MSSNG Research Portal Features

Prepared by DNAstack
December 2020

This presentation showcases the features of the MSSNG Research Portal, a project by Autism Speaks.

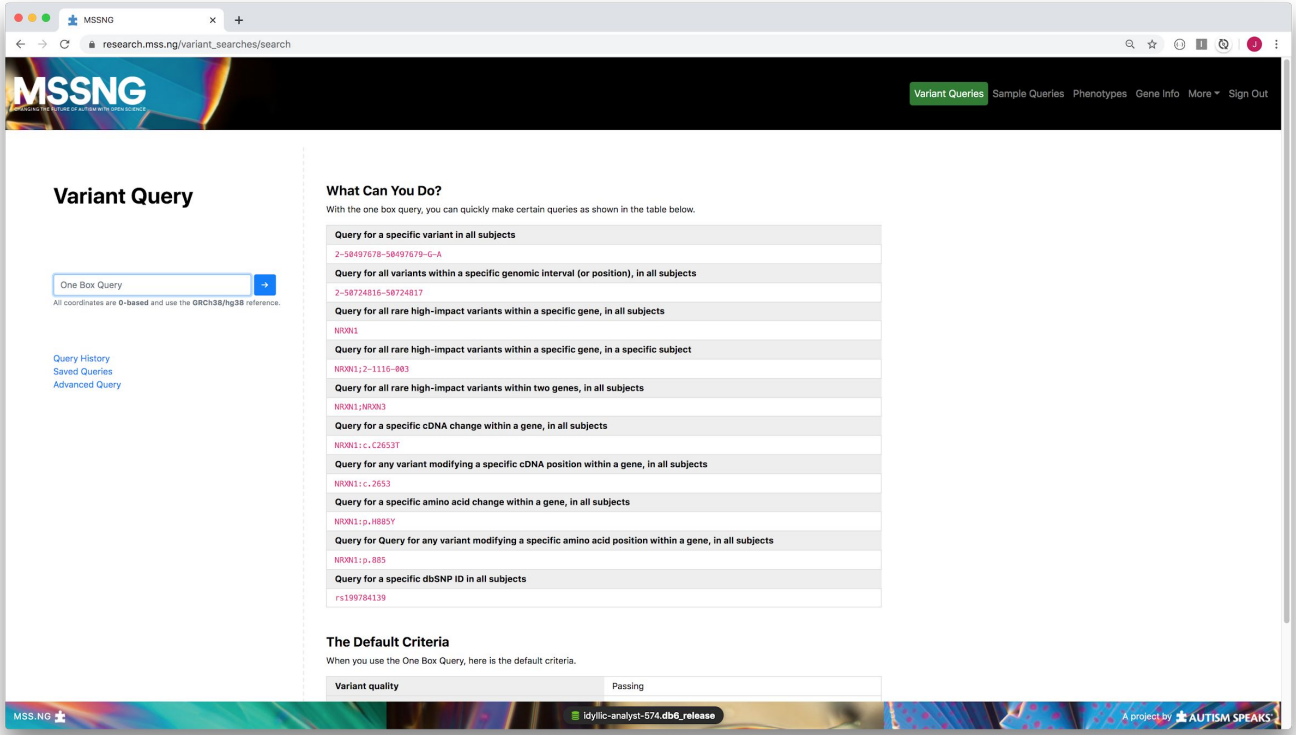
The MSSNG Research Portal is open source under the terms of the Apache License, version 2.0. The source code is available on GitHub.

We hope that this project will be useful beyond the MSSNG dataset, and that this presentation will help you to decide if it could be useful to your project.

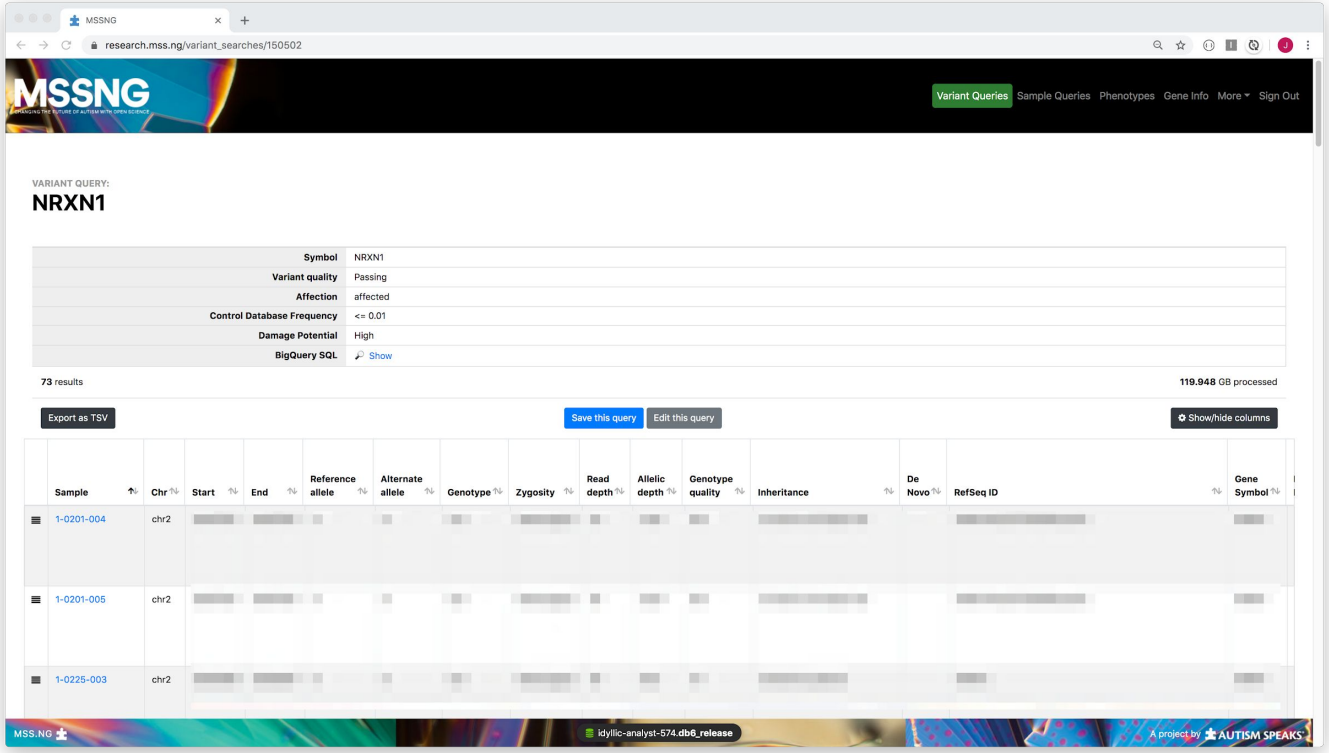
This presentation may be shared freely in its unaltered form.

