the lamppost



Genome



■ Genes ■ Non-Coding

Important findings in the darkness



AJHG

All Content
The American Journal of Human Genetics

Explore Online Now Current Issue Archive Journal Information For Authors

[< Previous Article](#)

Volume 98, Issue 1, p58–74, 7 January 2016

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



MSSNG A PROJECT BY AUTISM SPEAKS
Genetics and Genomics of Autism Research Network

Genes | Gene Lists | Variants | Tools | About

David Glazer ▾ Help

Editing Variant Search: SHANK1 frameshift

Variant Search: SHANK1 frameshift

Selected Variant(s)

By Format

By Genomic Location

By File

By Gene

Symbol
[x] SHANK1

File

By Sample(s)

By Subject(s)

By Significance

Quality Variants

De Novo

Frequency

Quality

Frequency Operator

or

Zygosity

Zygosity

Effects

[x] Frameshift

Start Date

Splice Site

LOF

Missense

QTL

Splicing Reg. Reg

Splicing Reg. Pos

Cancel Update Variant Search

MSSNG A PROJECT BY AUTISM SPEAKS
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Variant Search: SHANK1 frameshift

Symbol: SHANK1

Chromosome: chr11

Start Position: 51,170,827

End Position: 51,170,827

UCSC: chr11:51,170,827-51,170,827

CCV: chr11:51,170,827-51,170,827

Quality Variants: chr11:51,170,827-51,170,827

Frequency: chr11:51,170,827-51,170,827

or

or

Show 10 variants

Sample	Call Set ID	Annotation	Effects with Impact	Pathogenicity	Chromosome	Start Position
10-002-001	140097935/1120862752-650	19-51370827-51170827-650	Frameshift High, 1-CP-High		10	51170827
10-002-002	140097935/1120862752-661	19-51370827-51170827-661	Frameshift High, 1-CP-High		10	51170860
10-002-002	140097935/1120862752-681	19-51370827-51170827-681	Frameshift High, 1-CP-High		10	51207089

Showing 1 to 3 of 3 entries

Click variant Search Click variant Search

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Subject Sample: [REDACTED]

SUBMITTED: [REDACTED]

SNAGSOURCE: [REDACTED]

PLATFORM: [REDACTED]

READGROUPSETID: [REDACTED]

Show 10 variants

Relevance	Time	Question	Value	Legend
High		Client ID number		
High		Date completed		
Low	Timeline Autism Scale-20: 1992	Age at completion (months)		
Low	Timeline Autism Scale-20: 1992	Q1		
Low	Timeline Autism Scale-20: 1992	Q2		
Low	Timeline Autism Scale-20: 1992	Q3		
Low	Timeline Autism Scale-20: 1992	Q4		
Low	Timeline Autism Scale-20: 1992	Q5		
Low	Timeline Autism Scale-20: 1992	Q6		
Low	Timeline Autism Scale-20: 1992	Q7		
Low	Timeline Autism Scale-20: 1992	Q8		
Low	Timeline Autism Scale-20: 1992	Q9		
Low	Timeline Autism Scale-20: 1992	Q10		

Showing 1 to 10 of 10 entries

Subject Sample: [REDACTED]

