MSSNG - Researcher README

The MSSNG project makes data available to trusted researchers with the goal of improving our understanding of Autism Spectrum Disorder (ASD). An associated publication can be found at https://pubmed.ncbi.nlm.nih.gov/36368308

The purpose of this document is to provide an overview of the available data and associated tools, as well as basic examples of using the tools to access the data.

Notable updates to the data or portal can be found in the CHANGELOG.

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Data

Overview

Data are available for 13790 individuals (13837 genome samples[1]), including:

- 6002 affected individuals (4761 males, 1241 females)
- 7466 unaffected individuals (3647 males, 3819 females)
- 252 autism-related affected individuals (121 males, 131 females)
- 70 unknown affection individuals (31 males, 39 females)

[1]A few individuals were sequenced more than once.

Individuals typically belong to family trios (two parents and one affected child) or quads (two parents and two affected children). A few other family structures are also present. A total of 5069 families are available.

Family members	Families	Individuals
1	1305	1305
2	277	554
3	2329	6987
4	910	3640
5	202	1010
6	34	204
7	9	63
8	1	8
9	1	9
10	1	10

This provides, in summary:

Genome samples	Individuals	Affected individuals	Autism-related affected individuals	Unaffected individuals	Unknown affection individuals	Sequencing technology
1733	1727	726	9	992	0	Complete Genomics
8768	8766	4137	17	4542	70	Illumina HiSeqX

588	588	196	0	392	0	Illumina HiSeq2000
13	13	4	1	8	0	Illumina HiSeq2500
1081	1081	309	124	648	0	Illumina HiSeq
1654	1654	645	101	908	0	Illumina NovaSeq

A summary of the DNA source of the samples is as follows:

DNA source	Genome samples
Blood	11849
Cell line	362
Saliva	161
White blood cell	381
Unknown	1084

Types of data

The following types of data for these individuals are available:

- Sample/subject data
- Aligned reads
- Variants

Sample/subject data

Sample/subject data are divided into three tables: `subject`, `measures`, and `subject_sample`. These tables are available as BigQuery tables (`idyllic-analyst-574:db7_release` dataset); see the MSSNG Data Locations sub-section and the Examples section for how to access and query BigQuery tables.

subject

The `subject` table provides basic information about each individual, such as sex, date of birth, and whether they are affected:

Field	Description
INDEXID	Unique identifier for the individual
FATHERID	Identifier of the individual's father
MOTHERID	Identifier of the individual's mother
AFFECTION	"1" if unaffected, "2" if affected, "0" if autism-related affected, "9" if unknown
SEX	"M" (male), "F" (female)
FAMILYID	Family identifier
FAMILYTYPE	"SPX" (simplex), "MPX" (multiplex)
DOB	Date of birth; yyyy-mm-dd (if information available). Day set to "01" for anonymization.

subject_sample

The `subject_sample` table provides metadata about all genome samples available in the MSSNG database. Subjects may have multiple samples, and each will be referenced as a separate row in the `subject_sample` table. `SUBMITTEDID` is the genome sample identifier that you should use to join subject/sample data to the variant data '`call.name`' field.

Field	Description
SUBMITTEDID	Unique identifier for the genome sample. Note that while this value is usually the same as the INDEXID, that is not always the case. This corresponds to 'call.name' in the variant tables.
INDEXID	Unique identifier of the individual found in the `subject` table
DNASOURCE	Biological sample type used as DNA source: "Blood" (fresh blood), "White blood cell" (frozen as opposed to fresh white blood cells), "Cell line" (lymphoblastoid cell line), "Saliva"

