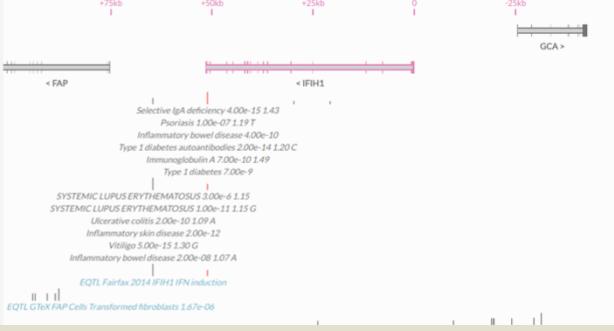


 Target Gene Notebook 1.0:1.0

About Terms Contact FAQ User Guide



Selective IgA deficiency 4.00e-15 1.43
 Porphyria 1.00e-07 1.19T
 Inflammatory bowel disease 2.00e-10 1.20C
 Type 1 diabetes associated variants 2.00e-14 1.20C
 Immunoglobulin A 7.00e-10 1.49
 Type 1 diabetes 7.00e-09

SYSTEMIC LUPUS ERYTHEMATOSUS 3.00e-6 1.15
 SYSTEMIC LUPUS ERYTHEMATOSUS 1.00e-11 1.15 G
 Ulcerative colitis 2.00e-10 1.09 A
 Inflammatory skin disease 2.00e-12
 Vitiligo 3.00e-15 1.30 G
 Inflammatory bowel disease 2.00e-08 1.07 A

EQTL Fairfax 2014 IFIH1 IFN induction 1.67e-06

Target Gene Notebook

Selective IgA deficiency 4.00e-15 1.43

Credible Set LD 1000GENOMES phase_3_EUR

Marker	r2	θ	Coding
rs2111485	0.889		
rs1990760		Y	
rs10255400	0.628		
rs13023380	0.612		

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

Summary IFIH1 TSS Ensembl 84chr2:142,318,709

Association Results

eQTL Results

Variants of Interest

LD Summary

Name	Non-Reference Allele	Location	Gene	Protein Change	Polyphen	R2 (FDR)	R2 (Max)
rs14726767	A	140,238,079	IFIH1	p.Arg777Trp	possibly damaging	0.001	0.003
rs146721166	C	162,317,872	IFIH1	p.Ile149Val	benign	0.000	0.004
rs41612073	C	162,330,909	IFIH1	p.Gln686Arg	probably damaging	0.001	0.001
rs2629664	C	162,288,184	IFIH1	p.Lys349Arg	benign	0.001	0.020
rs130317197	T	162,288,164	IFIH1	p.Pro354Thr	benign	0.001	0.001
rs117668083						0.000	0.008
rs140977021						0.000	0.004
rs145320044						0.001	0.001
rs10930046						0.013	0.426
rs147000317						0.000	0.008

Page: 1 2 3 ... 1 to 10 of 27 rows

Functional Variants

Expression Data

Protein and Structure Chemistry

Clinical Data

References

1st Author	Year	Journal/Site/File	Title/Description	Show/Add Details
Fairtan BP	2014	Science	Innate immune activity conditions the effect of regulatory variants upon monocyte gene expression.	Show/Add Details
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 autoimmune diseases.	Show/Add Details (1)
Pettersson M	2017	Am. J. Med. Genet. A	Further evidence for specific IFIH1 mutations as a cause of Singleton-Merten syndrome with phenotypic heterogeneity.	Show/Add Details
Wu B	2013	Cell	Structural basis for dsRNA recognition, filament formation, and local signal activation by MDAs.	Show/Add Details (2)
Barnett JC	2009	Nat. Genet.	Genome-wide association study and meta-analysis find that over 40 loci affect risk of type 1 diabetes.	Show/Add Details
Bauchtel H	2013	Am. J. Hum. Genet.	Genome-wide comparative analysis of atopic dermatitis and psoriasis gives insight into overlapping genetic mechanisms.	Show/Add Details
Bentham J	2013	Nat. Genet.	Genetic association analyses implicate aberrant regulation of innate and adaptive immunity genes in the pathogenesis of systemic lupus erythematosus.	Show/Add Details
Bronson PG	2016	Nat. Genet.	Common variants at PVT1, LATS1, AMBRA1, ANET1 and CLEC11A are associated with selective IgA deficiency.	Show/Add Details
Forrester RC	2010	Nat. Genet.	Association of IFIH1 and other autoimmunity risk alleles with selective IgA deficiency.	Show/Add Details
GTEx Consortium	2015	Science	Human genomics: The Genotype-Tissue Expression (GTEx) pilot analysis: multilevel gene regulation in humans.	Show/Add Details (1)