Variant Queries

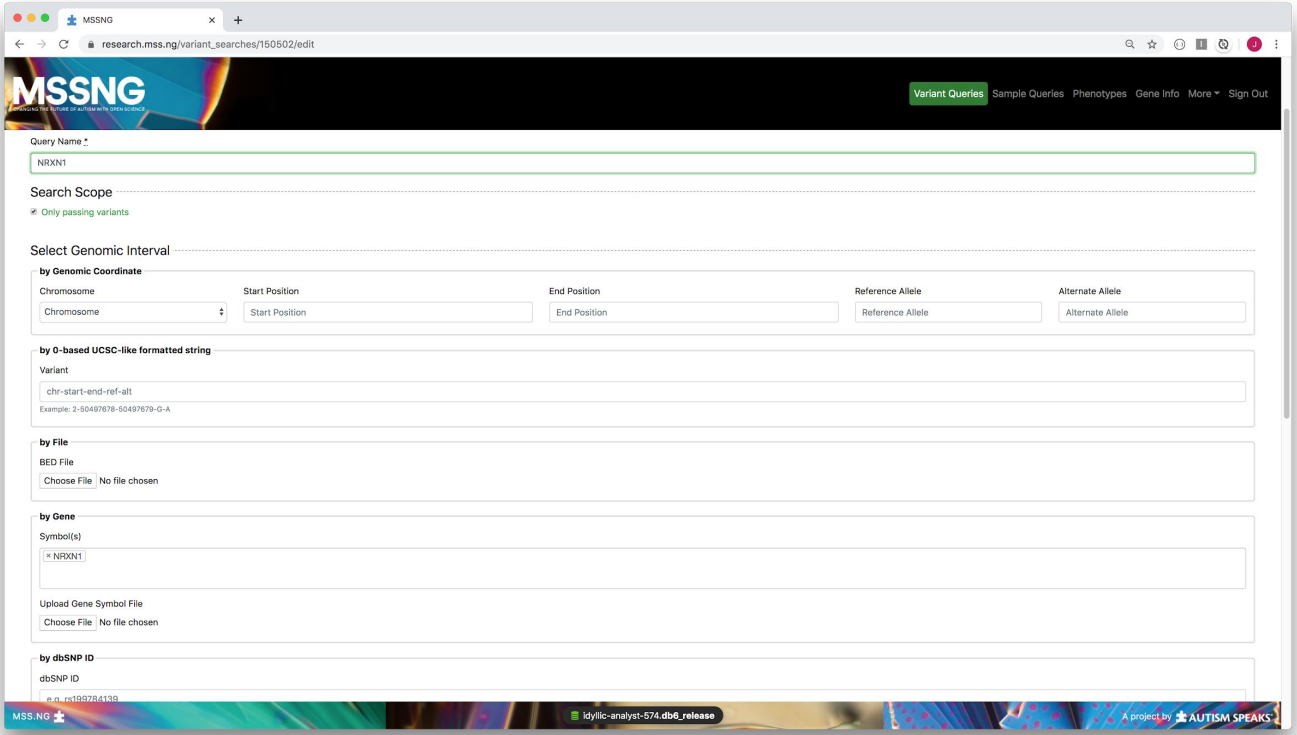
“One Box” Variant Search



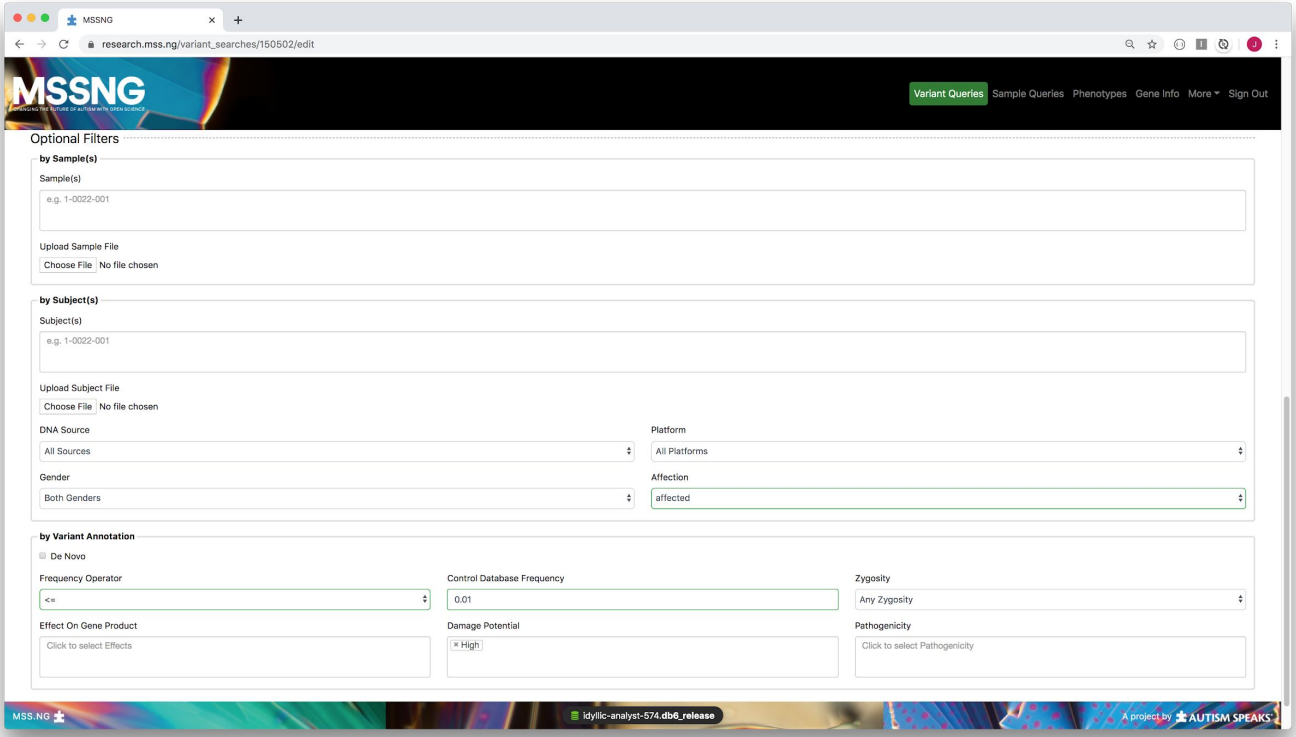
Variant Search Results



Advanced Variant Query Builder & Editor (1/2)

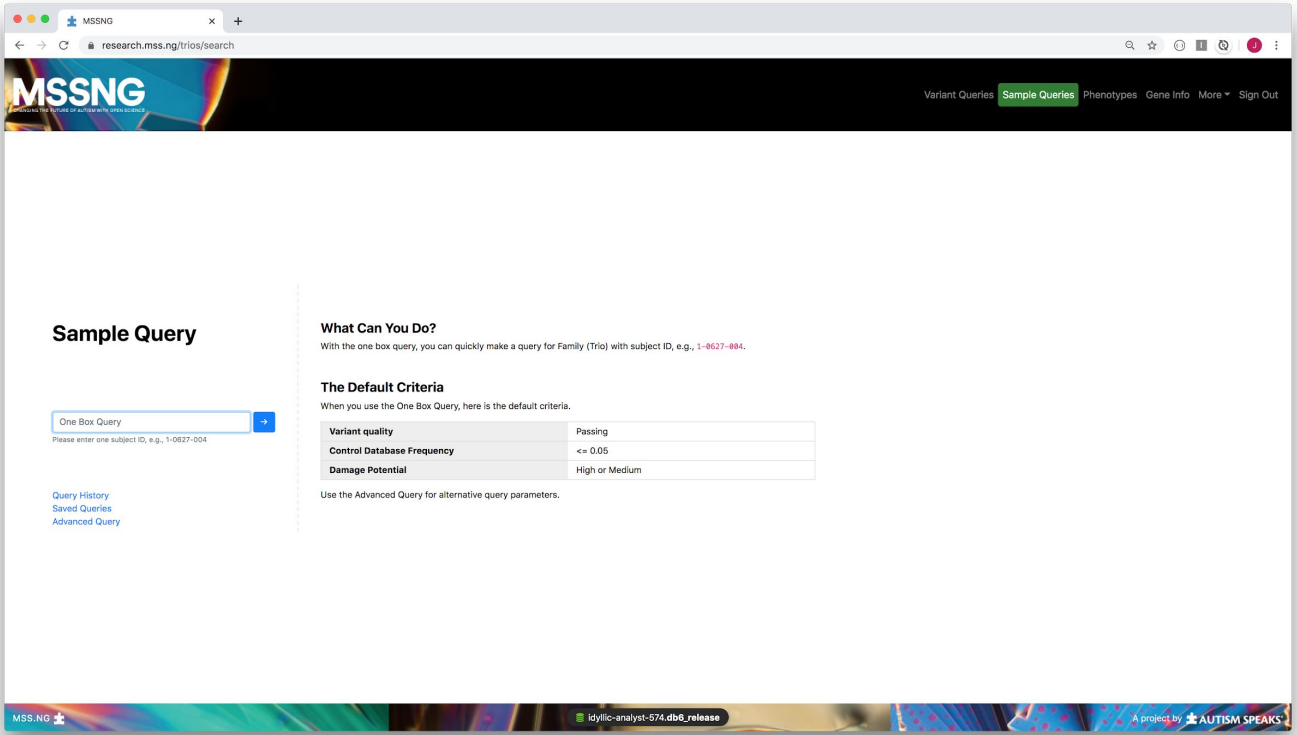


Advanced Variant Query Builder & Editor (2/2)

