

Target Gene Notebook

User Guide
1.2

Target Gene Notebook allows the curation of persistent, editable, and distributable databases of genomic, biological, chemical, and other data that pertain to the local neighborhood around a gene of interest.

The current version draws heavily on the many harmonized data sources available through Ensembl¹.

1. Daniel R. Zerbino, Premanand Achuthan, Wasiu Akanni, M. Ridwan Amode, Daniel Barrell, Jyothish Bhai, Konstantinos Billis, Carla Cummins, Astrid Gall, Carlos García Giro'n, Laurent Gil, Leo Gordon, Leanne Haggerty, Erin Haskell, Thibaut Hourlier, Osagie G. Izuogu, Sophie H. Janacek, Thomas Juettemann, Jimmy Kiang To, Matthew R. Laird, Ilias Lavidas, Zhicheng Liu, Jane E. Loveland, Thomas Maurel, William McLaren, Benjamin Moore, Jonathan Mudge, Daniel N. Murphy, Victoria Newman, Michael Nuhn, Denye Ogeh, Chuang Kee Ong, Anne Parker, Mateus Patricio, Harpreet Singh Riat, Helen Schuilenburg, Dan Sheppard, Helen Sparrow, Kieron Taylor, Anja Thormann, Alessandro Vullo, Brandon Walts, Amonida Zadissa, Adam Frankish, Sarah E. Hunt, Myrto Kostadima, Nicholas Langridge, Fergal J. Martin, Matthieu Muffato, Emily Perry, Magali Ruffier, Dan M. Staines, Stephen J. Trevanion, Bronwen L. Aken, Fiona Cunningham, Andrew Yates, Paul Flicek
Ensembl 2018.
PubMed PMID: 29155950.

Target Gene Notebook was originally supported and created at Eisai Inc.'s Andover Innovative Medicine Institute in Andover, MA.

We encourage further development of this resource.

Target Gene Notebook team

Eisai (design, implementation and testing):

Andrew Kirby

Janna Hutz

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Massachusetts General Hospital (design input):

Mark Daly

Hailiang Huang

Requirements

Server:
Java >= 1.7

Client:
Google Chrome
JavaScript enabled

Target Gene Notebook uses these frameworks/libraries:

Bootstrap 3.3.7 (MIT license)
FileSaver.js 1.3.2 (MIT license)
Huebee 2.0.0 (MIT license)
jQuery 2.2.4 (MIT license)
jQuery UI 1.12.1 (MIT license)
Lightbox2 2.9.0 (MIT license)
Select2 4.0.3 (MIT license)
tablesorter 2.28.14 (MIT license)
Spark micro framework (sparkjava.com, Apache 2 license)

Dependencies:
commons-io 2.4
commons-lang3 3.4
gson 2.7
jsoup 1.8.1
slf4j-simple 1.7.21
sqlite-jdbc 3.7.2

GUI Overview

Target Gene Notebook 1.1:1.1 User Guide About

View Mode ⓘ Credible Sets ⓘ LD Summary (0.6/2 threshold, 1KGp3.EUR)

Summary (IFIH1.TSS Ensembl 92;chr2:162,318,700) ⓘ Hide non-coding genes

Association Results

Association Details

- pQTL Results
- pQTL Details
- eQTL Results
- Variants of Interest

Marker	α	r ²	α	Coding
rs14572054	0.922			Y
rs10551540	0.734			
rs13023380	0.674			
rs7587426	0.616			

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Functional Variants Details

Expression Data Details

Protein and Structure Chemistry Details

Clinical Results

Clinical Data Details

References (18 out of 23 not yet reviewed)

Reviewed	1st Author	Year	Journal/Site/File	Title/Description	Curator Comment	Show/Add Details	X
<input checked="" type="checkbox"/>	Li Q.	2019	https://clue.io/repurposing-app	Curated information about drugs and drug targets from Broad, Genome-wide association study of paliperidone efficacy.	Enter comment	Show/Add Details	X
<input checked="" type="checkbox"/>		2017	Pharmacogenet. Genomics. http://www.ncbi.nlm.nih.gov/pmc/articles/PMC5501136/	UK Biobank Rapid GWAS	Enter comment	Show/Add Details	X
<input checked="" type="checkbox"/>	Petterson M.	2017	Am. J. Med. Genet. A	Further evidence for specific IFIH1 mutation as a cause of Singleton-Merten syndrome with phenotypic heterogeneity.	Enter comment	Show/Add Details	X
<input checked="" type="checkbox"/>	Gorman JA	2017	Nat. Immunol.	The A94G variant of the RNA sensor FIFH1 mediates an interferon program that limits viral infection but increases the risk for autoimmunity.	Enter comment	Show/Add Details (1)	X
<input checked="" type="checkbox"/>	Bronson PG	2016	Nat. Genet.	Common variants at PVT1, ATG13, AMBRA1, AHI1 and CLEC16A are associated with selective IgA deficiency.	Enter comment	Show/Add Details	
<input checked="" type="checkbox"/>	Morris DL	2016	Nat. Genet.	Genome-wide association meta-analysis in Chinese and European individuals identifies ten new loci associated with systemic lupus erythematosus.	Enter comment	Show/Add Details	
<input checked="" type="checkbox"/>	Jin Y.	2016	Nat. Genet.	Genome-wide association meta-analysis of autoimmune diseases identifies 23 new risk loci and highlights key pathways and regulatory variants.	Enter comment	Show/Add Details	
<input checked="" type="checkbox"/>	Bentham J.	2015	Nat. Genet.	Genetic association analyses implicate aberrant regulation of innate and adaptive immunity genes in the pathogenesis of systemic lupus erythematosus.	Enter comment	Show/Add Details	
<input checked="" type="checkbox"/>	Yin X.	2015	Nat. Commun.	Genome-wide meta-analysis identifies multiple novel associations and ethnic heterogeneity of psoriasis susceptibility.	Enter comment	Show/Add Details	

Page: 1 ⏪ ⏩ 1 to 10 of 23 rows ⏪ ⏩ 10 ⏪ Add Pubmed Reference Add File Reference Add Web Reference Add bioRxiv Reference

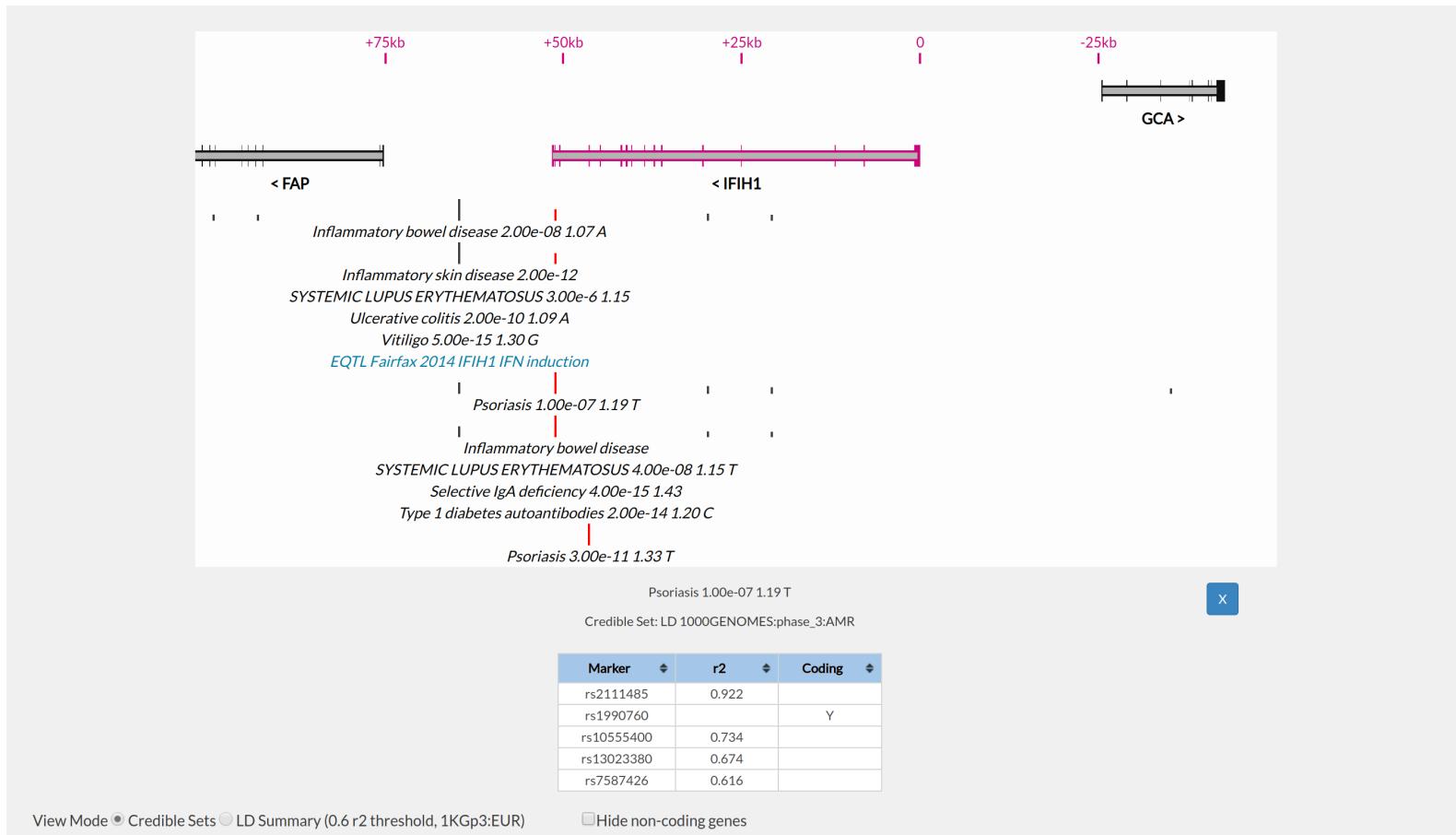
Interactive graphical view of curated data within a genomic context.

