

# step 1

select data set in Hub.iobio

The screenshot shows the Hub.iobio web interface. The browser tabs are 'Hub.iobio' and 'Cohort-Gene.iobio'. The address bar shows 'https://staging.frameshift.io/#/projects'. The page title is 'hub.iobio' and the user is logged in as 'John Doe'. The main heading is 'Projects' with a 'Create New Project' link. Below the heading is a 'Filter Results' section with a search query input. A table lists 8 records of projects. The 'SSC' project is highlighted with an orange box and an arrow. The table columns are: Name, Description, Sample count, Created On, and Last Updated.

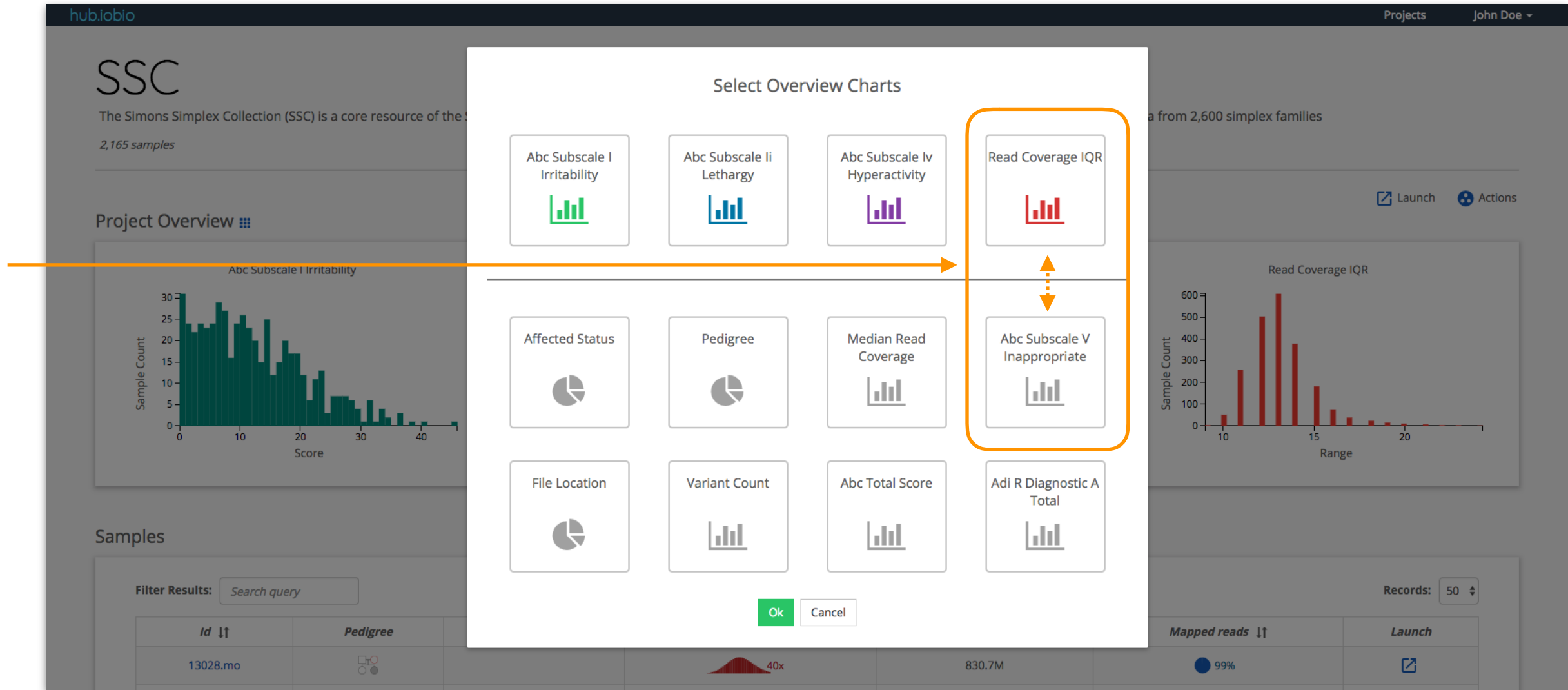
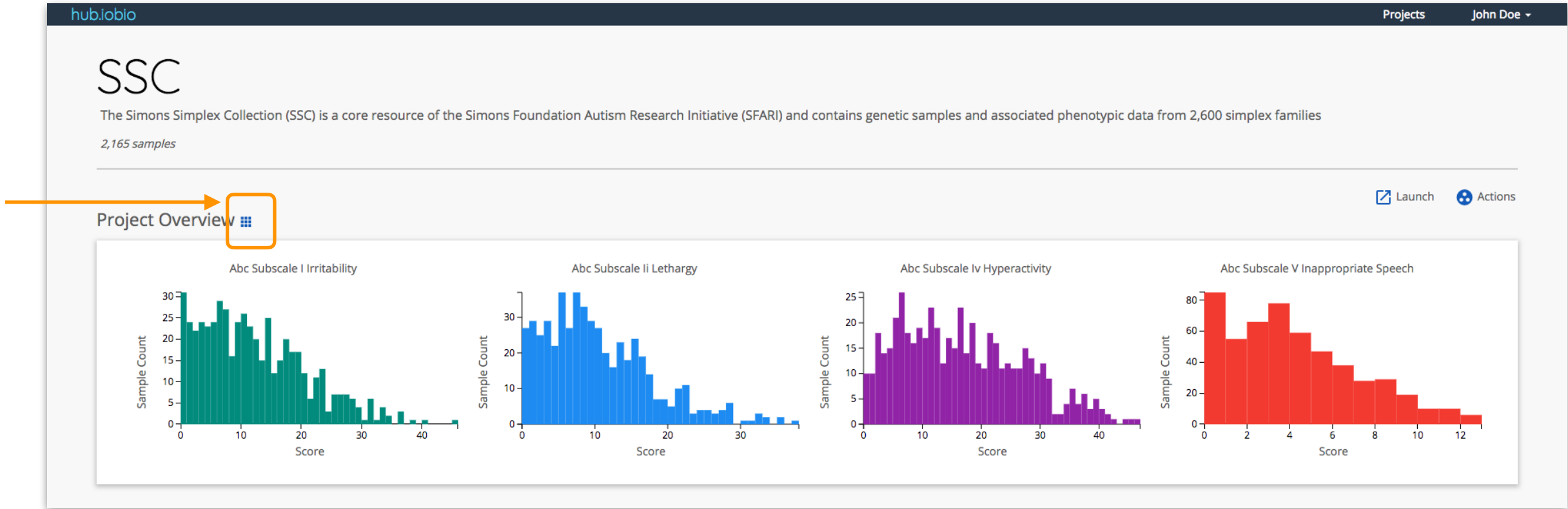
Name ↓↑	Description ↓↑	Sample count ↓↑	Created On ↓↑	Last Updated ▲
H1K	investigating 1k priority patients	753	25 June 2018	25 June 2018
Ewing's Sarcoma	A cancer that most often occurs in and around the bones	1,049	25 June 2018	25 June 2018
1000G WGS	1000+ samples from different ethnic groups around the world	851	25 June 2018	25 June 2018
SSC	The Simons Simplex Collection (SSC) is a core resource of the Simons Foundation Autism Research Initiative (SFARI) and contains genetic samples and associated phenotypic data from 2,600 simplex families	2,165	25 June 2018	25 June 2018
Platinum Data Set		4	25 June 2018	25 June 2018
Distributed Data	Data on S3 cloud and Simons hardware	4	25 June 2018	25 June 2018
All SFARI DATA	Big dataset	8,278	25 June 2018	25 June 2018
SPARK_Pilot	pilot samples	1,406	25 June 2018	25 June 2018

8 records

© 2018 Frameshift Labs, Inc. [Terms](#) [Privacy](#) [Security](#) [About](#) [Blog](#) [Contact](#) [Help](#)