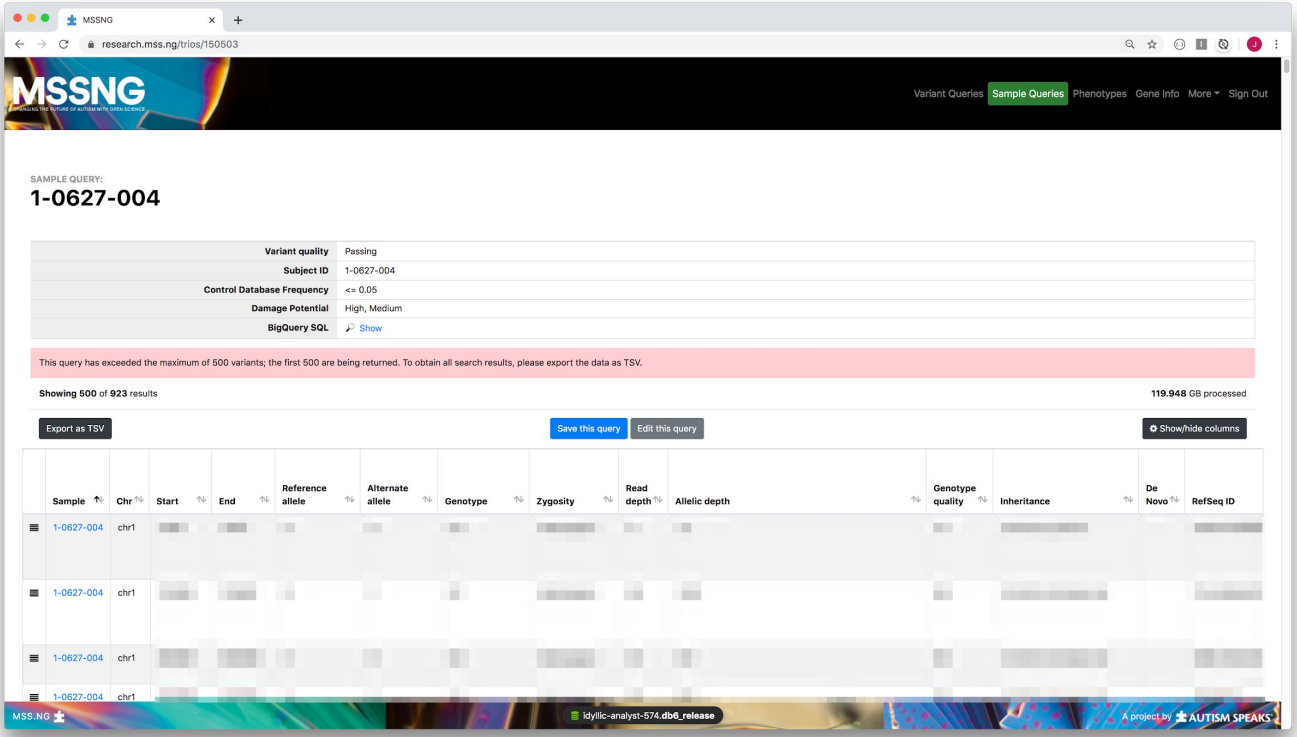


Sample Queries

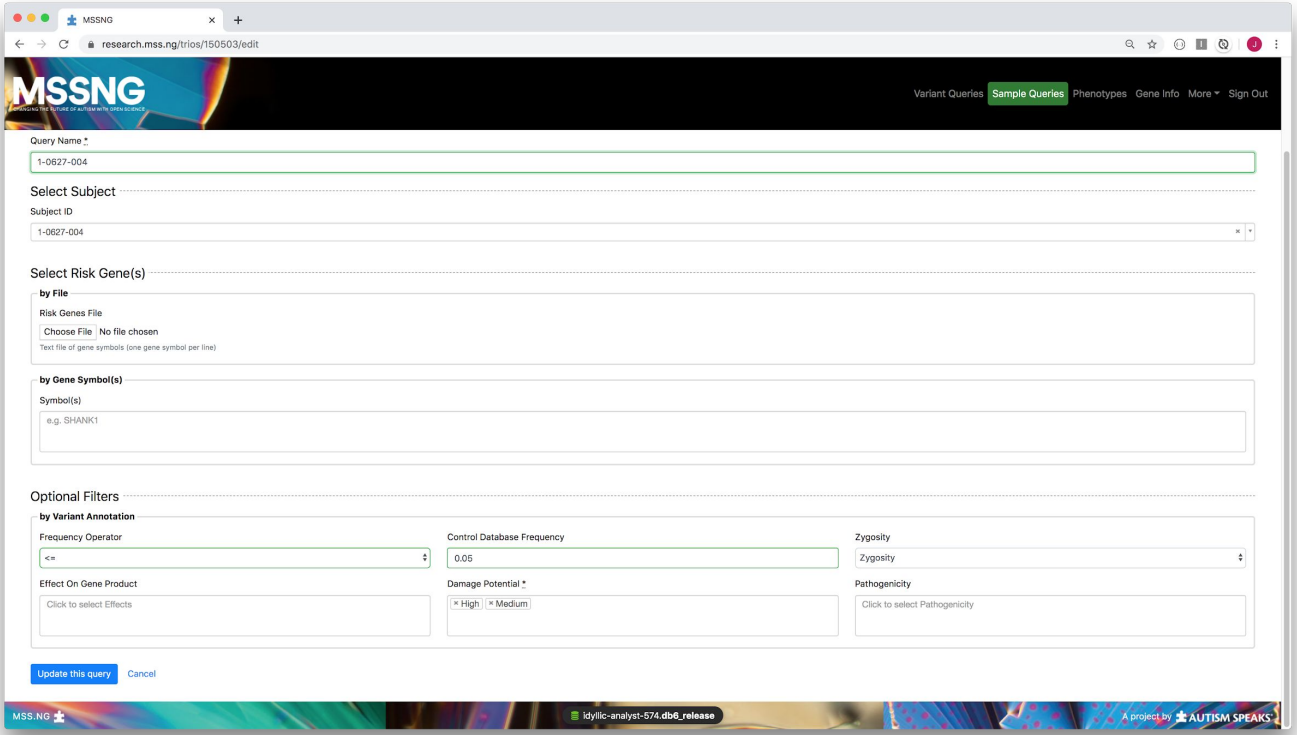
“One Box” Sample Query



Sample Query Results

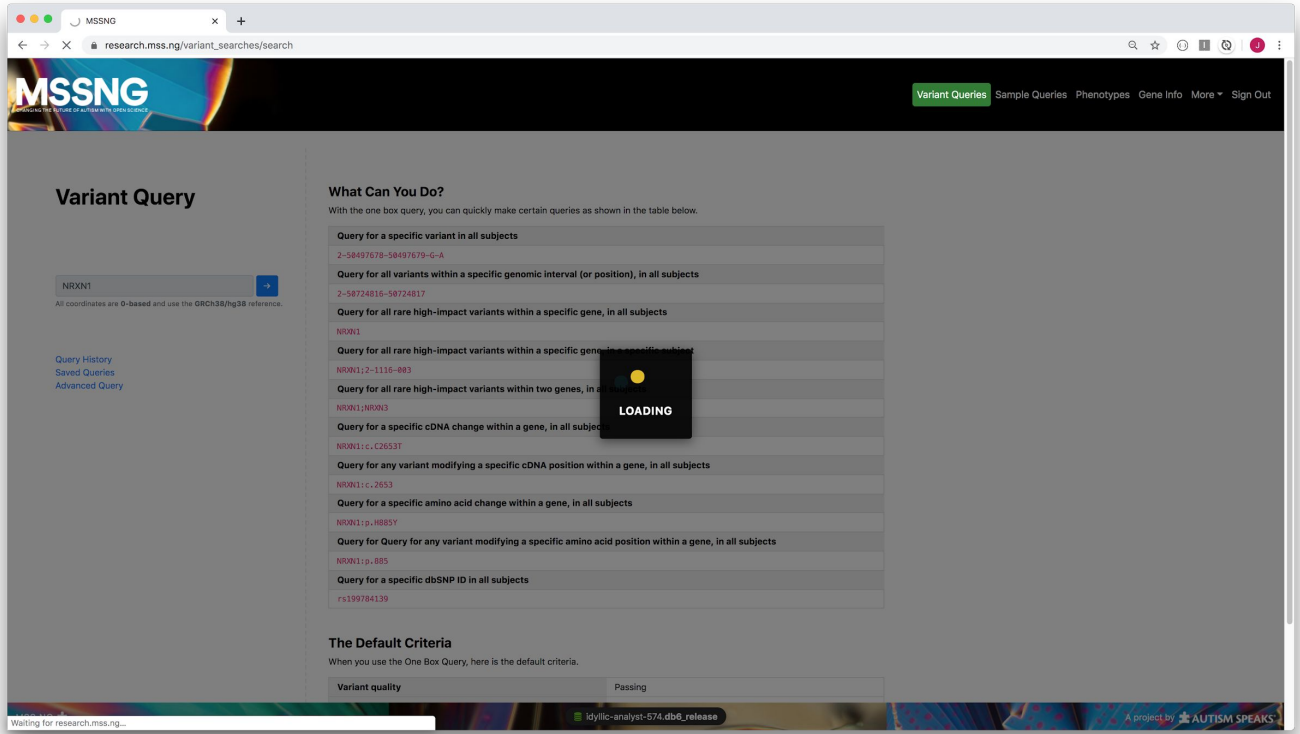


Advanced Sample Query Builder & Editor