Text-based/tabular view of curated data.

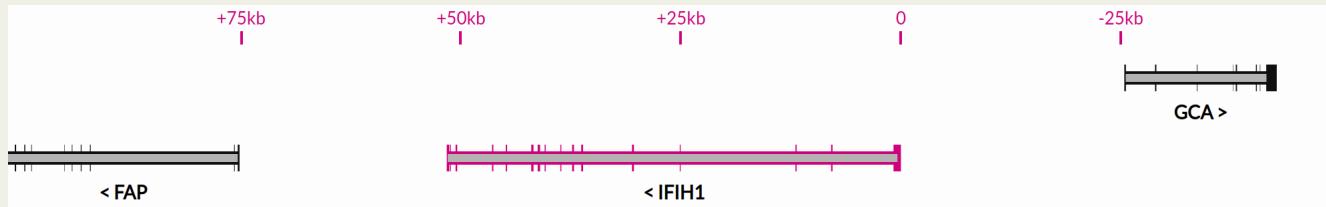
Interactive graphical view of curated data within a genomic context...

Interactive
graphical view of
curated data within
a genomic context.

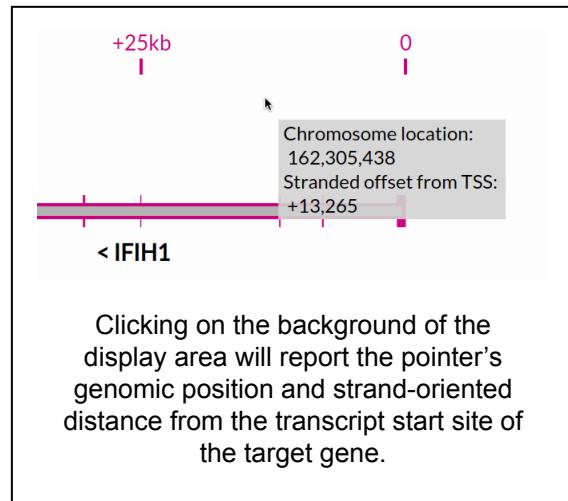
Overview



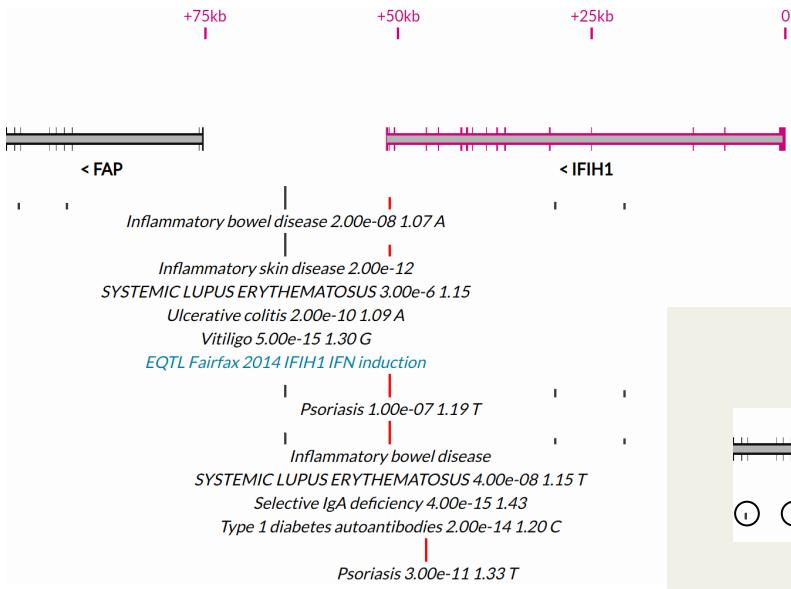
Interactive
graphical view of
curated data within
a genomic context.



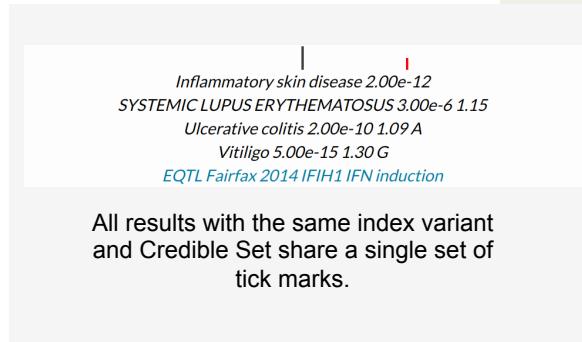
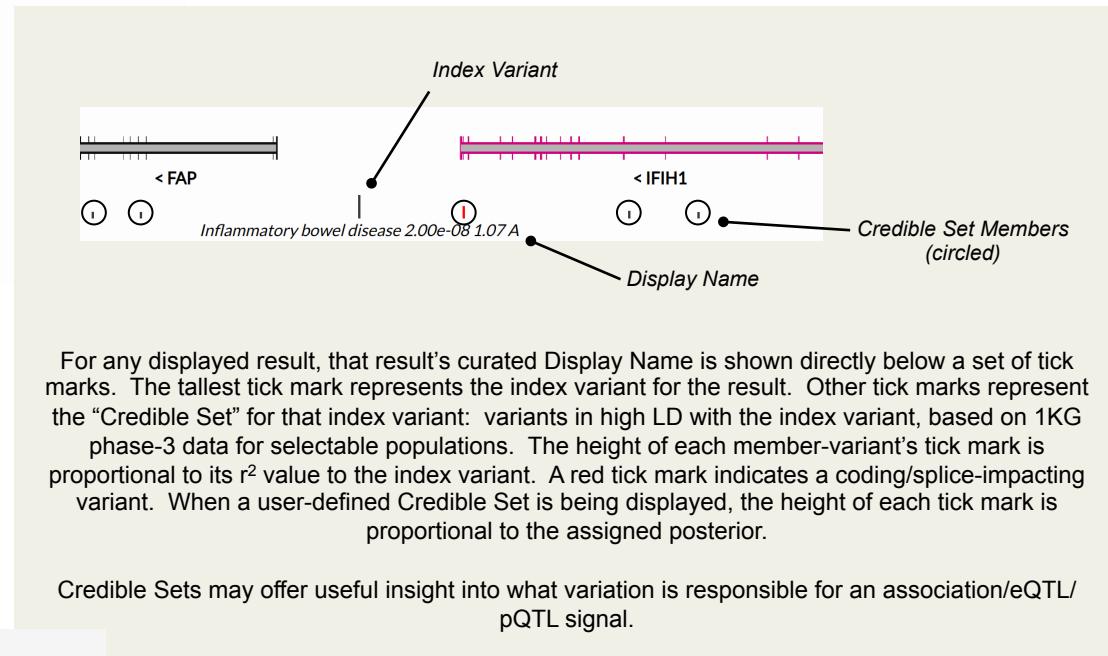
The genomic ruler is oriented relative to the start of the target-gene transcript.



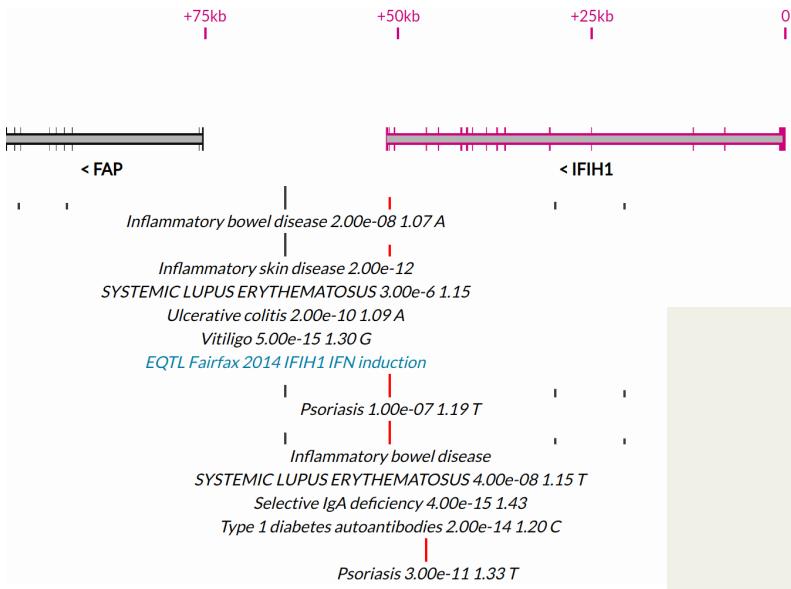
Interactive graphical view of curated data within a genomic context.