# step 2

choose data filters



step 3

filter data

hub.iobio

ProjectsJohn Doe

SSC

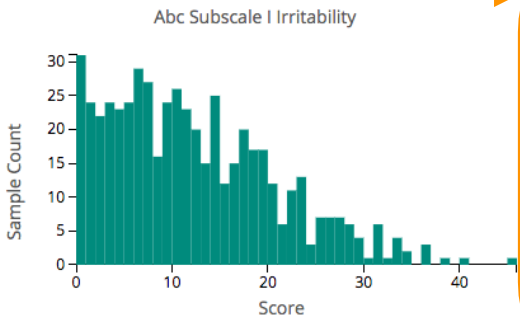
The Simons Simplex Collection (SSC) is a core resource of the Simons Foundation Autism Research Initiative (SFARI) and contains genetic samples and associated phenotypic data from 2,600 simplex families

2,165 samples

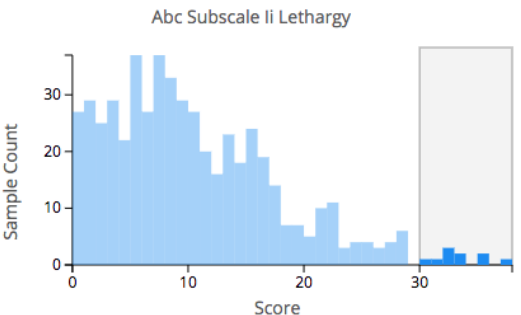
LaunchActions

Project Overview

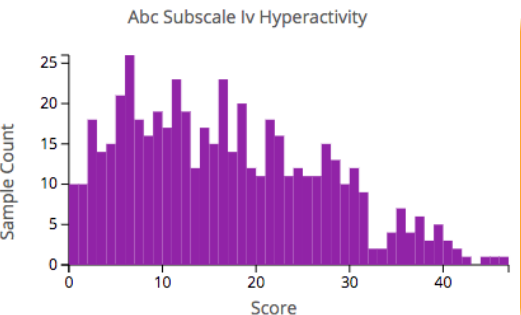
Abc Subscale I Irritability



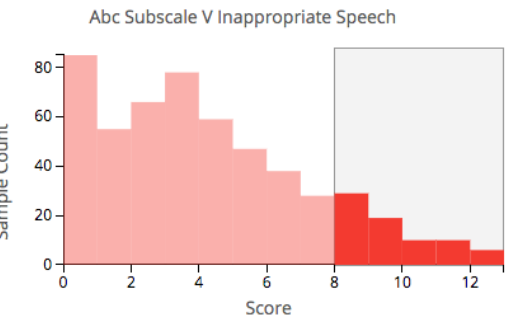
Abc Subscale II Lethargy



Abc Subscale IV Hyperactivity



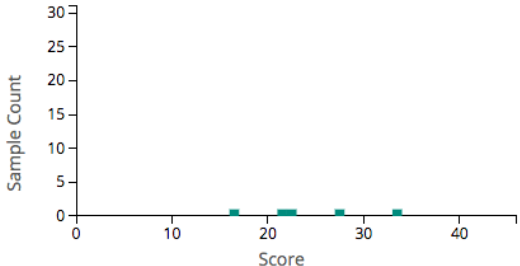
Abc Subscale V Inappropriate Speech



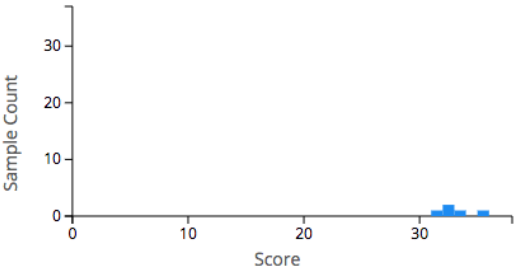
Filtered ChartsPedigree Charts

Clear FiltersUse same x-scaleUse same y-scale

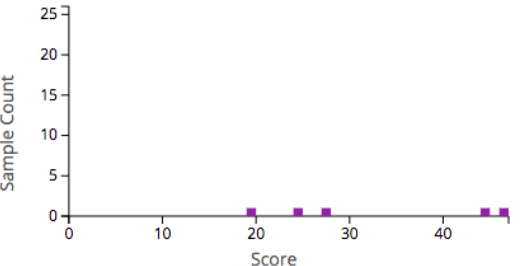
Abc Subscale I Irritability



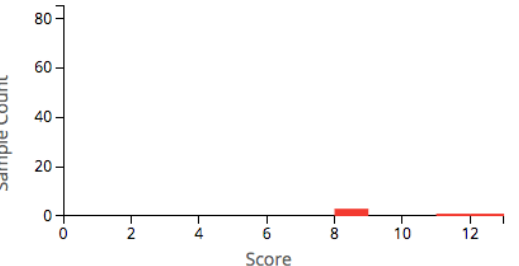
Abc Subscale II Lethargy



Abc Subscale IV Hyperactivity



Abc Subscale V Inappropriate Speech



Samples

Filter Results:

Search query

Id	Pedigree	Description	Read coverage	Total reads	Mapped reads	Launch
11948.p1			48x	999.6M	99%	
12891.p1			35x	740.3M	99%	
13539.p1			37x	789M	98%	
13835.p1			47x	973.7M	99%	
14314.p1			40x	839.4M	99%	

5 records

© 2018 Frameshift Labs, Inc.

TermsPrivacySecurity

AboutBlogContactHelp

# step 4

## launch cohort

hub.iobio

ProjectsJohn Doe

# SSC

The Simons Simplex Collection (SSC) is a core resource of the Simons Foundation Autism Research Initiative (SFARI) and contains genetic samples and associated phenotypic data from 2,600 simplex families

2,165 samples

Project Overview

Launch

Actions

Multibam.iobio

Cohort-Gene.iobio

Polygenic Risk Score

Abc Subscale I Irritability

Abc Subscale II Lethargy

Abc Subscale IV Hyperactivity

Abc Subscale V Inappropriate Speech

Filtered Charts

Pedigree Charts

Clear Filters

Use same x-scale

Use same y-scale

Abc Subscale I Irritability

Abc Subscale II Lethargy

Abc Subscale IV Hyperactivity

Abc Subscale V Inappropriate Speech

Samples

Filter Results:

Search query

Id	Pedigree	Description	Read coverage	Total reads	Mapped reads	Launch
11948.p1			48x	999.6M	99%	
12891.p1			35x	740.3M	99%	
13539.p1			37x	789M	98%	
13835.p1			47x	973.7M	99%	
14314.p1			40x	839.4M	99%	

5 records

© 2018 Frameshift Labs, Inc.

[Terms](#)[Privacy](#)[Security](#)

[About](#)[Blog](#)[Contact](#)[Help](#)

# step 5

select gene

hub.io

ProjectsJohn Doe

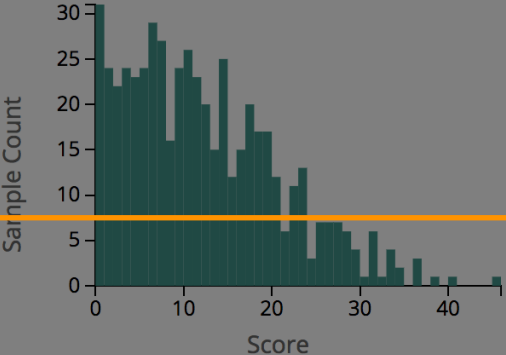
## SSC

The Simons Simplex Collection (SSC) is a core resource from 2,600 simplex families

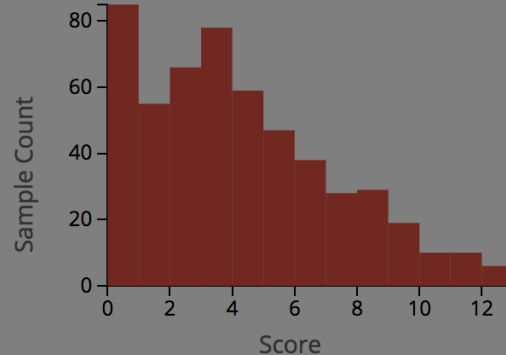
2,165 samples

### Project Overview

Abc Subscale I Irritability



Abc Subscale V Inappropriate Speech



Filtered Charts | Pedigree Charts

Clear FiltersUse same x-scaleUse same y-scale

Abc Subscale I Irritability

Abc Subscale II Lethargy

Abc Subscale IV Hyperactivity

Abc Subscale V Inappropriate Speech

cohort-gene.io

An application that identifies and visualizes variants that are enriched in a selected subset relative to all probands