PLATFORM	Sequencing platform: "Illumina HiSeq" (HiSeq2000), “Illumina HiSeq2500”, “Illumina HiSeqX”, "Complete Genomics" (different pipeline versions),”Illumina NovaSeq”
NIMHID	NIMH identifier
RUDCRID	Rutgers repository identifier
AFFECTIONCOMMENTS	Any specific comments regarding a sample
SOFTWARE_VERSION	For Complete Genomics samples only, the software version used to analyse sample
PREDICTED_ANCESTRY	Predicted ancestry of sample. Consensus of computationally derived predictions from two tools.
father_SUBMITTEDID	Unique identifier of the father’s genome sample, if it exists in the dataset and has been sequenced on the same platform as the genome sample
mother_SUBMITTEDID	Unique identifier of the mother’s genome sample, if it exists in the dataset and has been sequenced on the same platform as the genome sample
sample_QC	“ok”: sample passes all QC;”QC_FAIL”: sample failed multiple QC procedures; “CNV_QC_FAIL”: sample failed CNV QC; “CNV-SV_QC_FAIL “: sample failed CNV and SV QC; “SV_QC_FAIL “: sample failed SV QC; “DN_SNP_QC_FAIL”: sample failed de novo small variant QC; “DN_SNP-CNV-SV_QC_FAIL ”: sample failed de novo CNV and CNV and SV QC
Exclude	Samples to exclude from analyses. “YES” value denotes exclusion
Comments	Comments relating to whether a sample is a replicate, twin, or other important things to note

measures

Psychometric test results are typically available only for affected individuals and use established scales. Subjects are identified by `INDEXID`. Test results are linked to the date on which the tests were run (TESTDATE). For a subset of subjects, measurements for the same test performed on different dates are available and need to be collapsed if used for analysis. Please see this spreadsheet for a more detailed explanation of the measures available. The table is organized in tidy format (many records per subject)

Field	Description

INDEXID	Unique identifier for the individual
CODE	Identifier for the type of test
TESTDATE	Date on which the test was administered
MEASURE	Test result

Aligned reads

In this MSSNG database release, alignments in CRAM file format are available for 12,104 samples sequenced on Illumina platforms and aligned to the GRCh38 human reference assembly. For further information about the alignment pipeline for MSSNG Illumina samples, please read this document. For Complete Genomics samples, information about liftover and post-processing of variants can be found in this document.

CRAM and VCF files are available to researchers by following the Process for Researchers to Access MSSNG CRAM and VCF files.

Variants

An individual sample's variants can be found in BigQuery tables.

The `variants_ilmn_chr*` tables contain jointly genotyped variants for all samples sequenced on Illumina platforms. The `variants_cg_chr*` tables contain individually genotyped variants for all samples sequenced by Complete Genomics. In order to improve query performance and data organization, BigQuery's table clustering capabilities have been employed. Column descriptions are available by clicking on the table in BigQuery and viewing the schema.

For representing insertions and deletions, we follow the VCF convention of capturing the first reference base before the insertion or deletion within the variant locus; for example, a deletion of T would be represented by reference_bases = AT and alternate_bases = A.

Variants are available for all 13,837 Illumina and Complete Genomics genome samples. For further information about the variant calling pipeline for MSSNG Illumina samples, please read this document. Complete Genomics variant calls are processed using a custom pipeline

to liftover calls generated by Complete Genomics to GRCh38. More information can be found here.

When performing a variant query within the MSSNG Portal, a number of columns are viewable, some by default and some by modifying the column visibility. The following is a description of each field that is available in the variant query tab of the MSSNG Portal. These can be toggled by using the “Show/hide columns” feature.

Displayed by default?	Field	Description
yes	Sample	Sample name
yes	Sequencing platform	NGS platform; Illumina or Complete Genomics
yes	Sex	Sex of the sample
no	Family ID	Family ID
no	Sanger Validated	Whether confirmed by Sanger sequencing (Yes/No)
no	Sanger Inheritance	Inheritance by Sanger sequencing
yes	Chr	Chromosome (autosomes 1-22 and sex chromosomes X, Y)
yes	Start	Start position (0-positional system)
yes	End	End position
yes	Reference allele, Alternate allele	The reference allele and alternate allele(s) observed for this variant and represented in forward strand. For insertions, the alternate allele includes the inserted sequence as well as the base preceding the