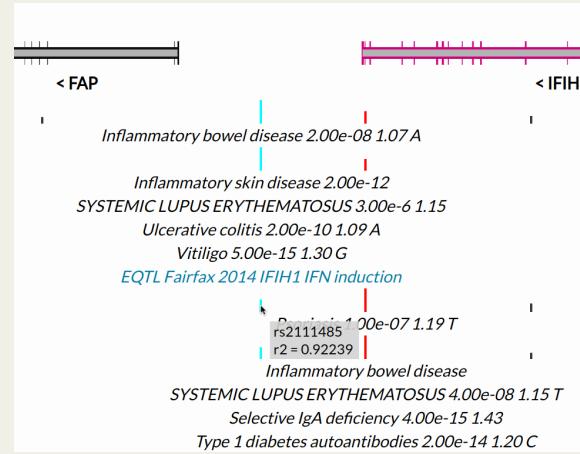
Curated association/eQTL/pQTL results are shown below any gene annotations.



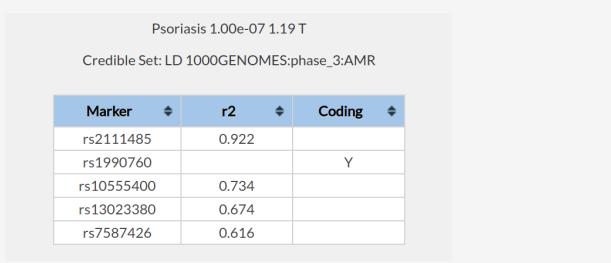
Interactive graphical view of curated data within a genomic context.



Curated association/eQTL/pQTL results are shown below any gene annotations.



Hovering the cursor over a variant's tick mark will display its name and its r² value to the index variant. Additionally, the same variant will be shaded teal where it appears in other association/eQTL/pQTL results.

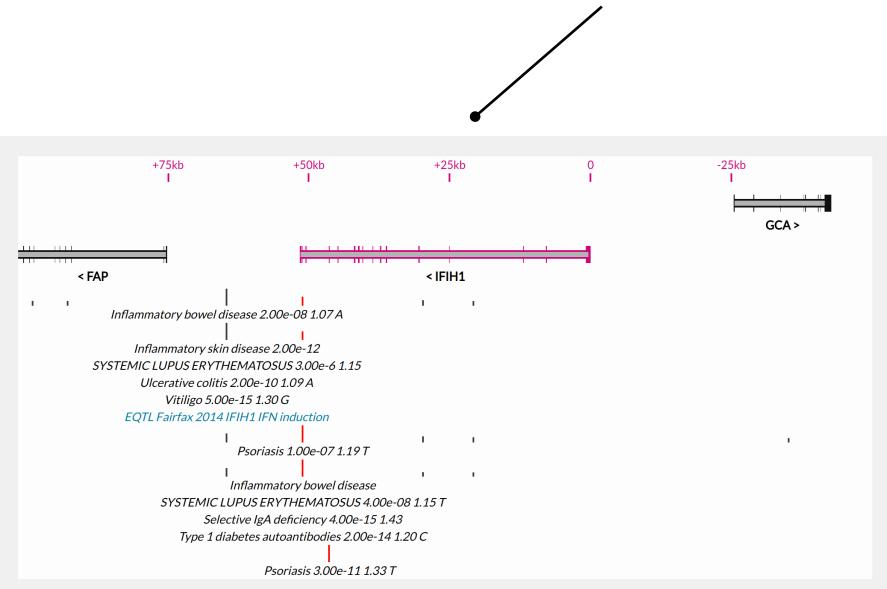


Clicking on the Display Name for an association/eQTL/pQTL result will reveal a dismissable table just below the display area summarizing the index variant and its Credible Set.

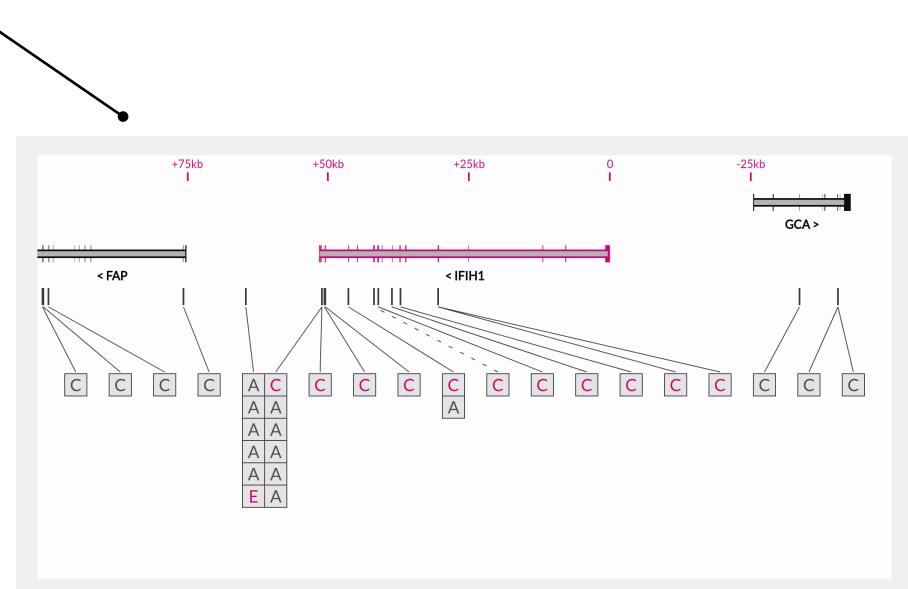
Interactive graphical view of curated data within a genomic context.

Two different graphical perspectives are available for the display area

View Mode Credible Sets LD Summary (0.6 r² threshold, 1KGp3:EUR)



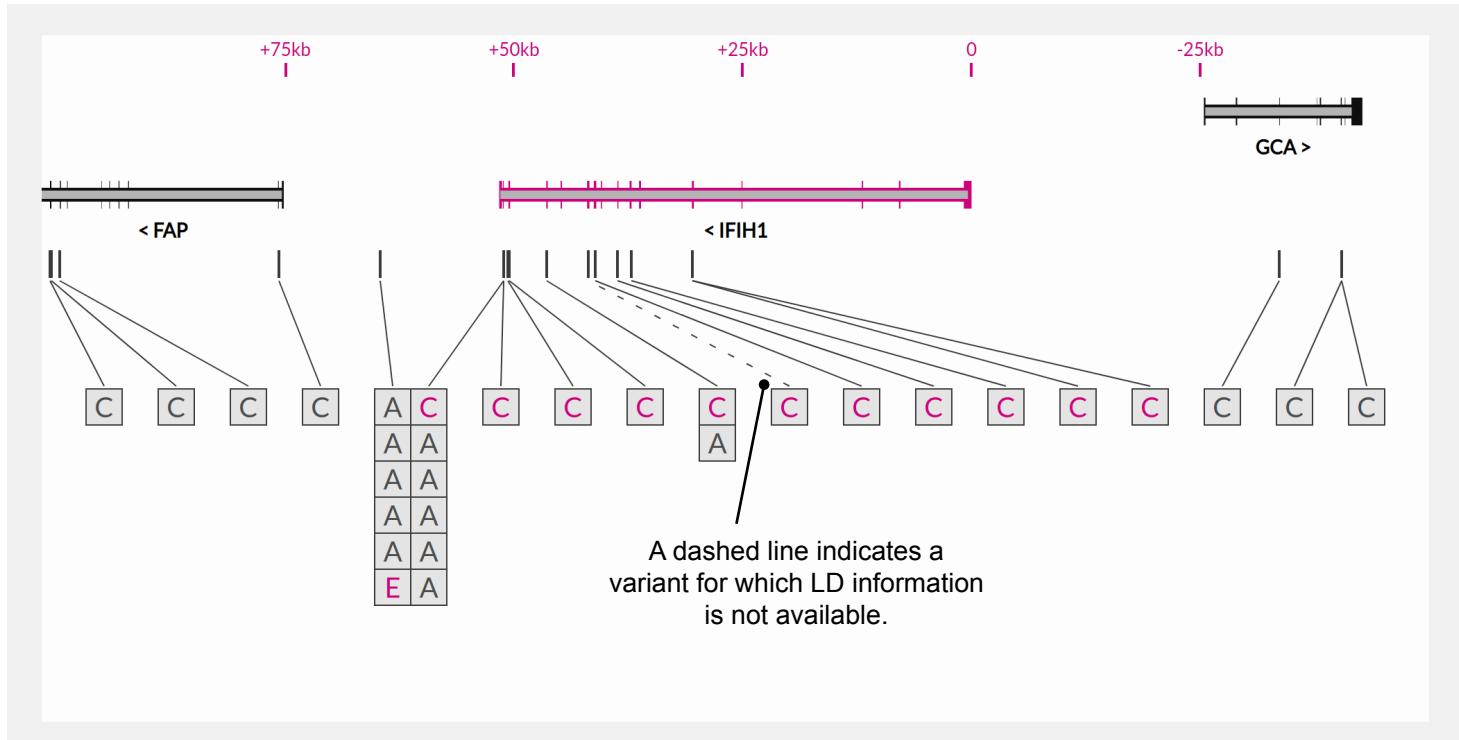
Credible Sets are shown for curated association, eQTL, and pQTL results.