Steps

1. Select a subset from the project overview charts

abc subscale ii lethargy

2. Number of samples selected: 74

3. Enter gene of interest

Please type in the exact name of the gene.

fam8a1

Launch cohort-gene.io

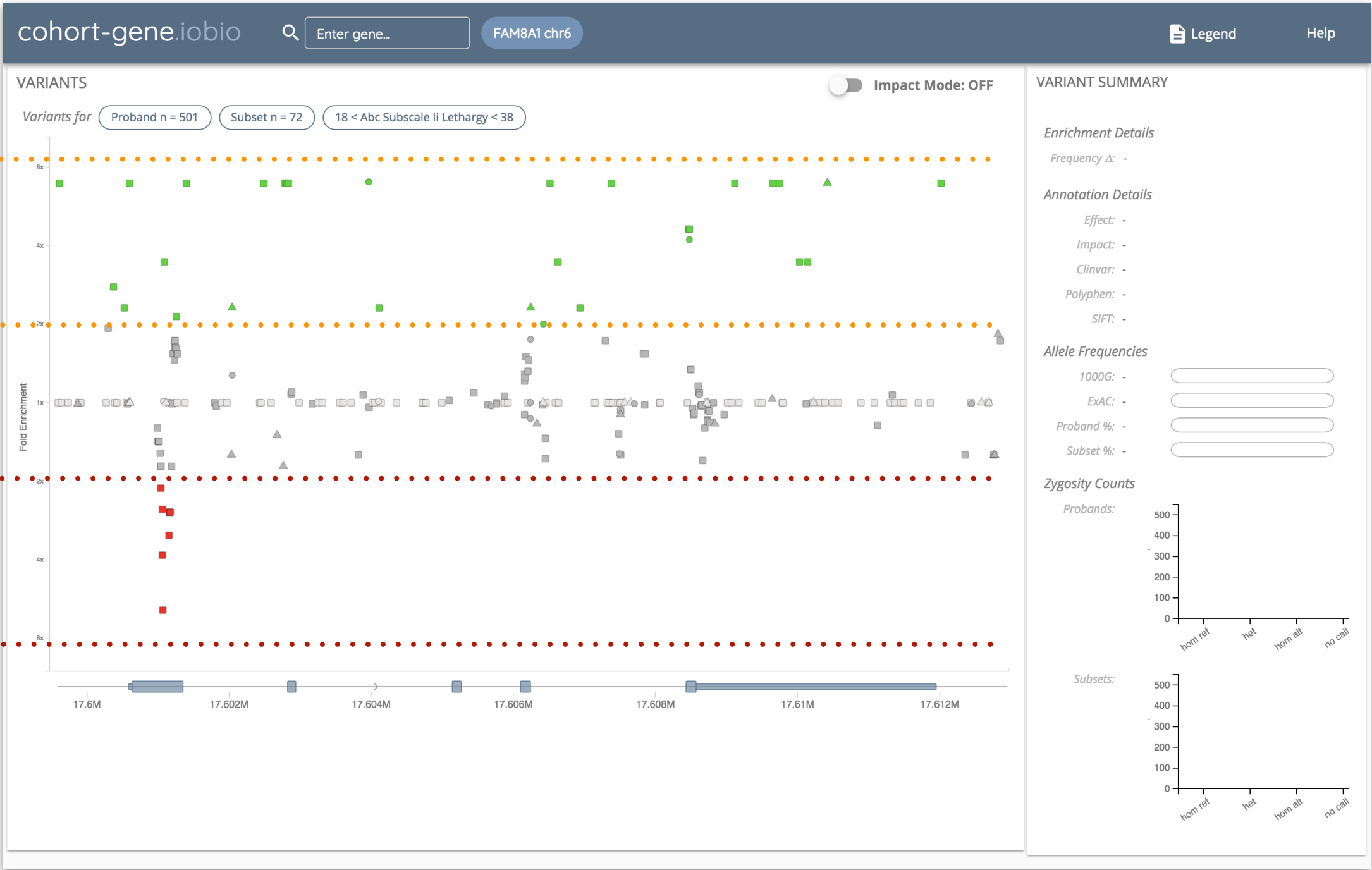
Cancel

# step 6

review enriched variants

green variants  
represent  
those enriched in  
the subset

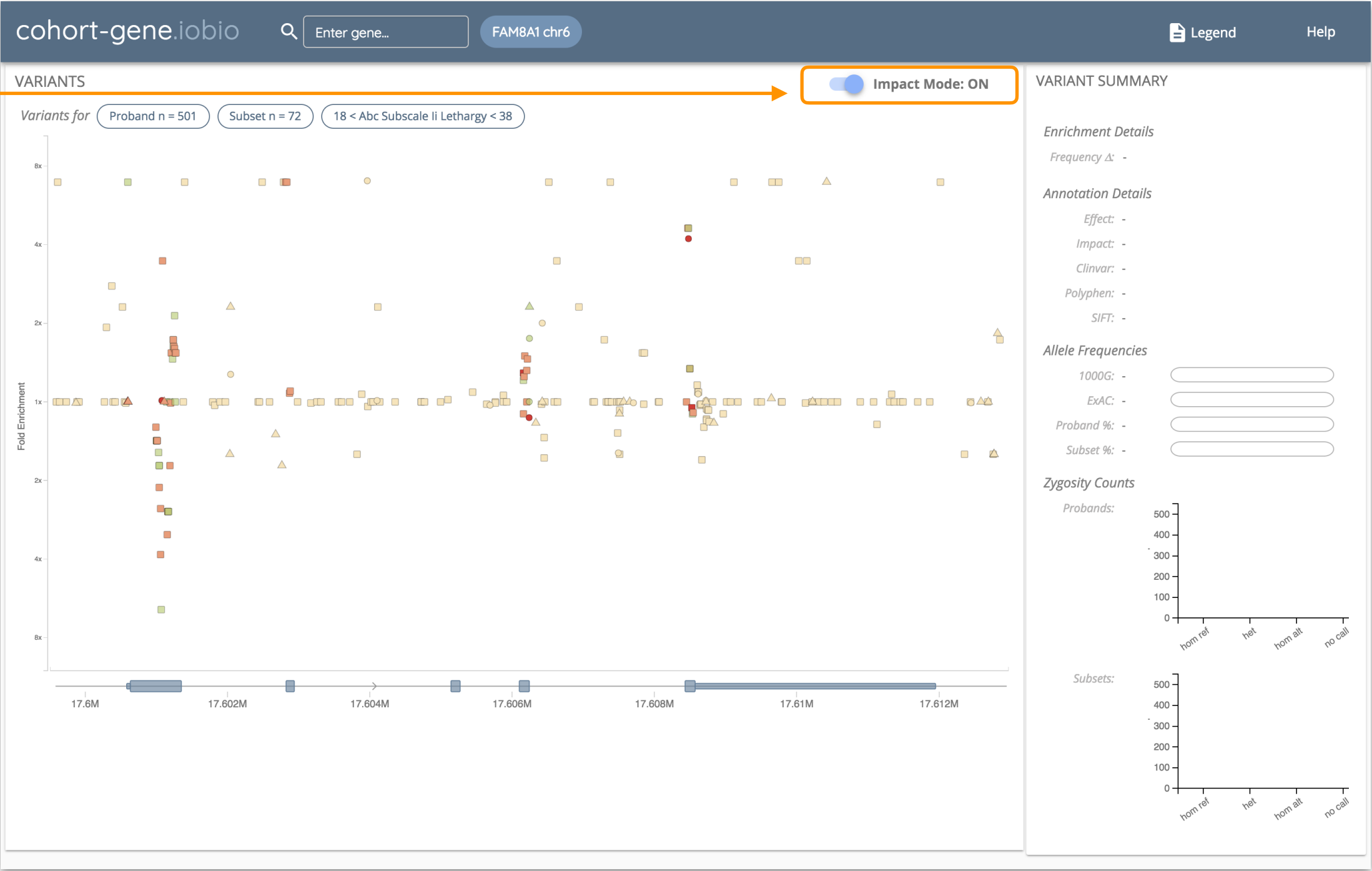
red variants  
represent  
those reduced in  
the subset