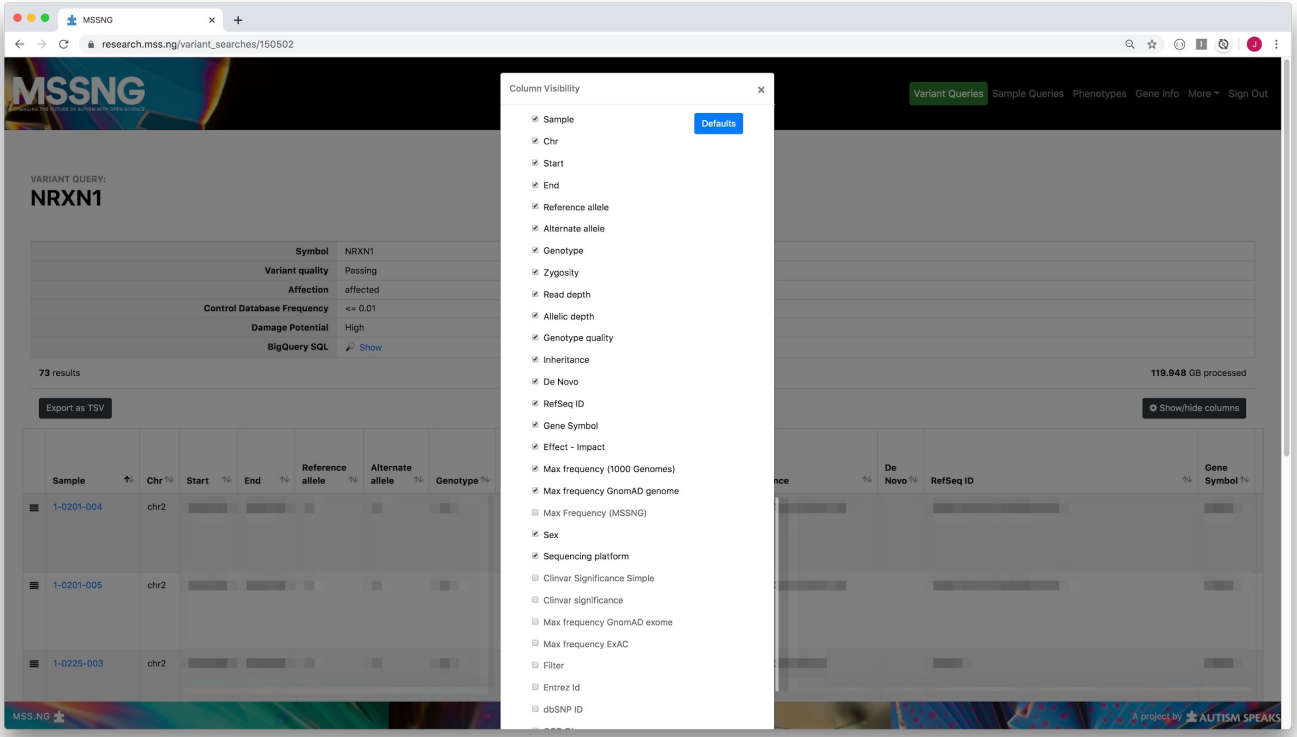


Common to Variant Queries and Sample Queries

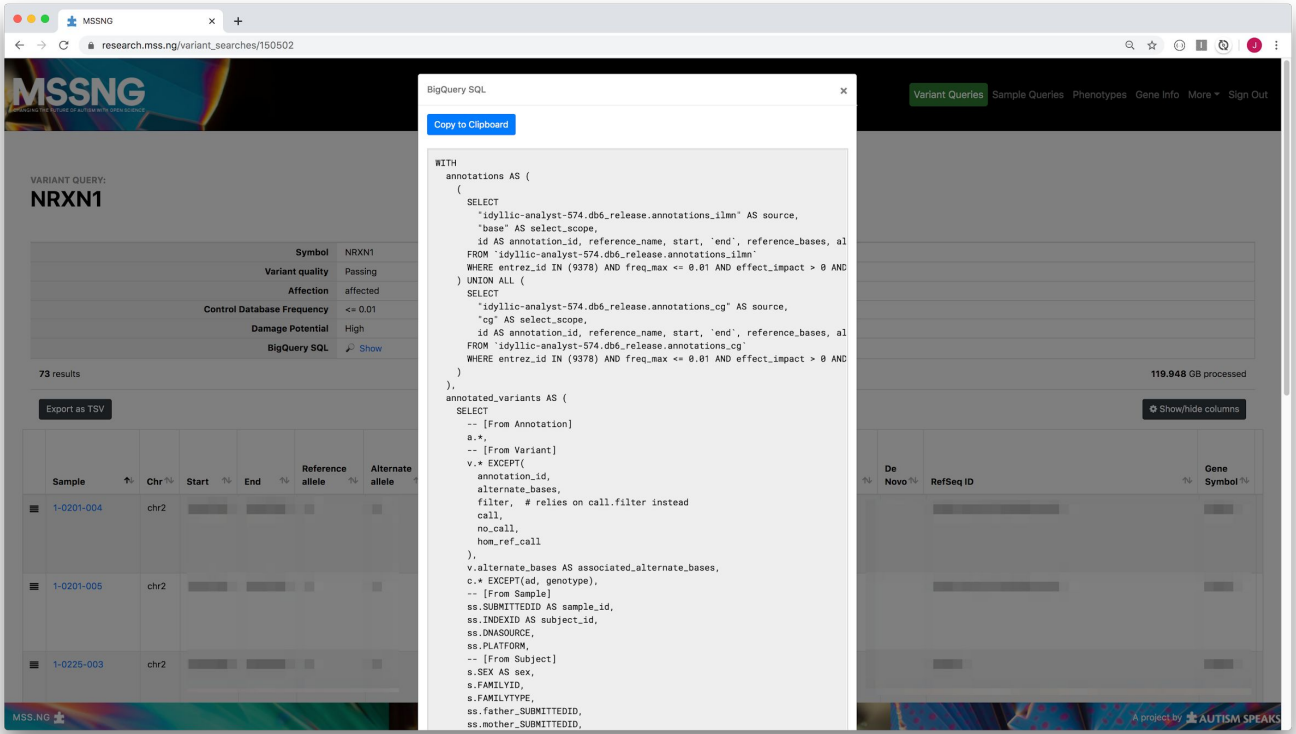
Animated Indicator While Query is Running



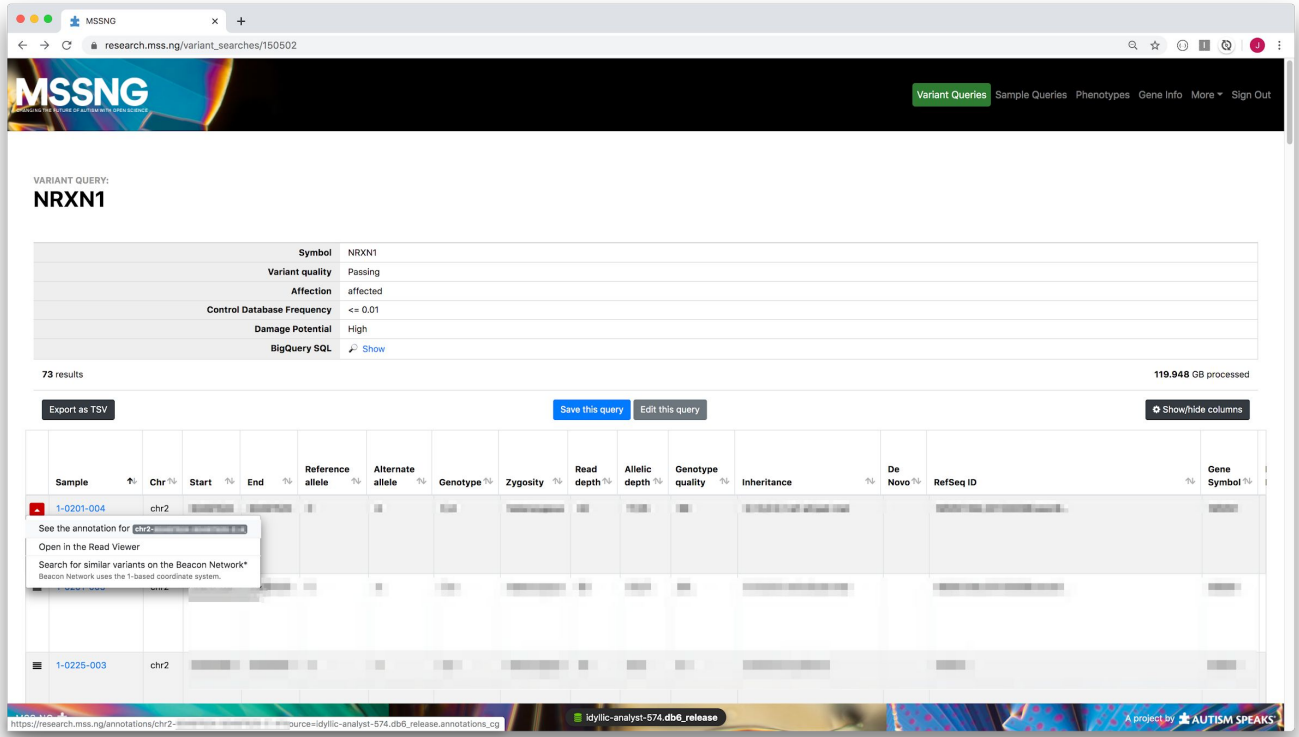
Choose Which Columns to Include in the Query