Association, eQTL, and pQTL index variants, as well as other curated variants are shown in high-LD groups.

Interactive graphical view of curated data within a genomic context.

LD-summary graphical perspective



Each box represents a particular association result (A), allele coding impact (C), eQTL result (E), or pQTL result (P). Results for the same variant are stacked vertically.

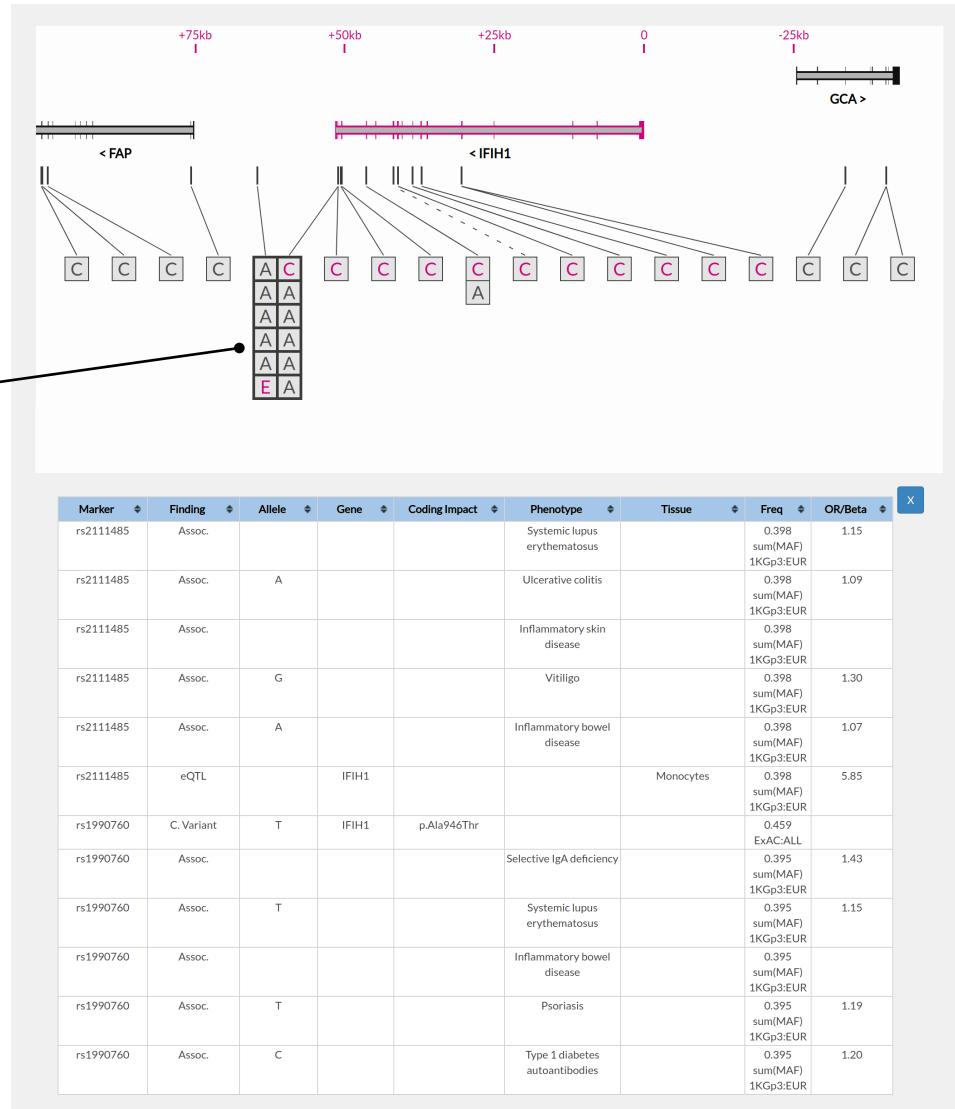
Variants are placed into groups where all members have $r^2 \geq 0.6$ (1KGp3:EUR) to at least one other member of the group.

eQTL results and allele coding impacts assigned to the target gene are highlighted in magenta.

Interactive
graphical view of
curated data within
a genomic context.

LD-summary graphical perspective

Clicking on an LD group will reveal a dismissable table below the display area summarizing the events in that group.



Interactive graphical view of curated data within a genomic context.

Text-based/tabular view of curated data...

Text-based/tabular
view of curated
data.

Overview

Summary (IFIH1 TSS Ensembl 93:chr2:162,318,703)

Association Results

Association Details

pQTL Results

pQTL Details

eQTL Results

▼ Variants of Interest

LD Summary	Name	Non-Reference Allele	Location	Gene	Protein Change	Polphen	f(NFE)	f(Max)
<input checked="" type="checkbox"/>	rs141572054	T	162,359,145	GCA	p.Arg186Ter		0.002	0.012
<input checked="" type="checkbox"/>	rs146161584	G	162,359,091	GCA	p.Ser168Gly	possibly damaging	0.000	0.010
<input type="checkbox"/>	rs758193923	T	162,352,402	GCA	p.Tyr86Phe	probably damaging	0.000	0.001
<input checked="" type="checkbox"/>	rs17783344	G	162,352,383	GCA	p.Ser80Ala	benign	0.126	0.209
<input type="checkbox"/>	rs754396914	C	162,352,371	GCA	p.Cys76Arg	probably damaging	0.000	0.001
<input type="checkbox"/>	rs79565841	T	162,352,367	GCA	p.Gln74His	probably damaging	0.004	0.004
<input type="checkbox"/>	rs545281061	-	162,347,739	GCA	p.Gln64ArgfsTer14		0.000	0.004
<input type="checkbox"/>	rs774170639	A	162,347,656	GCA	p.Asp36Asn	benign	0.000	0.001
<input type="checkbox"/>	rs1403164634	G	162,318,102	IFIH1	p.Gly9Ala	benign	0.000	0.001
<input type="checkbox"/>	rs147278787	A	162,318,079	IFIH1	p.Arg77Trp	possibly damaging	0.001	0.003

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Functional Variants Details

Expression Data Details

Protein and Structure Chemistry Details

Clinical Results

Clinical Data Details

▼ References (18 out of 23 not yet reviewed)

Reviewed	1st Author	Year	Journal/Site/File	Title/Description	Curator Comment
<input checked="" type="checkbox"/>		2019	https://clue.io/repurposing-app	Curated information about drugs and drug targets from Broad.	Enter comment
<input checked="" type="checkbox"/>	Li Q	2017	Pharmacogenet. Genomics	Genome-wide association study of paliperidone efficacy.	Enter comment
<input checked="" type="checkbox"/>		2017	http://www.nealelab.is/blog/2017/7/19/rapid-gwas-of-thousands-of-phenotypes-for-337000-samples-in-the-uk-biobank	UK Biobank Rapid GWAS	Enter comment
<input checked="" type="checkbox"/>	Pettersson M	2017	Am. J. Med. Genet. A	Further evidence for specific IFIH1 mutation as a cause of Singleton-Merten syndrome with phenotypic heterogeneity.	Enter comment
<input checked="" type="checkbox"/>	Gorman JA	2017	Nat. Immunol.	The A946T variant of the RNA sensor IFIH1 mediates an interferon program that limits viral infection but increases the risk for autoimmunity.	Enter comment
<input type="checkbox"/>	Bronson PG	2016	Nat. Genet.	Common variants at PVT1, ATG13-AMBRA1, AHI1 and CLEC16A are associated with selective IgA deficiency.	Enter comment
<input type="checkbox"/>	Morris DL	2016	Nat. Genet.	Genome-wide association meta-analysis in Chinese and European individuals identifies ten new loci associated with systemic lupus erythematosus.	Enter comment
<input type="checkbox"/>	Jin Y	2016	Nat. Genet.	Genome-wide association studies of autoimmune vitiligo identify 23 new risk loci and highlight key pathways and regulatory variants.	Enter comment
<input type="checkbox"/>	Bentham J	2015	Nat. Genet.	Genetic association analyses implicate aberrant regulation of innate and adaptive immunity genes in the pathogenesis of systemic lupus erythematosus.	Enter comment
<input type="checkbox"/>	Yin X	2015	Nat Commun	Genome-wide meta-analysis identifies multiple novel associations and ethnic heterogeneity of psoriasis susceptibility.	Enter comment

Page: 1 ▾ ⏴ 1 to 10 of 23 rows ⏴ ⏵ 10 ▾

Add Pubmed Reference

Add File Reference

Add Web Reference

Add bioRxiv Reference

Text-based/tabular view of curated data.