		insertion. For deletions, the alternate allele is the base before the deletion.
yes	Zygosity	Heterozygous, homozygous, hemizygous
yes	Genotype	Genotype, represented as “reference allele, alternate allele” or “alternate allele, alternate allele” or “alternate allele, del/ins/sub”
yes	De Novo	‘High-confidence’ if this variant is a high-confidence rare *de novo* variant
yes	Inheritance	string in the format “0,0:0,1:0,1:ref-alt|mat-pat”, where “0,0:0,1:0,1” is a colon-separated list of maternal (0,0: homozygous reference), paternal (0,1: heterozygous) and child (0,1: heterozygous) genotypes, and “ref-alt|mat-pat” indicates the inheritance (in this case, the child is heterozygous, where “ref” is maternally inherited and “alt” is paternally inherited).
no	FILTER	Filter status: PASS if this position has passed all variant-quality filters
yes	Read depth	Depth of coverage
yes	Allelic depth	Reference and alternate allele counts (comma separated)
yes	Genotype quality	Variant Confidence or Quality by Depth represents the Phred-scaled confidence that the genotype assignment is correct
no	Call.EHQ*	(Complete Genomics only) Calibrated haplotype quality based on equal allele fraction assumption
no	Call.HQ	(Complete Genomics only) Haplotype quality

yes	Max frequency 1000 Genomes	Maximum allele frequency from 1000 Genomes dataset. Dataset includes five populations (European, Admixed American, East Asian, South Asian, African American)
yes	Max frequency GnomAD genome	Maximum filtering allele frequency from the Genome Aggregation Database (gnomAD) for whole-genome data For further information see: https://gnomad.broadinstitute.org/help/faf
no	Max frequency GnomAD exome	Maximum filtering allele frequency from Genome Aggregation Database (gnomAD) for exome data
no	Max Frequency	Maximum of filtering allele frequencies from both gnomAD datasets and allele frequencies from Complete Genomics
yes	RefSeq ID	Combined ANNOVAR output on coding sequence mapping and effect; composed of: (a) for coding exonic changes (typeseq “exonic”): official gene symbol, RefSeq transcript isoform ID, position in the coding sequence, amino acid change; (b) for core splice site changes (typeseq "exonic"): official gene symbol, RefSeq transcript isoform ID, exon number, coding sequence position and change

no	Typeseq priority	Type of sequence overlapped with respect to known genes/transcripts and their coding/noncoding status: (a) “exonic” represents coding exons, (b) “exonic;splicing” represents exonic and splicing overlaps (for multi-transcript genes), (c) “splicing” represents core splicing site (2 bp on the intron side of intron-exon and exon-intron junctions), (d) "ncRNA_exonic" represents exons of non-coding RNA genes, (e) "ncRNA_splicing" represents core splicing sites of non-coding RNA genes, (f) “UTR5” represents 5’ untranslated region, (g) “UTR3” represents 3’ untranslated region, (h) “upstream” represents 1 kb upstream of transcription start site (TSS), (i) “downstream” represents 1 kb downstream of TSS, and (j) “intergenic” represents intergenic regions beyond the upstream/downstream threshold (1 kb). For variants with multiple sequence overlaps (for example, exonic for one transcript and intronic for other), we used the ANNOVAR prioritization scheme to prioritize them. (http://annovar.openbioinformatics.org/en/latest/user-guide/gene/).
no	Effect priority	Type of effect on the coding sequence: (a) “synonymous SNV”, (b) "nonsynonymous SNV", (c) "stopgain SNV", (d) "frameshift deletion", (e) "frameshift insertion", (f) "frameshift substitution", (g) "nonframeshift deletion", (h) "nonframeshift insertion", (i) "nonframeshift substitution" , (j) “stoploss SNV”. Prioritized effect is selected for variants with multiple effects (http://annovar.openbioinformatics.org/en/latest/user-guide/gene/).
yes	Gene Symbol	Official gene symbol
no	Entrez Id	Entrez-gene ID
no	OMIM Phenotype	OMIM disorder/disease description when available for the corresponding OMIM gene accession