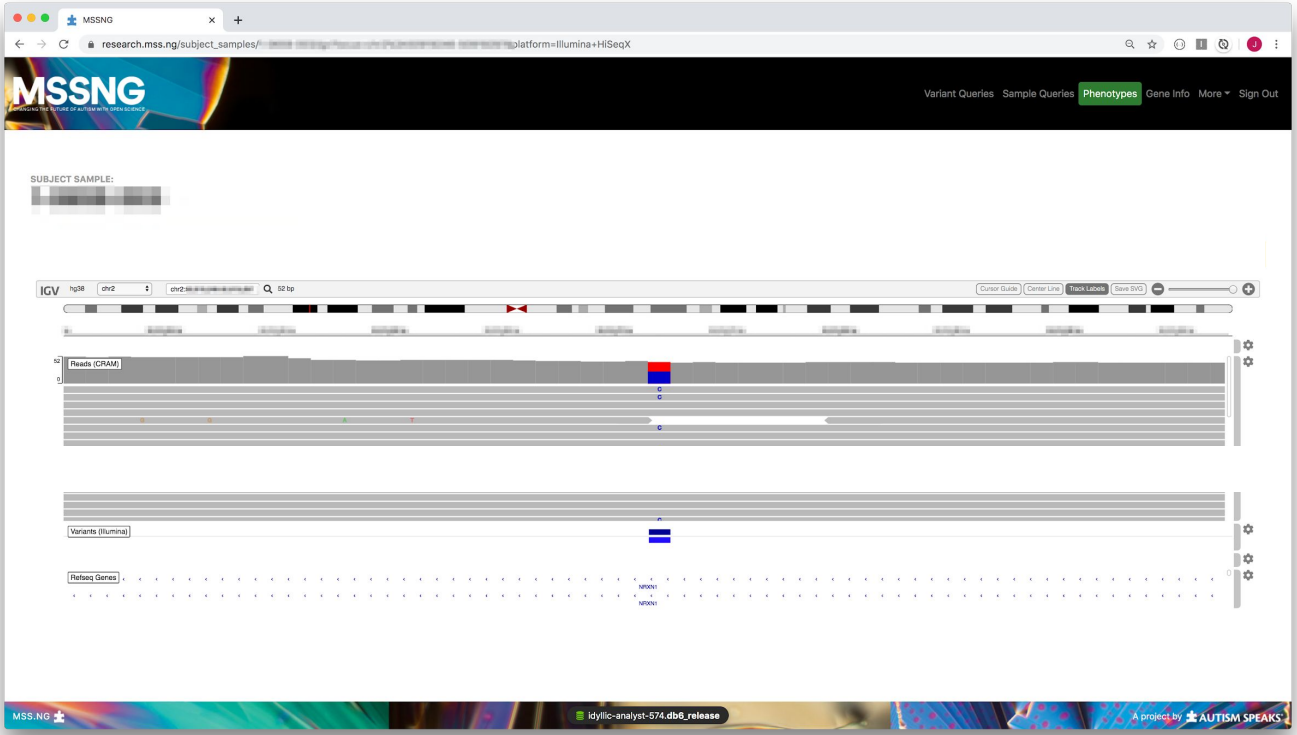
View/Copy Underlying BigQuery SQL Statement



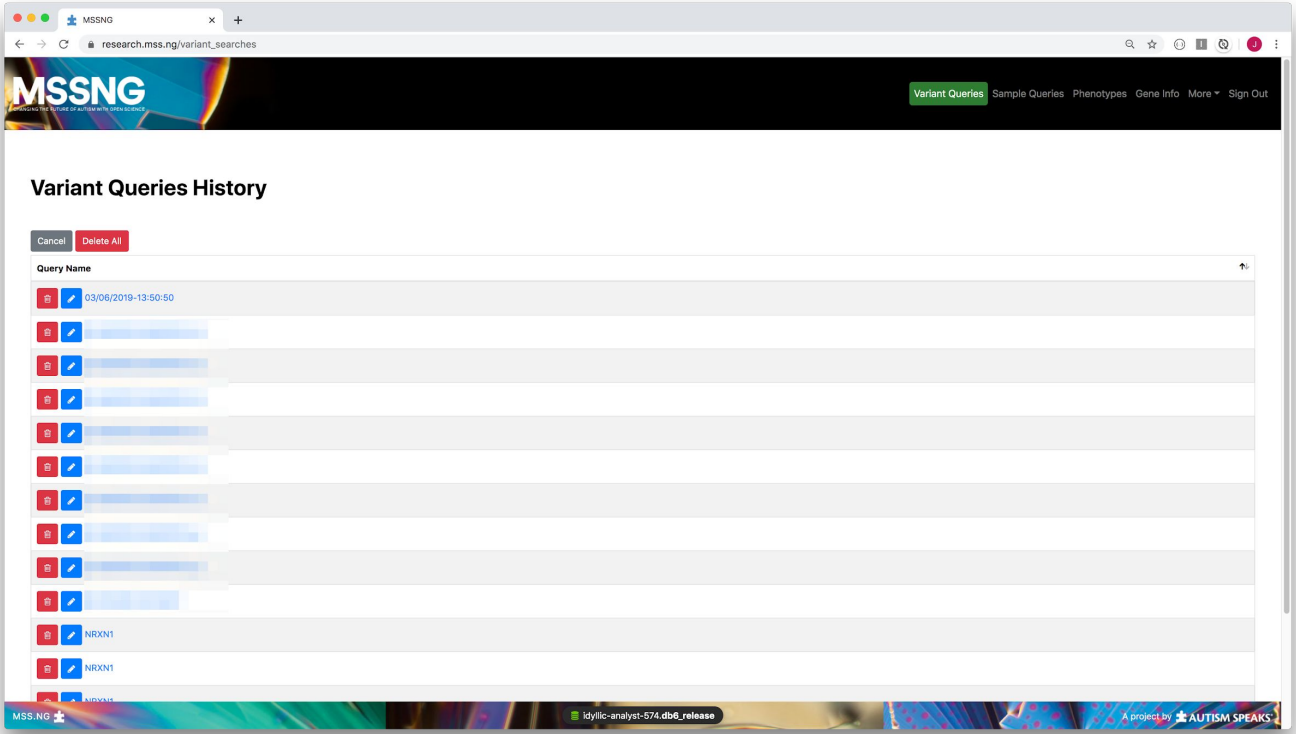
Jump to Related Resources for a Sample Variant