13 different sections hold curated data

- ▶ Summary (IFIH1 TSS Ensembl 93:chr2:162,318,703)
- ▶ Association Results
- ▶ Association Details
- ▶ pQTL Results
- ▶ pQTL Details
- ▶ eQTL Results
- ▶ Variants of Interest
- ▶ Functional Variants Details
- ▶ Expression Data Details
- ▶ Protein and Structure Chemistry Details
- ▶ Clinical Results
- ▶ Clinical Data Details
- ▶ References (18 out of 23 not yet reviewed)

Text-based/tabular
view of curated
data.

Summary section

The Ensembl version against which the data were curated is shown here, along with the start position of the canonical transcript for the target gene.

▼ Summary (IFIH1 TSS Ensembl 93:chr2:162,318,703)

Enter summary

Text additions/changes in this area are automatically saved to the target-gene database.

Text-based/tabular view of curated data.

Association Results section

Association Results													
Display Name	Phenotype	Source	Year	Index Variant	Allele	Pvalue	OR/Beta	Credible Set	Show	Curator Comment			
Inflammatory bowel disease	Inflammatory bowel disease	Liu JZ; Association analyses identify 38 susceptibility loci for inflammatory bowel disease and highlight shared genetic risk across populations.	2015	rs1990760		4.00e-10		LD 1KGp3:EUR (3)	<input checked="" type="checkbox"/>	Enter comment			
Inflammatory bowel disease 2.00e-08 1.07 A	Inflammatory bowel disease	Jostins L; Host-microbe interactions have shaped the genetic architecture of inflammatory bowel disease.	2012	rs2111485	A	2.00e-08	1.07	LD 1KGp3:AMR (5)	<input checked="" type="checkbox"/>	Enter comment			
Inflammatory skin disease 2.00e-12	Inflammatory skin disease	Baurecht H; Genome-wide comparative analysis of atopic dermatitis and psoriasis genes	2015	rs2111485		2.00e-12		LD 1KGp3:EUR (1)	<input checked="" type="checkbox"/>	Enter comment			
Marder score) 3.00e-06 5.87 T	schizophrenia (negative Marder score)							<p>LD 1KGp3:EUR (1) Unset None LD 1KGp3:AFR (1) LD 1KGp3:AMR (5) LD 1KGp3:EAS (3) LD 1KGp3:EUR (1) LD 1KGp3:SAS (0) Add custom</p>	<input checked="" type="checkbox"/>	Enter comment			
Selective IgA deficiency 7.0e-10 1.49	Immunoglobulin A	Ferreira RC; Association of IFIH1 and other autoimmunity risk alleles with selective IgA deficiency.	2010	rs1990760		7.00e-10	1.49	LD 1KGp3:AMR (4)	<input type="checkbox"/>	Enter comment			

Button to add a new association result (see next page).

Text-based/tabular view of curated data.

Editable Display Name used to label the result in the display area. Changes are automatically saved to the target-gene database.

Alleles with an "Audited" label are those for which strandedness is believed to be expressed as genome-forward-strand, based on data provided.



Toggle to indicate if a result should be shown in the display area (Credible Set must not be "Unset" for a result to be shown).

Editable curator comment. Changes are automatically saved to the target-gene database.

Page: 1 ▾ ⏪ ⏩ 1 to 10 of 36 rows ⏪ ⏩ 10 ▾

Association Results section

Adding an association result

Desired reference must already have been added to the References section to appear in the Source dropdown menu (see later description of the References section).

Please select

Barrett JC; Genome-wide association study and meta-analysis find that over 40 loci affect risk of type 1 diabetes.

Baurecht H; Genome-wide comparative analysis of atopic dermatitis and psoriasis gives insight into opposing genetic mechanisms.

Bentham J; Genetic association analyses implicate aberrant regulation of innate and adaptive immunity genes in the pathogenesis of systemic lupus erythematosus.

Bronson PG; Common variants at PVT1, ATG13-AMBRA1, AHI1 and CLEC16A are associated with selective IgA deficiency.

Curated information about drugs and drug targets from Broad.

Add Association

New Association

Phenotype: Eg. Schizophrenia

Source: Please select

Index variant: Eg. rs123

Allele (optional): Eg. T

Pvalue: Eg. 3.0e-13

OR/Beta: Eg. 1.11

Enter comment

Comment:

Save

highlights the role of innate immunity.

New row added to association-results table.

Button to remove association result.

Text-based/tabular view of curated data.

Association Results section

Creating a custom Credible Set

Association Results													
Display Name	Phenotype	Source	Year	Index Variant	Allele	Pvalue	OR/Beta	Credible Set	Show	Curator Comment			
Inflammatory bowel disease	Inflammatory bowel disease	Liu JZ; Association analyses identify 38 susceptibility loci for inflammatory bowel disease and highlight shared genetic risk across populations.	2015	rs1990760		4.00e-10		LD 1KGp3:EUR (3)	<input checked="" type="checkbox"/>	Enter comment			
Inflammatory bowel disease 2.00e-08 1.07 A	Inflammatory bowel disease	Jostins L; Host-microbe interactions have shaped the genetic architecture of inflammatory bowel disease.	2012	rs2111485	A	2.00e-08	1.07	LD 1KGp3:AMR (5)	<input checked="" type="checkbox"/>	Enter comment			
Inflammatory skin disease 2.00e-12	Inflammatory skin disease	Baurecht H; Genome-wide comparative analysis of atopic dermatitis and psoriasis gives	2015	rs2111485		2.00e-12		LD 1KGp3:EUR (1)	<input checked="" type="checkbox"/>	Enter comment			

Custom Credible Set

Name of credible set: MyCredibleSet

Index-variant (rs2111485) posterior: 0.87

Other credible-set members:

Marker	Posterior
rs1990760	0.11
Eg. rs123	Eg. 0.123

Add Row Create Credible Set

LD 1KGp3:EUR (1)
Unset
None
LD 1KGp3:AFR (1)
LD 1KGp3:AMR (5)
LD 1KGp3:EAS (3)
LD 1KGp3:EUR (1)
LD 1KGp3:SAS (0)
MyCredibleSet (1)
Delete custom

The custom Credible Set and ability to delete the custom Credible Set are now selectable options in the dropdown menu.

Select "Add custom" dropdown option.

LD 1KGp3:EUR (1)
Unset
None
LD 1KGp3:AFR (1)
LD 1KGp3:AMR (5)
LD 1KGp3:EAS (3)
LD 1KGp3:EUR (1)
LD 1KGp3:SAS (0)
Add custom

Define custom Credible Set.

Text-based/tabular view of curated data.