no	CGD disease	The Clinical Genomics Database (CGD) is compiled by curators and maintained by the National Human Genome Research Institute (NHGRI); for every gene in the database, the CGD provides a list of one or more genetic disorders and a mode of inheritance; this field reports the genetic disorder(s)
no	CGD inheritance	This field reports the CGD mode of inheritance (AD, AR, AD/AR, XL, more complex modes). Since the CGD mode of inheritance is directly added by a curator and is tied to specific genetic disorder(s), it could be considered more accurate than the mode of inheritance for top-level HPO phenotypes.
no	Comment	Annotation database issues (for example, ambiguous liftover or incomplete ORF for gene transcript)
no	gnomAD_lof_oe_ci_upper	gnomadV4 constraint: LOEUF - upper bound of 90% confidence interval for o/e ratio for high confidence pLoF variants (lower values indicate more constrained)
no	gnomAD_mis_oe_ci_upper	gnomadV4 constraint: LOEUF - upper bound of 90% confidence interval for o/e ratio for high confidence pLoF variants (lower values indicate more constrained)
no	gnomAD_lof_pLI	GnomAD pLI score
no	gnomAD_lof_pRec	GnomAD pRec score
yes	ClinVar significance	Overall ClinVar significance code; “pathogenic” is the code of interest for rare disorders (https://www.ncbi.nlm.nih.gov/clinvar/docs/clinsig/)

yes	ClinVar Significance Simple	Expected values 0,1 or -1; 0 = no current value of pathogenic; 1 = at least one record submitted with pathogenic/likely pathogenic; -1 = no values for clinical significance at all for this variant or set of variants. Used for the "included" variants that are only in ClinVar because they are included in a haplotype or genotype with an interpretation
yes	Effect - Impact	Pre-computed effects and impacts for the variant. This was computed using various annotation factors such as sequence overlap, coding effect, prediction scores, etc.
no	Affection	0 = Autism-related affected, 1 = unaffected, 2 = affected
no	dbSNP ID	dbSNP identifier
yes	Sample QC	replace with - “ok”: sample passes all QC;”QC_FAIL”: sample failed multiple QC procedures; “CNV_QC_FAIL”: sample failed CNV QC; “CNV-SV_QC_FAIL “: sample failed CNV and SV QC; “SV_QC_FAIL “: sample failed SV QC; “DN_SNP_QC_FAIL”: sample failed de novo small variant QC; “DN_SNP-CNV-SV_QC_FAIL ”: sample failed de novo CNV and CNV and SV QC

Copy Number Variants (CNVs)

For samples sequenced on Illumina platforms, copy number variants (CNVs) were detected using ERDS (Zhu et al, 2012) and CNVnator (Abyzov et al, 2011) as previously described (Trost et al, 2018). For CNVnator, calls for which > 50% of the reads in the CNV region had zero mapping quality were removed (q0 filter), except for in homozygous autosomal deletions or X-linked deletions in males (with normalized read depth (NRD) < 0.03). For samples sequenced by Complete Genomics (CG), CNV calls were used as provided, with all CNVs being >= 2 kb. Quality and frequency of CNVs were defined as follows:

- **High-quality CNVs** for Illumina samples were defined as those that are >=1 kb, are detected by both ERDS and CNVnator with >= 50% reciprocal overlap, and have <= 70% overlap with telomeres, centromeres, and segmental duplications (“Unclean genome

overlap" column below). High-quality CNVs for Complete Genomics samples were defined as those with <= 70% overlap with telomeres, centromeres, and segmental duplications.

- **Rare CNVs** were defined as those detected at <=1% frequency in the QC-passing parental samples in MSSNG according to ERDS, CNVnator, and Complete Genomics, as well as <=1% frequency according to both ERDS and CNVnator in QC-passing samples from 2,504 unrelated individuals from the 1000 Genomes Project sequenced on the Illumina NovaSeq platform.

A CNV was tagged as "high quality rare" (see "High quality rare" column below) if it was both high-quality and rare according to the criteria defined above. The table below describes the columns found when querying the CNV table, as well as noting column visibility. Column visibility can be modified under "Show/hide columns" in the MSSNG Portal website.

Displayed by default?	Column name (BigQuery)	Column name (UI)	Description
yes	sample	Sample	sample identifier
yes	chr	Chr	chromosome
yes	start	Start	start position
yes	end	End	end position
yes	CNV_type	CNV type	CNV type (DEL or DUP)
yes	size	Size	size of CNV in base pairs
yes	overlap	Overlap	overlap between CNV calls (Illumina samples only; format is overlap of ERDS call with CNVnator call | overlap of CNVnator call with ERDS calls)
no	copy_number	Copy number	estimated copy number (for Illumina, the copy number reported by ERDS is given)