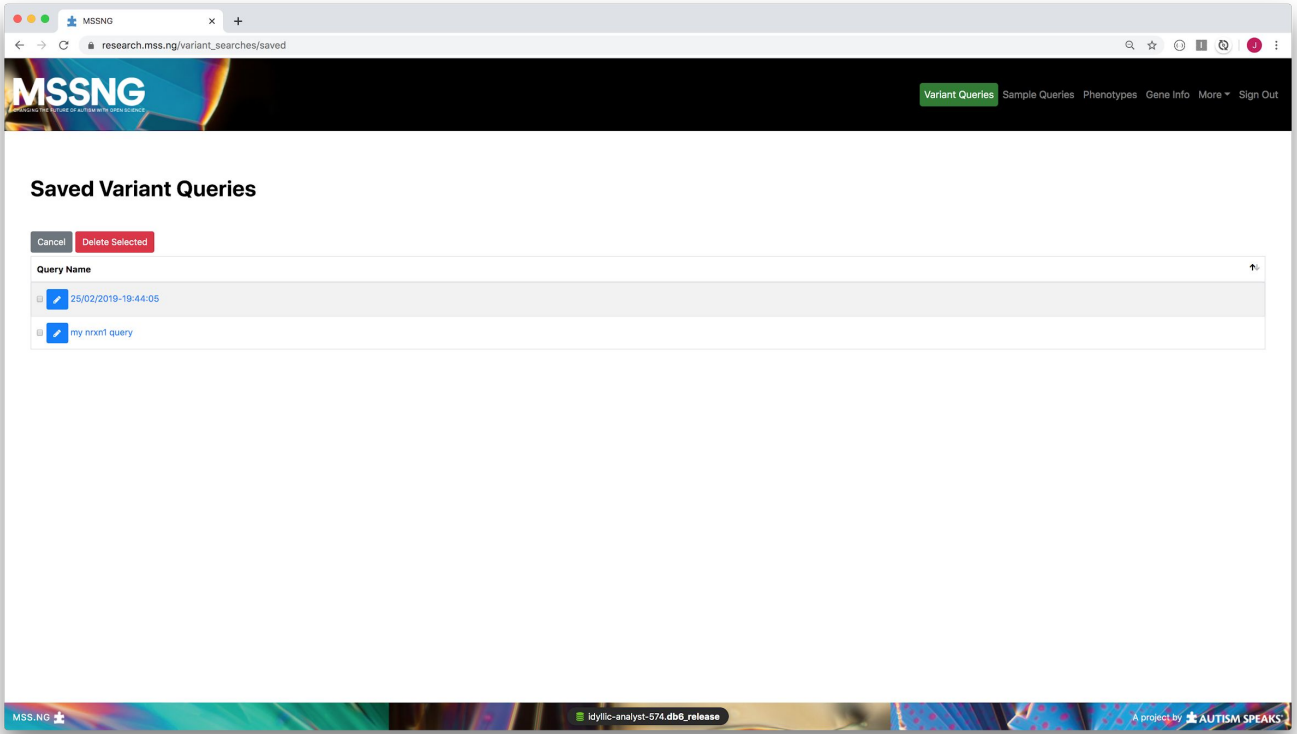
IGV Read Viewer



History of Past Queries

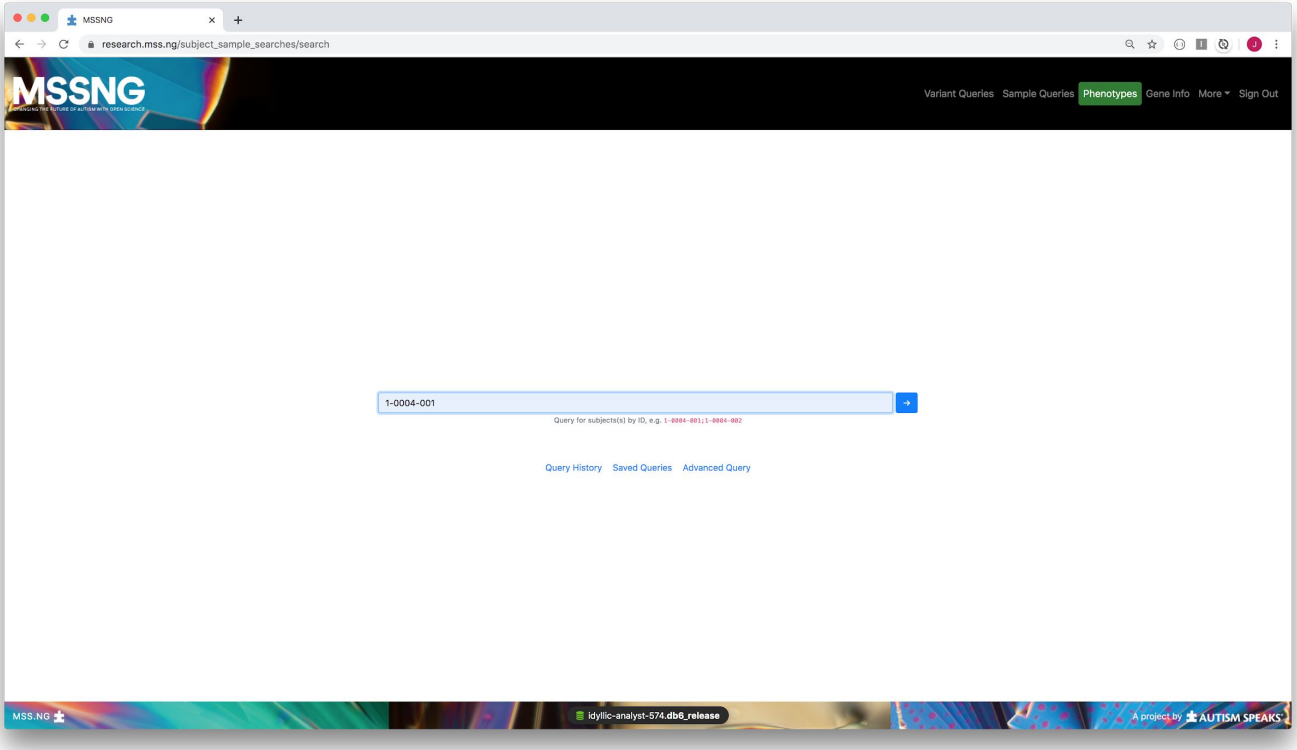


Saved Queries (separate from history)