pQTL Results and eQTL Results sections

These sections are very similar to the Association Results section

eQTL Results													
Display Name	Tissue	Gene	Source	Year	Index Variant	Pvalue	Beta	Effect Allele	Credible Set	Show	Curator Comment		
EQTL GTex FAP Nerve Tibial 2.55e-05	Nerve Tibial	FAP	GTEx Consortium; Human genomics. The Genotype-Tissue Expression (GTEx) pilot analysis: multitissue gene regulation in humans.	2015	rs3788967	2.55e-05	0.56	Unset		<input type="checkbox"/>	Tissue pval threshold = 4.11e-05	0	
EQTL GTex FAP Artery Tibial 5.19e-07	Artery Tibial	FAP	GTEx Consortium; Human genomics. The Genotype-Tissue Expression (GTEx) pilot analysis: multitissue gene regulation in humans.	2015	rs3827491	5.19e-07	0.43	Unset		<input type="checkbox"/>	Tissue pval threshold = 3.54e-05	0	
EQTL GTex FAP Cells Transformed fibroblasts 1.67e-06	Cells Transformed fibroblasts	FAP	GTEx Consortium; Human genomics. The Genotype-Tissue Expression (GTEx) pilot analysis: multitissue gene regulation in humans.	2015	rs6746339	1.67e-06	0.34	Unset		<input type="checkbox"/>	Tissue pval threshold = 3.54e-05	0	
EQTL GTex FAP Cells Transformed fibroblasts 7.80e-06	Cells Transformed fibroblasts	FAP	GTEx Consortium; Human genomics. The Genotype-Tissue Expression (GTEx) pilot analysis: multitissue gene regulation in humans.	2015	rs12468578	7.80e-06	0.36	Unset		<input type="checkbox"/>	Tissue pval threshold = 3.54e-05	0	
EQTL GTex FAP Thyroid 2.67e-05	Thyroid	FAP	GTEx Consortium; Human genomics. The Genotype-Tissue Expression (GTEx) pilot analysis: multitissue gene regulation in humans.	2015	rs2389683	2.67e-05	0.45	Unset		<input type="checkbox"/>	Tissue pval threshold = 3.29e-05	0	
EQTL GTex FAP Thyroid 2.91e-05	Thyroid	FAP	GTEx Consortium; Human genomics. The Genotype-Tissue Expression (GTEx) pilot analysis: multitissue gene regulation in humans.	2015	rs11455810	2.91e-05	0.44	Unset		<input type="checkbox"/>	Tissue pval threshold = 3.29e-05	0	
EQTL GTex FAP Thyroid 4.53e-06	Thyroid	FAP	GTEx Consortium; Human genomics. The Genotype-Tissue Expression (GTEx) pilot analysis: multitissue gene regulation in humans.	2015	rs16846600	4.53e-06	0.51	Unset		<input type="checkbox"/>	Tissue pval threshold = 3.29e-05	0	
EQTL GTex FAP Muscle Skeletal 1.58e-05	Muscle Skeletal	FAP	GTEx Consortium; Human genomics. The Genotype-Tissue Expression (GTEx) pilot analysis: multitissue gene regulation in humans.	2015	rs35544136	1.58e-05	0.67	Unset		<input type="checkbox"/>	Tissue pval threshold = 1.85e-05	0	
EQTL GTex GCA Adrenal Gland 6.34e-06	Adrenal Gland	GCA	GTEx Consortium; Human genomics. The Genotype-Tissue Expression (GTEx) pilot analysis: multitissue gene regulation in humans.	2015	rs34977319	6.34e-06	-0.77	Unset		<input type="checkbox"/>	Tissue pval threshold = 1.75e-05	0	
EQTL Fairtax 2014 IFIH1 IFN induction	Monocytes	IFIH1	Fairfax BP; Innate immune activity conditions the effect of regulatory variants upon monocyte gene expression.	2014	rs2111485	1.19e-08	5.85	LD 1KGp3:EUR (1)	<input checked="" type="checkbox"/>	Enter comment	X	10	

The eQTL Results table contains an extra column showing the count of displayed association and pQTL results that have intersecting Credible-Set members with the 1KGp3:EUR Credible Set of each eQTL result.

Text-based/tabular view of curated data.

Variants of Interest section

In this section are listed those variant alleles which confer a coding or likely splice change to any gene in the neighborhood of the target gene

▼ Variants of Interest

LD Summary	Name	Non-Reference Allele	Location	Gene	Protein Change	Polyphen	f(NFE)	f(Max)
<input checked="" type="checkbox"/>	rs141572054	T	162,359,145	GCA	p.Arg186Ter		0.002	0.012
<input checked="" type="checkbox"/>	rs146161584	G	162,359,091	GCA	p.Ser168Gly	possibly damaging	0.000	0.010
<input type="checkbox"/>	rs758193923	T	162,352,402	GCA	p.Tyr86Phe	probably damaging	0.000	0.001
<input checked="" type="checkbox"/>	rs17783344	G	162,352,383	GCA	p.Ser80Ala	benign	0.126	0.209
<input type="checkbox"/>	rs754396914	C	162,352,371	GCA	p.Cys76Arg	probably damaging	0.000	0.001
<input type="checkbox"/>	rs79565841	T	162,352,367	GCA	p.Gln74His	probably damaging	0.004	0.004
<input type="checkbox"/>	rs545281061	-	162,347,739	GCA	p.Gln64ArgfsTer14		0.000	0.004
<input type="checkbox"/>	rs774170639	A	162,347,656	GCA	p.Asp36Asn	benign	0.000	0.001
<input type="checkbox"/>	rs1403164634	G	162,318,102	IFIH1	p.Gly69Ala	benign	0.000	0.001
<input type="checkbox"/>	rs147278787	A	162,318,079	IFIH1	p.Arg77Trp	possibly damaging	0.001	0.003

Page: 1 ▼ 1 to 10 of 68 rows ⏪ ⏩ 10 ▼



Toggle for inclusion of the variant in the LD-Summary perspective of the display area.

To be included in this table, an allele must have a frequency of at least 0.001 in one of the ExAC sub-populations.

Text-based/tabular view of curated data.

Clinical Results section

▼ Clinical Results

Phenotype/Gene Associations					Curator Comment
Phenotype	Gene	Source			
SINGLETON-MERTEN SYNDROME 1	IFIH1	MIM morbid	Enter comment		
Aicardi-Goutieres syndrome 7	IFIH1	MIM morbid	Enter comment		

Page: 1 ▼ 1 to 2 of 2 rows 10 ▼

 Editable curator comment. Changes are automatically saved to the target-gene database.

Phenotype/Allele Associations						
Phenotype	Variant	Unaudited Risk Allele	Clinical Significance	Transcript-Impact Genes	Source	
SINGLETON-MERTEN SYNDROME 1	rs6748554	G	benign	IFIH1	ClinVar	
SINGLETON-MERTEN SYNDROME 1	rs13418718	A	benign	IFIH1	ClinVar	
SINGLETON-MERTEN SYNDROME 1	rs35337543	G	benign	IFIH1	ClinVar	
SINGLETON-MERTEN SYNDROME 1	rs35667974	C	benign	IFIH1	ClinVar	
SINGLETON-MERTEN SYNDROME 1	rs41399348	T	benign	IFIH1	ClinVar	
SINGLETON-MERTEN SYNDROME 1	rs74162087	T	uncertain significance	IFIH1	ClinVar	
SINGLETON-MERTEN SYNDROME 1	rs74162089	A	benign	IFIH1	ClinVar	
SINGLETON-MERTEN SYNDROME 1	rs79324540	T	uncertain significance	IFIH1	ClinVar	
SINGLETON-MERTEN SYNDROME 1	rs140562355	T	benign	IFIH1	ClinVar	
SINGLETON-MERTEN SYNDROME 1	rs143870870	C	benign	IFIH1	ClinVar	

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Text-based/tabular view of curated data.

References section

▼ References (18 out of 23 not yet reviewed)								
Reviewed	1st Author	Year	Journal/Site/File	Title/Description	Curator Comment			
<input checked="" type="checkbox"/>		2019	https://clue.io/repurposing-app	Curated information about drugs and drug targets from Broad.	Enter comment	Show/Add Details	X	
<input checked="" type="checkbox"/>	Li Q	2017	Pharmacogenet. Genomics	Genome-wide association study of paliperidone efficacy.	Enter comment	Show/Add Details		
<input checked="" type="checkbox"/>		2017	http://www.nealelab.is/blog/2017/7/19/rapid-gwas-of-thousands-of-phenotypes-for-337000-samples-in-the-uk-biobank	UK Biobank Rapid GWAS	Enter comment	Show/Add Details	X	
<input checked="" type="checkbox"/>	Pettersson M	2017	Am. J. Med. Genet. A	Further evidence for specific IFIH1 mutation as a cause of Singleton-Merten syndrome with phenotypic heterogeneity.	Enter comment	Show/Add Details	X	
<input checked="" type="checkbox"/>	Gorman JA	2017	Nat. Immunol.	The A946T variant of the RNA sensor IFIH1 mediates an interferon program that limits viral infection but increases the risk for autoimmunity.	Enter comment	Show/Add Details (1)	X	
<input type="checkbox"/>	Bronson PG	2016	Nat. Genet.	Common variants at PVT1, ATG13-AMBRA1, AH1 and CLEC16A are associated with selective IgA deficiency.	Enter comment	Show/Add Details		
<input type="checkbox"/>	Morris DL	2016	Nat. Genet.	Genome-wide association meta-analysis in Chinese and European individuals identifies ten new loci associated with systemic lupus erythematosus.	Enter comment	Show/Add Details		
<input type="checkbox"/>	Jin Y	2016	Nat. Genet.	Genome-wide association studies of autoimmune vitiligo identify 23 new risk loci and highlight key pathways and regulatory variants.	Enter comment	Show/Add Details		
<input type="checkbox"/>	Bentham J	2015	Nat. Genet.	Genetic association analyses implicate aberrant regulation of innate and adaptive immunity genes in the pathogenesis of systemic lupus erythematosus.	Enter comment	Show/Add Details		
<input type="checkbox"/>	Yin X	2015	Nat Commun	Genome-wide meta-analysis identifies multiple novel associations and ethnic heterogeneity of psoriasis susceptibility.	Enter comment	Show/Add Details		

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[Add Pubmed Reference](#)

[Add File Reference](#)

[Add Web Reference](#)

[Add bioRxiv Reference](#)

Checkboxes to declare references having been reviewed.