no	GC_content_percent	GC content (%)	% GC content
no	cytoband	Cytoband	cytoband
yes	gene_symbol	Gene symbol	official gene symbols with transcript overlap
no	gene_entrez_id	Gene ID	Entrez gene IDs with transcript overlap
yes	exon_symbol	Exon symbol	official gene symbols with exon overlap
no	exon_entrez_id	Exon ID	Entrez gene IDs with exon overlap
no	CDS_symbol	CDS symbol	Official gene symbols with CDS overlap
no	CDS_entrez_id	CDS ID	Entrez gene IDs with CDS overlap
yes	gnomAD_pLI	GnomAD pLI	gnomAD pLI value
no	gnomAD_oe_lof_upper	gnomAD o/e LoF upper	upper bound of gnomAD observed/expected ratio for loss of function variants
no	RepeatMasker_percent_overlap	RepeatMasker overlap (%)	% overlap with RepeatMasker repeats
yes	unclean_genome_percent_overlap	Unclean genome overlap (%)	% overlap with segmental duplications, centromeres, and telomeres
no	MPO_nervous_system	MPO nervous system	mouse phenotype ontology terms related to nervous system phenotypes
no	HPO_nervous_system	HPO nervous system	human phenotype ontology terms related to nervous system phenotypes
no	CGD_disease_inheritance	CGD disease inheritance	gene symbols with transcript overlap in the Clinical Genomic Database
yes	OMIM_morbid_map	OMIM Morbid Map	OMIM Morbid Map
no	ISCA_region	ISCA region	genomic disease region from the International Standards for Cytogenomic Arrays database

no	CNV_ISCA_percent_overlap	CNV ISCA overlap (%)	% length of CNV overlapped by International Standards for Cytogenomic Arrays region
no	DECIPHER_region	DECIPHER region	genomic disease region from the DECIPHER database
no	CNV_DECIPHER_percent_overlap	CNV DECIPHER overlap (%)	% length of CNV overlapped by DECIPHER region
no	DGV_N_studies	DGV N studies	number of studies in which there is an overlapping CNV in the Database of Genomic Variants (DGV) (50% reciprocal overlap)
no	DGV_percent_freq_subjects_all_studies	DGV freq subjects all studies (%)	DGV frequency, any study (50% reciprocal overlap)
no	DGV_percent_freq_subjects_coverage_studies	DGV freq subjects coverage studies (%)	DGV frequency, only studies with coverage (50% reciprocal overlap)
no	DGV_percent_freq	DGV freq (%)	DGV frequency (50% reciprocal overlap)
yes	CG_percent_freq_MSSNG_parents	CG freq parents (%)	percentage of MSSNG parents sequenced on the Complete Genomics platform in which the CNV is detected (50% reciprocal overlap)
yes	CNVnator_percent_freq_MSSNG_parents_HiSeqX	CNVN freq parents HiSeqX (%)	percentage of MSSNG parents sequenced on Illumina HiSeq X in which the CNV is detected by CNVnator (50% reciprocal overlap)
no	CNVnator_percent_freq_MSSNG_parents_HiSeq2000	CNVN freq parents HiSeq (%)	percentage of MSSNG parents sequenced on Illumina HiSeq 2000/2500 in which the CNV is detected by CNVnator (50% reciprocal overlap)
yes	ERDS_percent_freq_MSSNG_parents_HiSeqX	ERDS freq parents HiSeqX (%)	percentage of MSSNG parents sequenced on Illumina HiSeq X in which the CNV is detected by ERDS (50% reciprocal overlap)

no	ERDS_percent_freq_MSSNG_parents_HiSeq2000	ERDS freq parents HiSeq (%)	percentage of MSSNG parents sequenced on Illumina HiSeq 2000/2500 in which the CNV is detected by ERDS (50% reciprocal overlap)
no	ERDS_percent_freq_1000G	ERDS freq 1000G (%)	percentage of 1000 Genomes Project samples in which the CNV is detected by ERDS (50% reciprocal overlap)
no	CNVnator_percent_freq_1000G	CNVN freq 1000G (%)	percentage of 1000 Genomes Project samples in which the CNV is detected by CNVnator (50% reciprocal overlap)
yes	putative_inheritance	Putative inheritance	putative inheritance - possible values are “Maternal”, “Paternal”, “NA” (no parents available), “Inherited_Ambiguous” (both parents have the CNV), “One_parent_sequenced” (only one parent available), “Ambiguous” (inheritance is ambiguous), and “P_denovo” (putative *de novo* variant)
yes	high_quality_rare	High quality rare	“High quality rare” if the CNV meets the criteria for this described above, or “-” otherwise.
yes	sample_QC	Sample QC	“ok”: sample passes all QC;”QC_FAIL”: sample failed multiple QC procedures; “CNV_QC_FAIL”: sample failed CNV QC; “CNV-SV_QC_FAIL “: sample failed CNV and SV QC; “SV_QC_FAIL “: sample failed SV QC; “DN_SNP_QC_FAIL”: sample failed de novo small variant QC; “DN_SNP-CNV-SV_QC_FAIL ”: sample failed de novo CNV and CNV and SV QC
no	FAMILYID	Family ID	family identifier