# step 7

switch to impact mode

variants colored  
by predicted  
functional impact



step 8

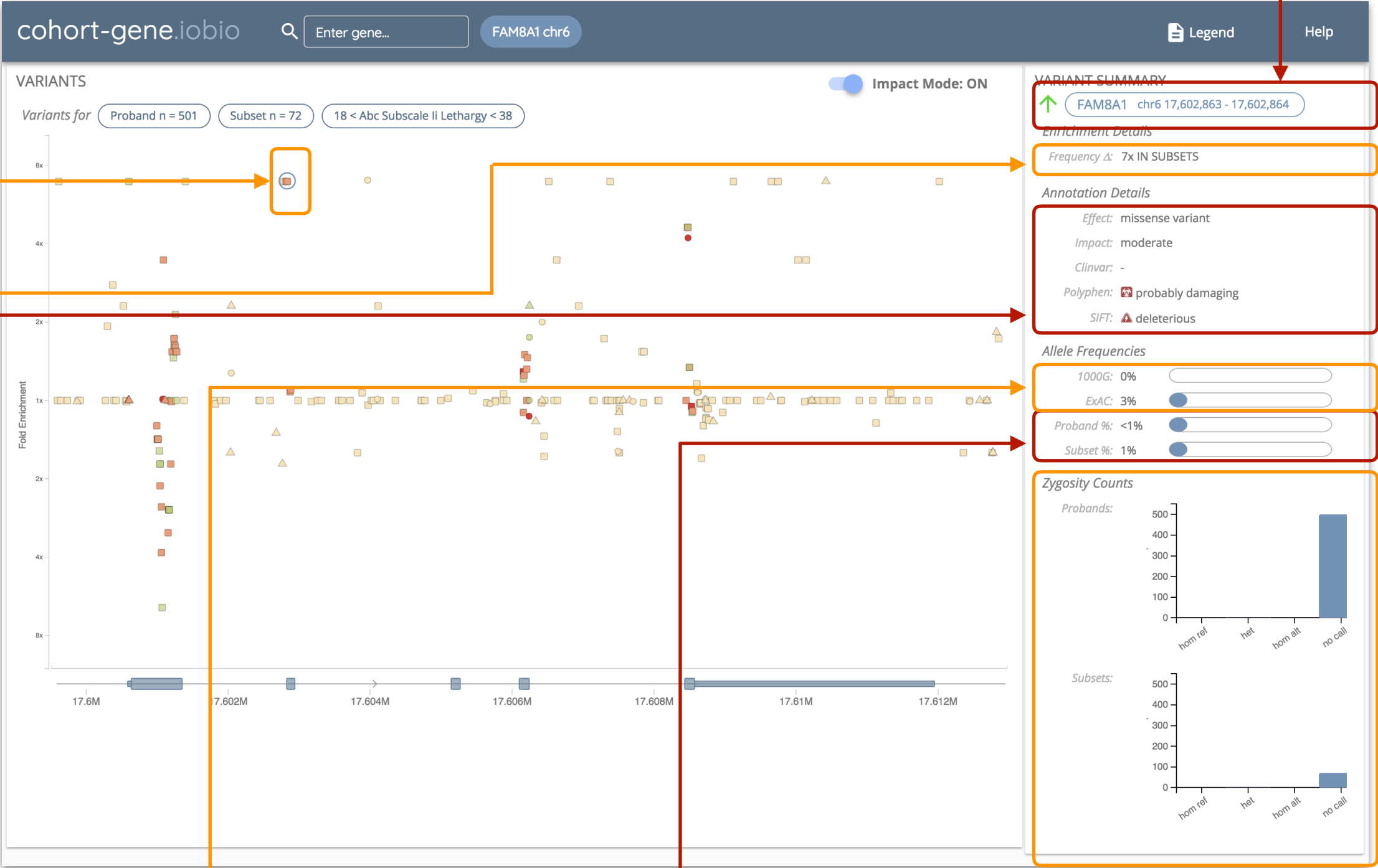
review individual variant details

genomic coordinates

select variant

fold enrichment

functional annotations



population frequencies

cohort frequencies

tallies for homozygous & heterozygous instances of the variant



step 9

continue searching loci