Buttons to add new references of different types.

Editable curator comment.
Changes are automatically saved to the target-gene database.

Button to remove user-added references.

Toggles to show/hide Detail elements attached to each reference (see next page). Number of Detail elements shown in parentheses.

Text-based/tabular view of curated data.

References section

Graphic elements (Details) can be saved with each reference

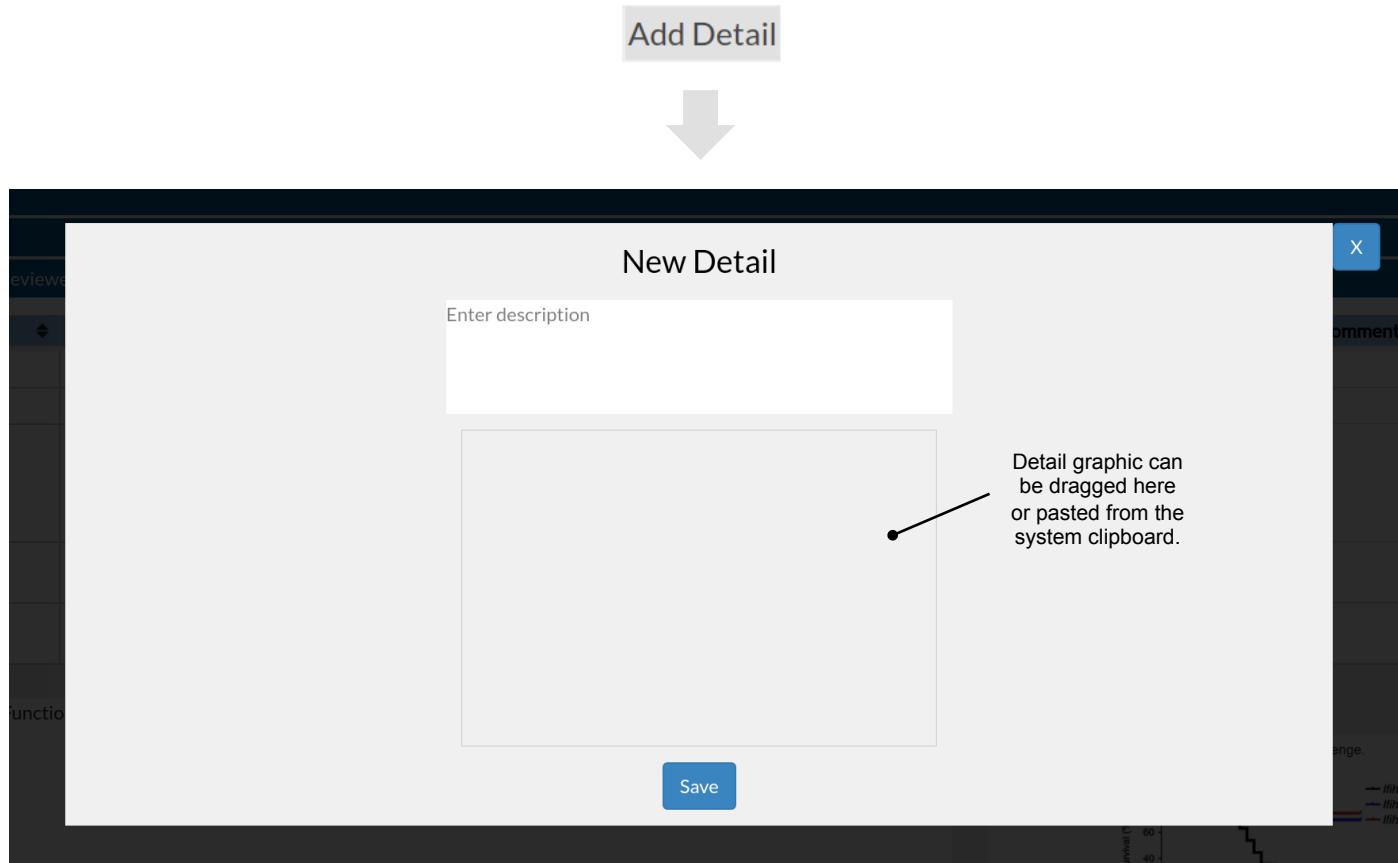
▼ References (18 out of 23 not yet reviewed)								
Reviewed	1st Author	Year	Journal/Site/File	Title/Description	Curator Comment			
<input checked="" type="checkbox"/>		2019	https://clue.io/repurposing-app	Curated information about drugs and drug targets from Broad.	Enter comment	Show/Add Details	X	
<input checked="" type="checkbox"/>	Li Q	2017	Pharmacogenet. Genomics	Genome-wide association study of paliperidone efficacy.	Enter comment	Show/Add Details		
<input checked="" type="checkbox"/>		2017	http://www.nealab.is/blog/2017/7/19/rapid-gwas-of-thousands-of-phenotypes-for-337000-samples-in-the-uk-biobank	UK Biobank Rapid GWAS	Enter comment	Show/Add Details	X	
<input checked="" type="checkbox"/>	Pettersson M	2017	Am. J. Med. Genet. A	Further evidence for specific IFIH1 mutation as a cause of Singleton-Merten syndrome with phenotypic heterogeneity.	Enter comment	Show/Add Details	X	
<input checked="" type="checkbox"/>	Gorman JA	2017	Nat. Immunol.	The A946T variant of the RNA sensor IFIH1 mediates an interferon program that limits viral infection but increases the risk for autoimmunity.	Enter comment	Hide Details (1)		
<p>Functional data linking rs1990760 (A946T) and protection from infection challenge in mice.</p> <p>Section Assignment <input type="button" value="Functional"/></p> <p>Editable description field. Changes are automatically saved to the target-gene database.</p> <p>Zoomable thumbnail of Detail.</p> <p>Figure 4: <i>Ifih1^R</i> mice exhibit protection from EMCV challenge.</p> <p>Remove</p> <p>Button to remove Detail</p> <p>Button to add new Detail (see next page). <input type="button" value="Add Detail"/></p>								
<input type="checkbox"/>	Bronson PG	2016	Nat. Genet.	Common variants at PVT1, ATG13-AMBRA1, AHI1 and CLEC16A are associated with selective IgA deficiency.	Enter comment	Show/Add Details		
<input type="checkbox"/>	Morris DL	2016	Nat. Genet.	Genome-wide association meta-analysis in Chinese and European individuals identifies ten new loci associated with systemic lupus erythematosus.	Enter comment	Show/Add Details		
<input type="checkbox"/>	Jin Y	2016	Nat. Genet.	Genome-wide association studies of autoimmune vitiligo identify 23 new risk loci and highlight key pathways and regulatory variants.	Enter comment	Show/Add Details		
<input type="checkbox"/>	Bentham J	2015	Nat. Genet.	Genetic association analyses implicate aberrant regulation of innate and adaptive immunity genes in the pathogenesis of systemic lupus erythematosus.	Enter comment	Show/Add Details		
<input type="checkbox"/>	Yin X	2015	Nat Commun	Genome-wide meta-analysis identifies multiple novel associations and ethnic heterogeneity of psoriasis susceptibility.	Enter comment	Show/Add Details		

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Text-based/tabular view of curated data.

References section

Adding a Detail



Text-based/tabular view of curated data.

Association Details, pQTL Details, Functional Variants Details, Expression Data Details, Protein and Structure Chemistry Details, and Clinical Data Details sections

Each section contains those Details assigned to that particular section, organized by reference

▼ Protein and Structure Chemistry Details			
1st Author	Year	Journal/Site/File	Title/Description
Wu B	2013	Cell	Structural basis for dsRNA recognition, filament formation, and antiviral signal activation by MDA5.
<p>Figure 7. Oligomerization of 2CARD in the Full-Length MDA5 Filament</p> <p>B</p> <p>I II III</p> <p>Periodic path distance: 22 nm (200 nm) Periodic path width: 4.5 nm Periodic path length: 8 nm</p> <p>Initial trajectory: ~5 nm Helical trajectory: ~1.5 nm Full trajectory: <25 nm (100 nm)</p> <p><5 monomers 6.5 nm (helical trajectory) <11 monomers</p>			
<p>MDA5 homologs</p> <p>Editable description field. Changes are automatically saved to the target-gene database.</p>			
<p>MDA5 filament formation</p> <p>MDA5 RIG-I</p> <p>dsRNA dsRNA stem-binding cooperative filament assembly (head-to-tail) tandem CARD oligomerization MAVS monomer MAVS filament antiviral signaling</p>			

Page: 1 ▼ 1 to 1 of 1 rows 10 ▼

Text-based/tabular view of curated data.