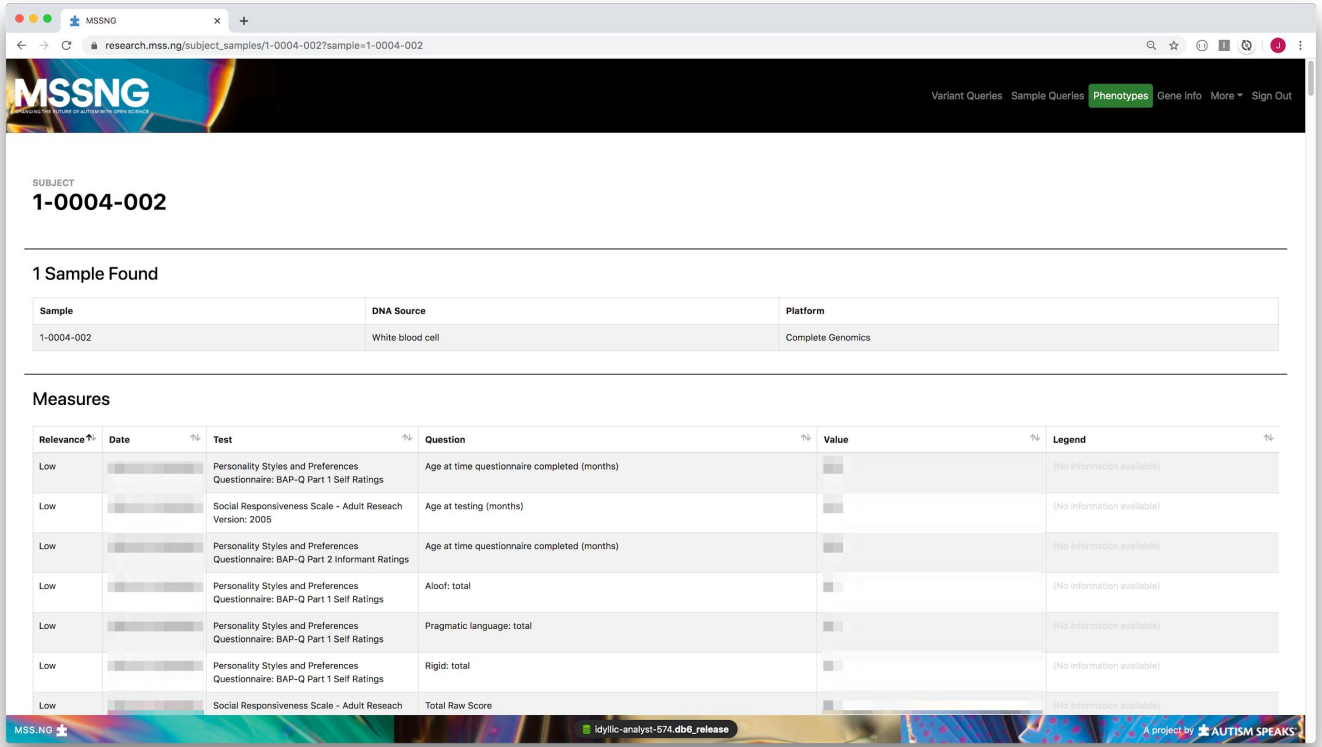


Subject Phenotype Query

“One Box” Subject Query

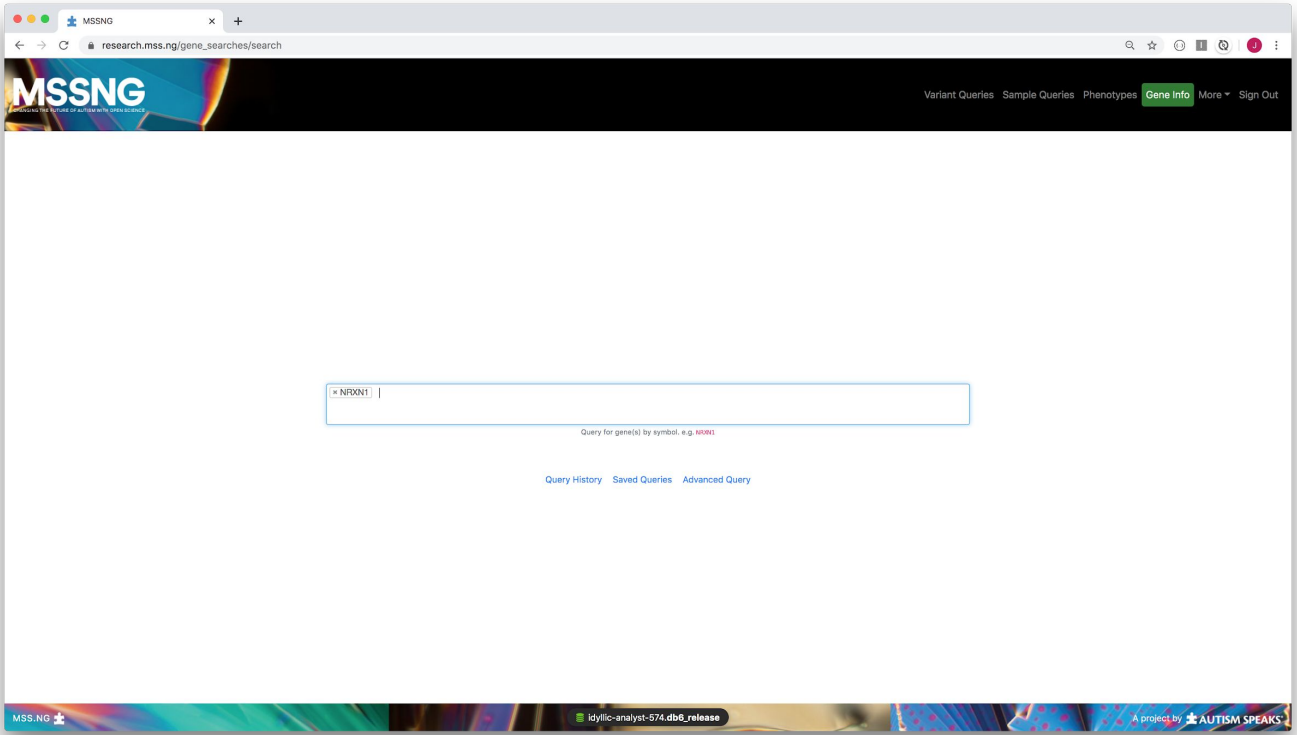


Sample Info and Subject Phenotypes

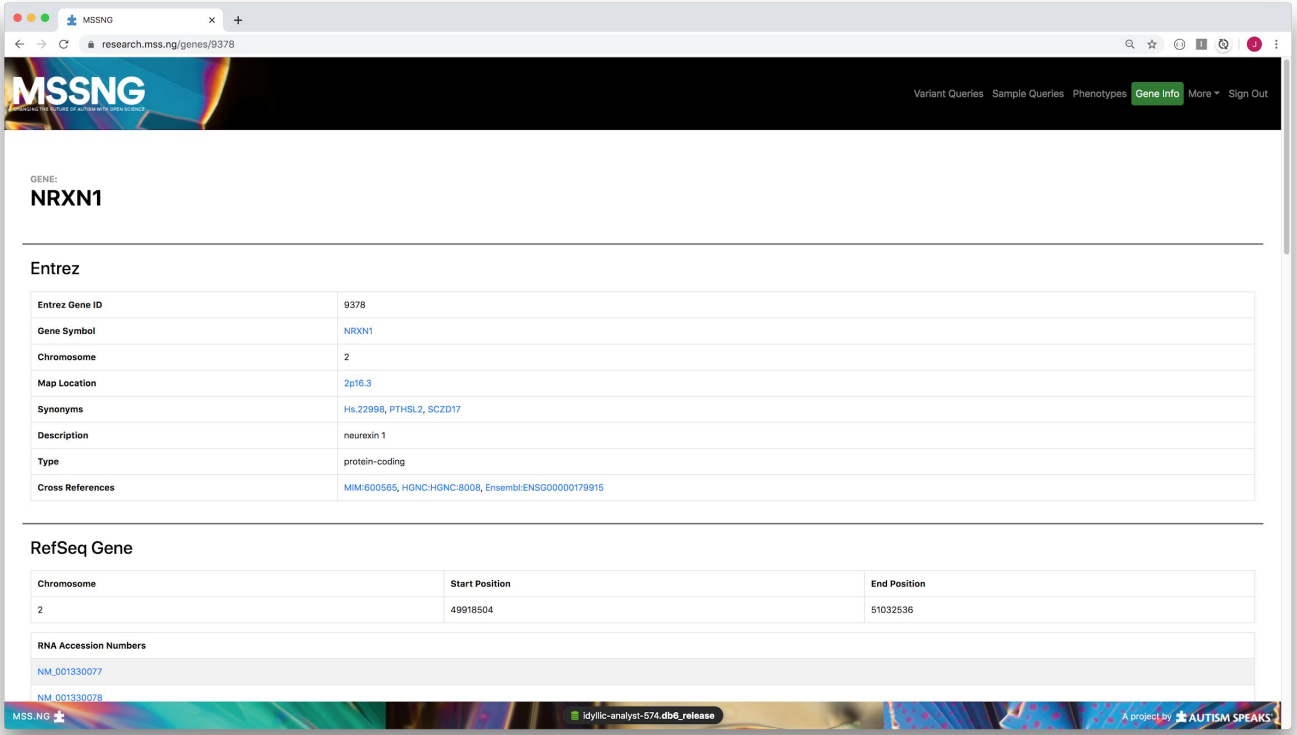


Gene Info

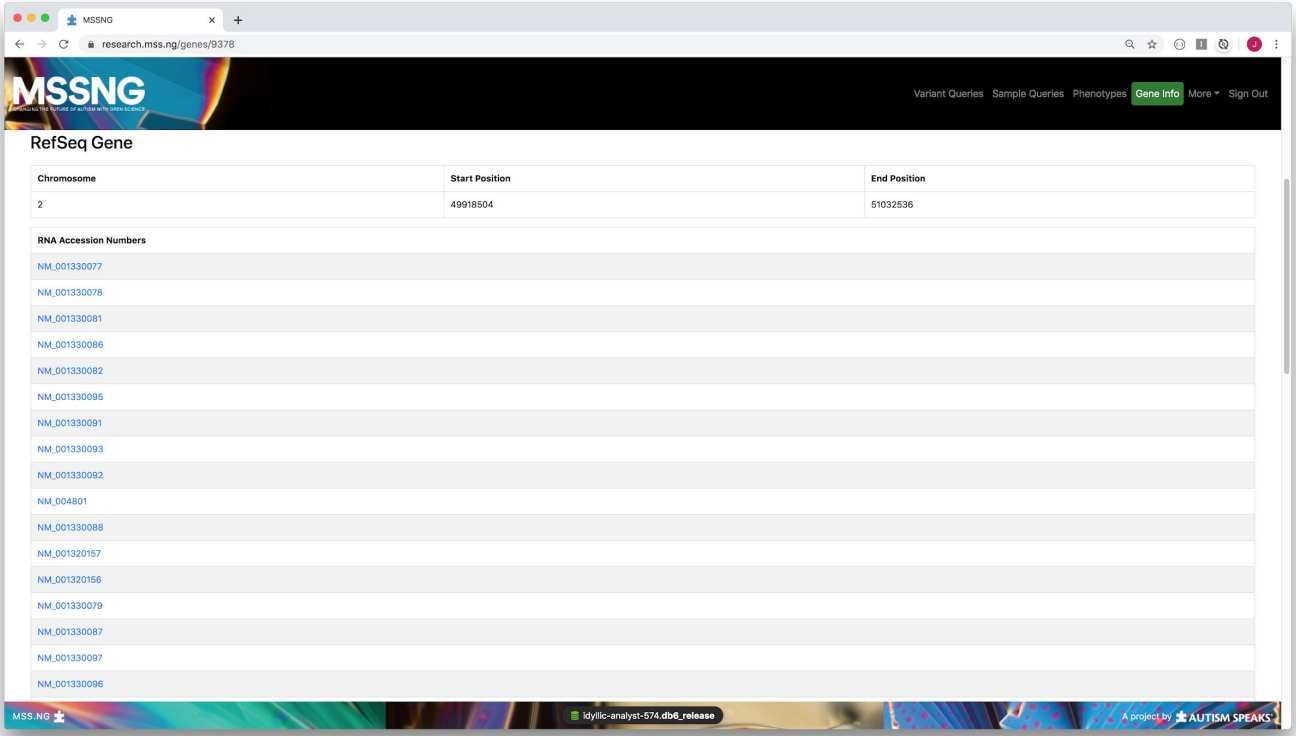
“One Box” Gene Info Query



Gene Info (1/4)


MSSNG
research.mss.ng/genes/9378
MSSNG
Variant Queries
Sample Queries
Phenotypes
Gene Info
More
Sign Out
GENE:
NRXN1
Entrez
Entrez Gene ID 9378
Gene Symbol NRXN1
Chromosome 2
Map Location 2p16.3
Synonyms Hs.22998, PTHSL2, SCZD17
Description neurexin 1
Type protein-coding
Cross References MIM:600565, HGNC:HGNC:8008, Ensembl:ENSG00000179915
RefSeq Gene
Chromosome
Start Position
End Position
2
49918504
51032536
RNA Accession Numbers
NM_001330077
NM_001330078
MSS.NG
idyllic-analyst-574.db6_release
A project by AUTISM SPEAKS

Gene Info (2/4)