chr11:51,707,041-51,707,041

chr11

10-002-001

10-002-002

10-002-003

10-002-004

10-002-005

10-002-006

10-002-007

10-002-008

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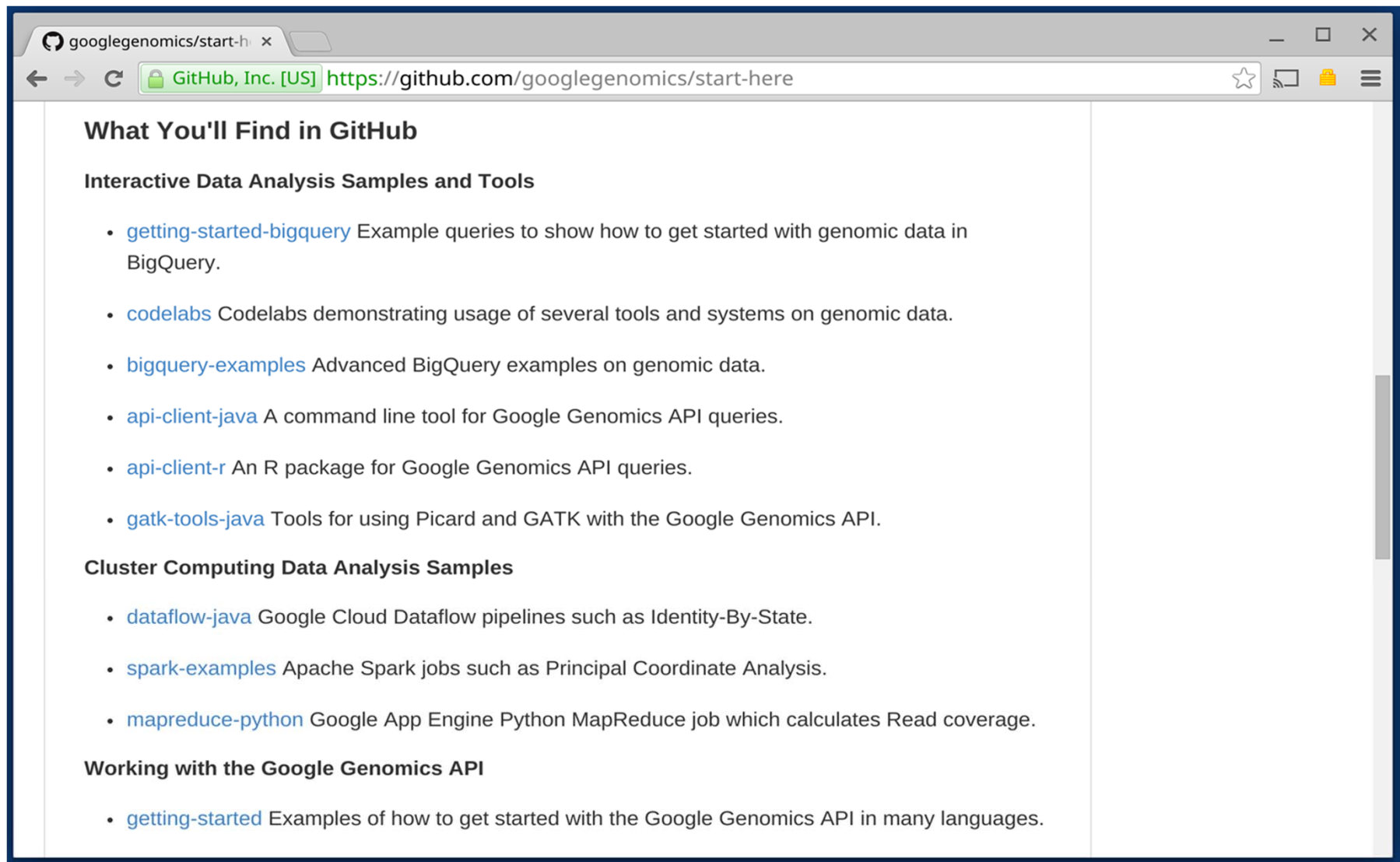
10-002-689

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10-00

Google Genomics on GitHub

A screenshot of a web browser displaying the GitHub repository page for Google Genomics. The browser's address bar shows the URL "https://github.com/googlegenomics/start-here". The page content is organized into sections with headings and bulleted lists of links to various tools and examples.

What You'll Find in GitHub

Interactive Data Analysis Samples and Tools

- [getting-started-bigquery](#) Example queries to show how to get started with genomic data in BigQuery.
- [codelabs](#) Codelabs demonstrating usage of several tools and systems on genomic data.
- [bigquery-examples](#) Advanced BigQuery examples on genomic data.
- [api-client-java](#) A command line tool for Google Genomics API queries.
- [api-client-r](#) An R package for Google Genomics API queries.
- [gatk-tools-java](#) Tools for using Picard and GATK with the Google Genomics API.

Cluster Computing Data Analysis Samples

- [dataflow-java](#) Google Cloud Dataflow pipelines such as Identity-By-State.
- [spark-examples](#) Apache Spark jobs such as Principal Coordinate Analysis.
- [mapreduce-python](#) Google App Engine Python MapReduce job which calculates Read coverage.

Working with the Google Genomics API

- [getting-started](#) Examples of how to get started with the Google Genomics API in many languages.

Genetic diagnosis provides certainty and enables action



Why it matters