Notebook utilities...

Notebook utilities.

Overview page of available Target Gene Notebooks

This is the TGN server's index.html page

The screenshot shows the 'Target Gene Notebook' index page at localhost:8083/index.html. The page has a header with 'User Guide', 'Manage Tags', and 'About' links. Below the header is a navigation bar with three tabs: 'IBD' (highlighted in blue), 'Chol Metab' (grey), and 'CAD' (pink). A callout box labeled 'Links to available Target Gene Notebooks.' points to the table below. Another callout box labeled 'Tags assigned to each Target Gene Notebook' points to the 'Tags' column. A third callout box labeled 'Toggles to filter TGN list according to assigned Tags.' points to the three tabs. The table lists two entries:

Gene	Chr	Mb Location	Neighborhood Coding Genes	References Status	Last Modified Time	Tags
IFIH1	2	162.3	FAP GCA GCG KCNH7	18 out of 23 not reviewed	2019-04-11T19:04:36.353346Z	IBD
PCSK9	1	55.1	BSND TMEM61 USP24	22 out of 22 not reviewed	2019-04-13T18:43:48.814601Z	Chol Metab CAD

Notebook utilities.

Tag-management tools

Manage Tags

The diagram illustrates the relationship between tag management and the Target Gene Notebook interface. A central 'Manage Tags' button is connected by a downward arrow to the 'Target Gene Notebook' interface. The interface features a header with 'Target Gene Notebook', 'User Guide', and 'About' links. Below the header is a table titled 'Existing Tags' containing three entries: CAD (Disease association), Chol Metab (Pathway), and IBD (Disease association). Each entry includes columns for Tag, Tag Class, Short Name, Long Name, Description, and Edit/Delete buttons. Below the table is a 'New Tag' button. The interface also includes sections for 'Assign by Tag', 'Push Web Reference to Notebooks', and 'Assign by TGN', each with its own descriptive text and associated icons.

Tag	Tag Class	Short Name	Long Name	Description	
CAD	Disease association	CAD	Coronary artery disease		Edit Delete
Chol Metab	Pathway	Chol Metab	Cholesterol metabolism		Edit Delete
IBD	Disease association	IBD	Inflammatory Bowel Disease		Edit Delete

Existing Tags

Button to create a new Tag. New Tag

Assign by Tag

Assign by TGN

Push Web Reference to Notebooks

Button to assign specific Tags to Target Gene Notebooks.

Button to add web references to notebooks *en masse*.

Button to assign specific Target Gene Notebooks to Tags.

Notebook utilities.

Tag-management tools

Creating a Tag

New Tag

Existing Tags

Create/Edit Tag

Class:

Short name: Eg. Tag Name

Long name: Eg. Longer Tag Name

Description:

Save

Tag Color

Color selector window:

- Tag Color button
- Color palette grid
- Close button (X)

Annotations:

- A callout box points from the "New Tag" button to the "Create/Edit Tag" form.
- A callout box points from the "Class" dropdown to its description: "Dropdown menu to select Tag class, which determines Tag shape." It also points to the "Please select" button in the dropdown.
- A callout box points from the "Tag Color" button to its description: "Button to open color selector." It also points to the color palette grid and the close button.

The screenshot shows a 'Create/Edit Tag' interface. At the top, there's a 'Class:' dropdown menu with options like 'Please select', 'Collaboration', 'Disease association', etc. Below it are fields for 'Short name:' (Eg. Tag Name) and 'Long name:' (Eg. Longer Tag Name). There's a large text area for 'Description:' and a 'Save' button at the bottom. A 'Tag Color' button is located next to the 'Long name:' field. A color selector dialog is open, showing a grid of colors and a close button. Annotations with callout boxes explain the 'Class:' dropdown and the 'Tag Color' button.

Notebook utilities.

Tag-management tools

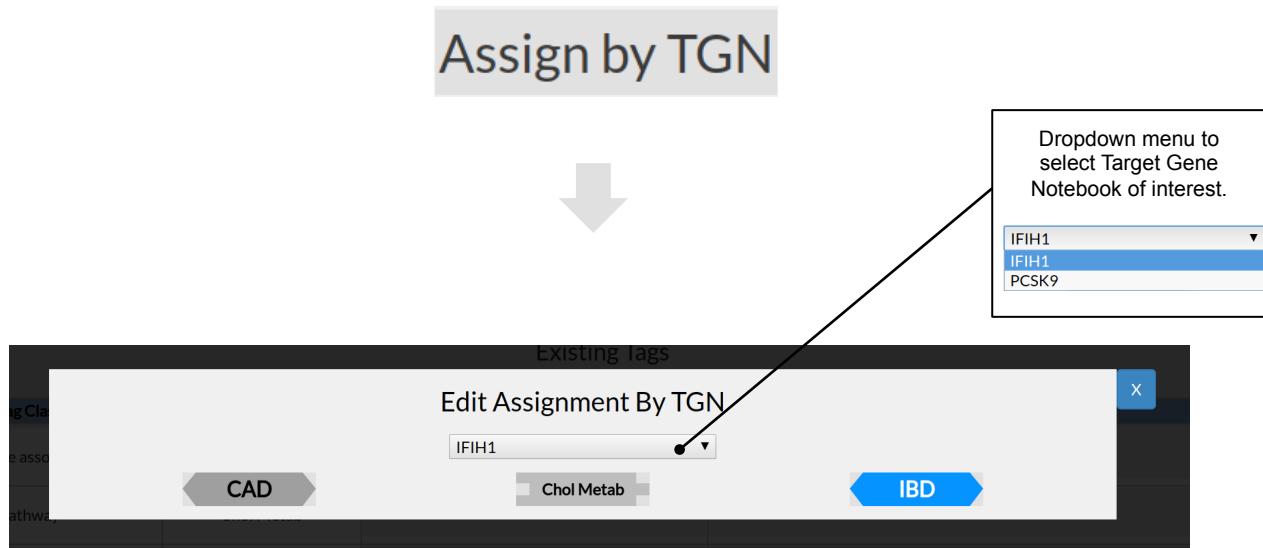
Assigning specific Tags to Target Gene Notebooks



Use Notebook buttons to toggle Tag assignment: if the Notebook button is the same color as the Tag, then that Tag is assigned to the Notebook.

Tag-management tools

Assigning specific Target Gene Notebooks to Tags

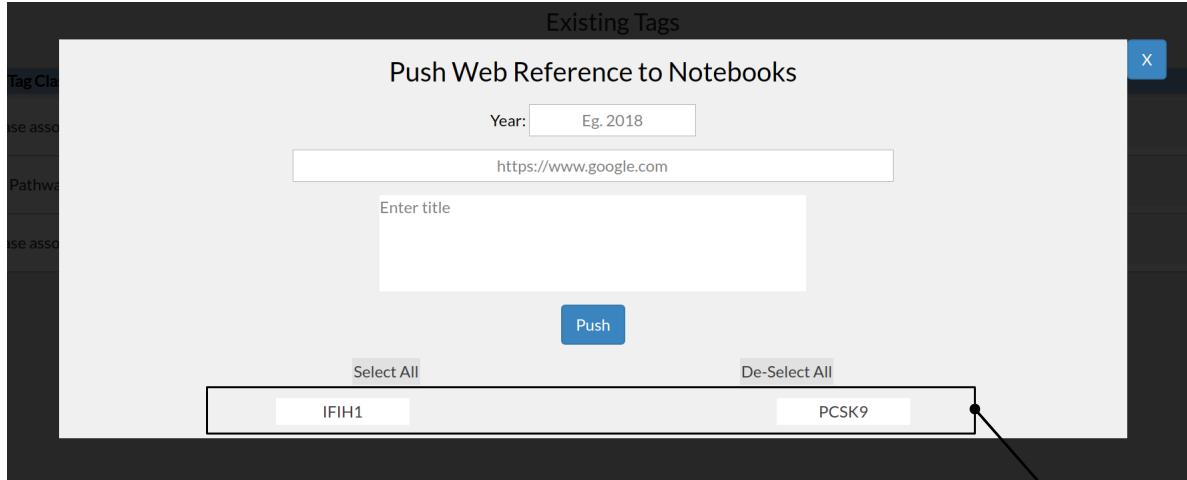


Use Tag buttons to toggle Tag assignment: if the Tag button is the same color as the Tag, then the Notebook is assigned to that Tag.

Tag-management tools

Pushing a web reference to existing Notebooks
(yes, we know this is technically not Tag management)

Push Web Reference to Notebooks



Toggle buttons to indicate Notebooks to receive the web reference. Those Notebooks shaded blue will be sent the reference.

Notebooks that already have the web reference will ignore the push.

Notebook utilities.