MSSNG
research.mss.ng/genes/9378
MSSNG
Variant Queries
Sample Queries
Phenotypes
Gene Info
More
Sign Out
RefSeq Gene
Chromosome
Start Position
End Position
2
49918504
51032536
RNA Accession Numbers
NM_001330077
NM_001330078
NM_001330081
NM_001330086
NM_001330082
NM_001330095
NM_001330091
NM_001330093
NM_001330092
NM_004801
NM_001330088
NM_001320157
NM_001320156
NM_001330079
NM_001330087
NM_001330097
NM_001330096
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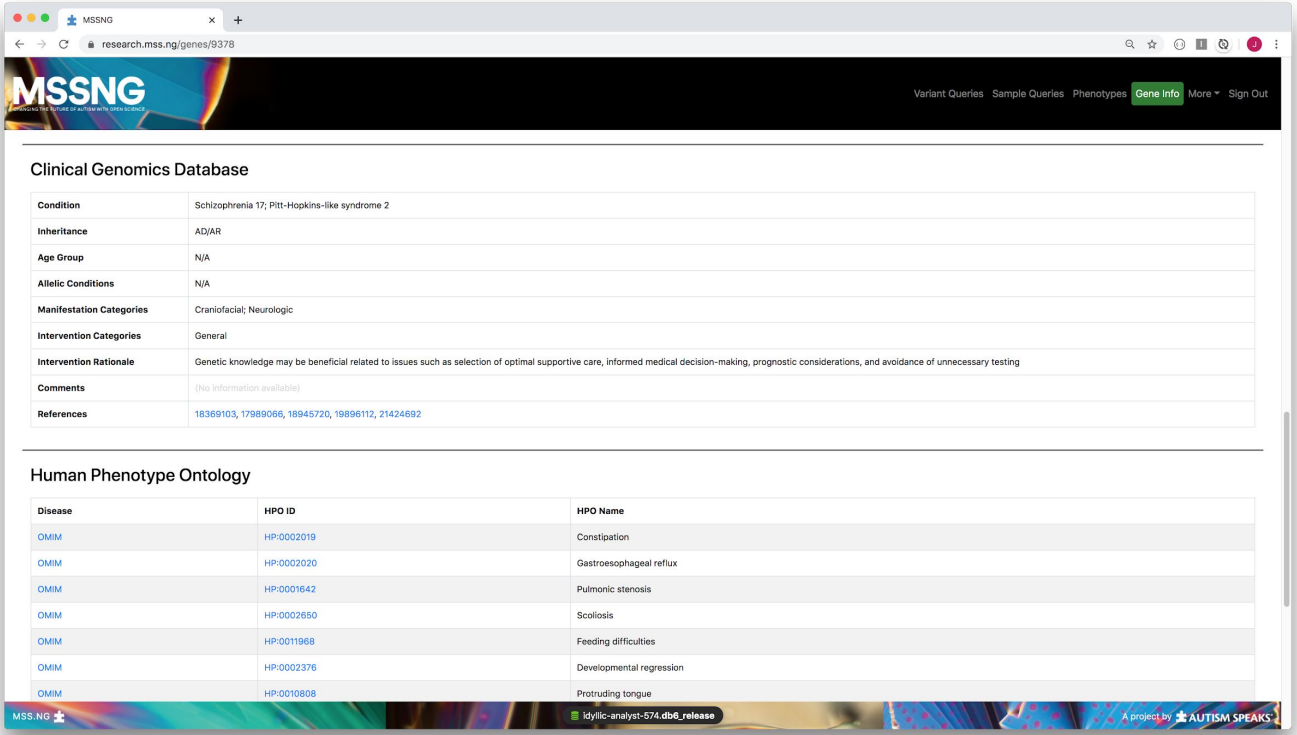
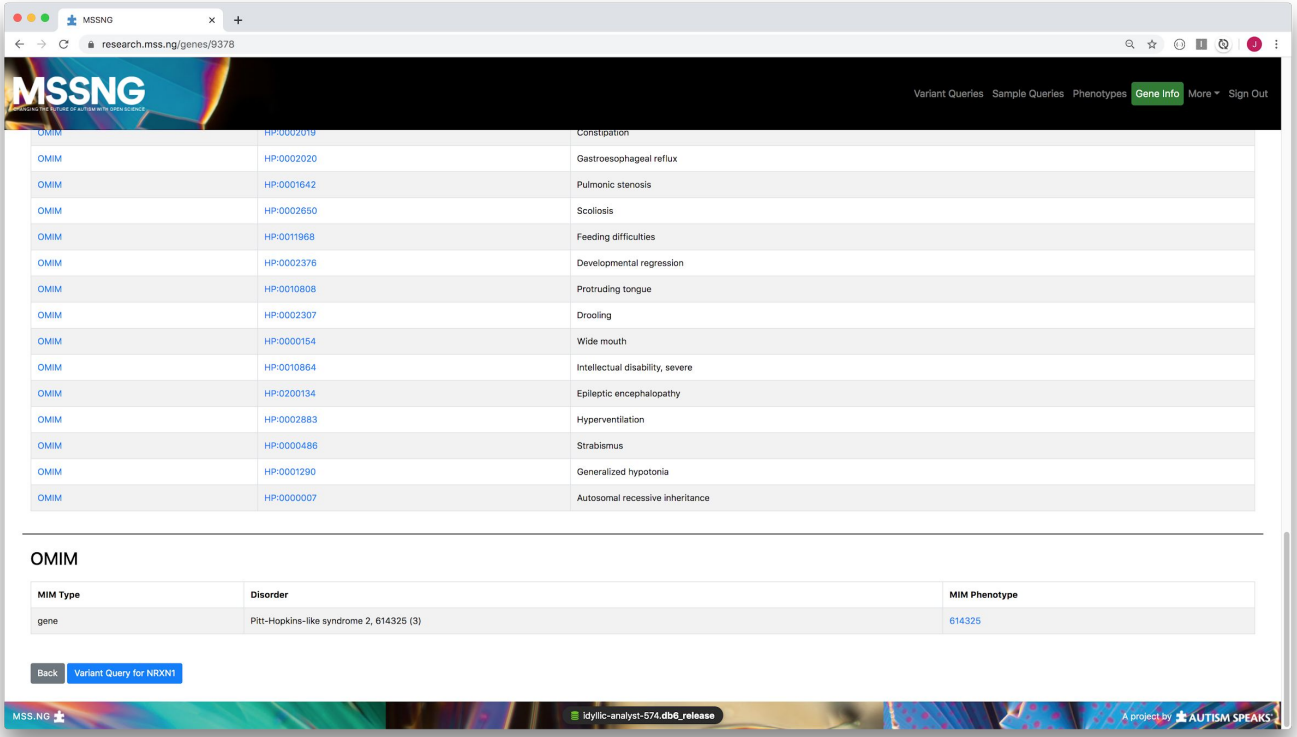
idyllic-analyst-574.db6_release
A project by AUTISM SPEAKS

Gene Info (3/4)


MSSNG
research.mss.ng/genes/9378
MSSNG
Variant Queries
Sample Queries
Phenotypes
Gene Info
More
Sign Out
Clinical Genomics Database
Condition
Schizophrenia 17; Pitt-Hopkins-like syndrome 2
Inheritance
AD/AR
Age Group
N/A
Allelic Conditions
N/A
Manifestation Categories
Craniofacial; Neurologic
Intervention Categories
General
Intervention Rationale
Genetic knowledge may be beneficial related to issues such as selection of optimal supportive care, informed medical decision-making, prognostic considerations, and avoidance of unnecessary testing
Comments
(No information available)
References
18369103, 17989066, 18945720, 19896112, 21424692
Human Phenotype Ontology
Disease
HPO ID
HPO Name
OMIM
HP:0002019
Constipation
OMIM
HP:0002020
Gastroesophageal reflux
OMIM
HP:0001642
Pulmonic stenosis
OMIM
HP:0002650
Scoliosis
OMIM
HP:0011968
Feeding difficulties
OMIM
HP:0002376
Developmental regression
OMIM
HP:0010808
Protruding tongue
MSS.NG
idyllic-analyst-574.db6_release
A project by AUTISM SPEAKS

Gene Info (4/4)


MSSNG
research.mss.ng/genes/9378
MSSNG
Variant Queries
Sample Queries
Phenotypes
Gene Info
More
Sign Out
OMIM HP:0002019 Constipation
OMIM HP:0002020 Gastroesophageal reflux
OMIM HP:0001642 Pulmonic stenosis
OMIM HP:0002650 Scoliosis
OMIM HP:0011968 Feeding difficulties
OMIM HP:0002376 Developmental regression
OMIM HP:0010808 Protruding tongue
OMIM HP:0002307 Drooling
OMIM HP:0000154 Wide mouth
OMIM HP:0010864 Intellectual disability, severe
OMIM HP:0200134 Epileptic encephalopathy
OMIM HP:0002883 Hyperventilation
OMIM HP:0000486 Strabismus
OMIM HP:0001290 Generalized hypotonia
OMIM HP:0000007 Autosomal recessive inheritance
OMIM
MIM Type
Disorder
MIM Phenotype
gene
Pitt-Hopkins-like syndrome 2, 614325 (3)
614325
Back
Variant Query for NRXN1
MSS.NG
idyllic-analyst-574.db6_release
A project by AUTISM SPEAKS