yes	platform	Platform	Sequencing platform: "Illumina HiSeq" (HiSeq2000), “Illumina HiSeq2500”, “Illumina HiSeqX”, "Complete Genomics", ”Illumina NovaSeq”

Annotations

The `annotations_ilmn` (Illumina) and `annotations_cg` (Complete Genomics) variant annotations are generated using an ANNOVAR-based custom pipeline, following ANNOVAR priority rules to report variants. For further information on the databases/data sources and versions used to derive annotations, please read this document. Column descriptions are available for both tables by clicking on either one in BigQuery and viewing the schema.

See the example section for how to join this table to the variant table.

Putative *de novo* variants

De novo variants are those called in probands but not in the parents’ genomes. *De novo* variants are available for families for which both parents were sequenced (4795/6002 affected). The `variants_de_novo` table lists all putative *de novo* variants. The variants with annotations can also be viewed as a spreadsheet.

Field	Description
id	As in `annotation` table
reference_name	As in `variants*` table
start	As in `variants*` table
end	As in `variants*` table
reference_bases	As in `variants*` table
alternate_bases	As in `variants*` table
PLATFORM	As in `subject_sample` table
COMMENT	Any comments about this variant
SUBMITTEDID	As in `subject_sample` table

Sanger-validated variants

For the variants in the `variants_sanger` table, Sanger validation results are available; positive as well as negative results are reported. Only a small fraction of variants have undergone Sanger validation.

Field	Description
id	As in `annotations_*` table
reference_name	As in `variants*` table
start	As in `variants*` table
end	As in `variants*` table
reference_bases	As in `variants*` table
alternate_bases	As in `variants*` table
Sanger_validated	Possible values: YES (variant found), NO (variant not found), NULL
Sanger_inheritance	Possible values: *null*, DE NOVO, HEMIZYGOUS, HETEROZYGOUS, HOMOZYGOUS, INHERITED, LIKELY PATERNAL, MATERNAL, NOT MATERNAL, NOT PATERNAL, PATERNAL
PLATFORM	As in `subject_sample` table
SUBMITTEDID	As in `subject_sample` table

MSSNG data locations

MSSNG data are hosted on Google Cloud Platform services as follows:

	Google BigQuery	Google Cloud Storage
Aligned reads		X
Called variants	X	X

Sample/subject data	X	

Google BigQuery is a service designed for storing generic structured data and allowing for querying over massive datasets in seconds. BigQuery supports standard SQL queries, which can be accessed via the BigQuery web-based interface, command line tool, or programmatic API. This allows access to data from any data analysis tool (such as Python) that supports the Google BigQuery API.

Google Cloud Storage is a repository for storing and sharing files. Cloud Storage supports storing and retrieving files using a web-based interface, command-line tool, or programmatic API.

Some data are also available as a direct download via the MSSNG Portal. These include complete subject/sample information as well as annotated *de novo* variants.

Access

The following describes ways to access the MSSNG data stored in the Google Cloud:

- To get started, you can access the MSSNG researcher portal at https://research.mss.ng
- If you would like to issue custom queries against the MSSNG BigQuery tables, then you will need to create a Google Cloud Project. See the instructions below for getting started using BigQuery.
- If you would like to download the CRAM and VCF files, please refer to the document on the process for researchers to access MSSNG CRAM and VCF files.

Once you have created your own Google Cloud project, you can try some of the examples in the next section.

BigQuery Examples

Subject/sample data

Subject/sample and variant data are stored in Google BigQuery. Genomics data in BigQuery is most commonly accessed through the BigQuery web interface. Subject/sample data are also available for download from the MSSNG Portal.

BigQuery web interface

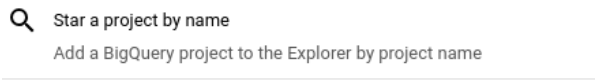
The BigQuery web interface can be used for issuing ad hoc queries over the genomic variant data and subject/sample data.

Setup

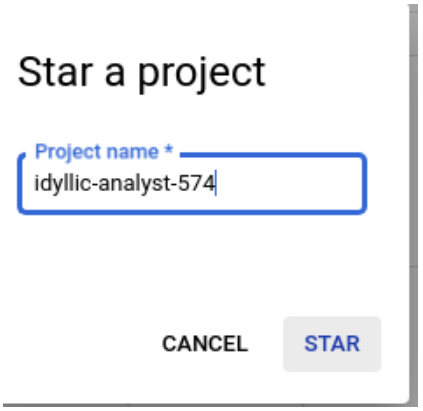
The following steps demonstrate accessing the MSSNG subject/sample data.

1. Go to https://console.cloud.google.com/bigquery
2. The current project shown on the page (at the top, next to the Google Cloud logo), should be your own project, which will be used to run queries. To view the db7_release dataset:
    a. Click the Add button on the explorer pane

    b. Select "Star a project by name"



    c. Enter "idyllic-analyst-574" here



    d. Click on the STAR button. You should now see the project and the db7_release dataset in the Explorer pane.

If you click on the `db7_release` dataset it should expand to show (among many others) the tables:

- `annotations_cg`
- `annotations_ilmn`
- `variants_cg*`
- `variants_ilmn*`
- `subject`
- `subject_sample`
- `variants_de_novo`
- `variants_sanger`

Subject/sample data example

Your first example query will be on the subject table. Clicking on the `subject` table, and then the Query button will open the query pane on the right hand side.

In the query text area enter the query:

```
#standardSQL
SELECT
  sex,
  COUNT(INDEXID) AS count
FROM
  `idyllic-analyst-574.db7_release.subject`
GROUP BY
  sex
ORDER BY
  sex
```

Clicking on the Run Query button should generate results in a few seconds which looks like:

Row	SEX	count	
1	F	5230	
2	M	8560	

To see the number of autism-affected individuals, change the query to:

```
#standardSQL
SELECT
```

```
  sex,
  COUNT (INDEXID) AS count
FROM
  `idyllic-analyst-574.db7_release.subject`
WHERE
  affection = '2'
GROUP BY
  sex
ORDER BY
  sex
```

(note that `affection = '2'` means autism affected)

Clicking on the Run Query button should generate results in a few seconds which looks like:

Row	gender	count	
1	F	1241	
2	M	4761	

Genomic variants examples

Genomic variants are stored in the `db7_release.variants_cg*` and `db7_release.variants_ilmn*` tables (described above). This table uses some features of Google BigQuery not commonly seen in relational databases (which you may already be familiar with), namely Array fields.

Each record in the `variants_*` tables describes a variant that has been called at least once within the set of samples. Within the variant record is a `call` field, which contains a reference to all calls of this variant.

The schema for the `variants_*` tables can be found by:

1. Selecting one of the `variants_*` tables in the left hand pane of the BigQuery interface
2. The Schema button in the right hand pane should be selected by default.

A button for Table Details should also be displayed. Select this to view information such as the size of the table and number of rows. To see a sampling of the data, select the Preview button.

Many example queries that can be used on the `variants_*` tables can be found here.

To build your own, more sophisticated queries, see the BigQuery Query Reference.

References

Zhu, M. et al. Using ERDS to infer copy-number variants in high-coverage genomes. American Journal of Human Genetics 91:408–421 (2012).

Abyzov, A. et al. CNVnator: an approach to discover, genotype, and characterize typical and atypical CNVs from family and population genome sequencing. Genome Research 21:974–984 (2011).

Trost, B. et al. A comprehensive workflow for read depth-based identification of copy-number variation from whole-genome sequence data. American Journal of Human Genetics 4:142-155 (2018).