Page: 1 2 3 ... 1 to 10 of 17 rows Add Pubmed Reference Add File Reference Add Web Reference Add bioRxiv Reference

TargetGeneNotebook@eisai.com

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

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

1. Andrew Yates, Wasiu Akanni, M. Ridwan Amode, Daniel Barrell, Konstantinos Billis, Denise Carvalho-Silva, Carla Cummins, Peter Clapham, Stephen Fitzgerald, Laurent Gil, Carlos García Girón, Leo Gordon, Thibaut Hourlier, Sarah E. Hunt, Sophie H. Janacek, Nathan Johnson, Thomas Juettemann, Stephen Keenan, Ilias Lavidas, Fergal J. Martin, Thomas Maurel, William McLaren, Daniel N. Murphy, Rishi Nag, Michael Nuhn, Anne Parker, Mateus Patrício, Miguel Pignatelli, Matthew Rahtz, Harpreet Singh Riat, Daniel Sheppard, Kieron Taylor, Anja Thörmann, Alessandro Vullo, Steven P. Wilder, Amonida Zadissa, Ewan Birney, Jennifer Harrow, Matthieu Muffato, Emily Perry, Magali Ruffier, Giulietta Spudich, Stephen J. Trevanion, Fiona Cunningham, Bronwen L. Aken, Daniel R. Zerbino, Paul Flicek Ensembl 2016.

Nucleic Acids Res. 2016 44 Database issue:D710-6.

Requirements

Server:

Java >=1.7

Client:

Google Chrome

JavaScript enabled

Cookies enabled

Target Gene Notebook uses these libraries/frameworks:

jQuery 2.2.4 (MIT license)

FileSaver.js 1.3.2 (MIT license)

tablesorter 2.28.14 (MIT license)

Select2 4.0.3 (MIT license)

Lightbox2 2.9.0 (MIT license)

jQuery UI 1.12.1 (MIT license)

Bootstrap 3.3.7 (MIT license, ©2016 Twitter)

Spark micro framework (sparkjava.jcom, Apache 2 license)

Dependencies:

sqlite-jdbc 3.7.2

gson 2.7

commons-lang3 3.4

commons-io 2.4

jsoup 1.8.1

slf4j-simple 1.7.21

GUI Overview

The screenshot displays the Target Gene Notebook interface. At the top, there's a navigation bar with links to About, Terms, Contact, FAQ, and User Guide. Below the navigation is a genomic track for the *IFIH1* gene, showing its location relative to other genes (+75kb, +50kb, +25kb, 0, -25kb) and its transcription direction (< FAP). The track highlights several SNPs associated with various diseases:

- Selective IgA deficiency: 4.00e-15 1.43
- Pсоріаз: 1.00e-07 1.19 T
- Inflammatory bowel disease: 4.00e-10
- Type 1 diabetes autoantibodies: 2.00e-14 1.20 C
- Immunoglobulin A: 7.00e-10 1.49
- Type 1 diabetes: 7.00e-9

Below the track, a legend indicates the color coding for different association studies: SYSTEMIC LUPUS ERYTHEMATOSUS (blue), SYSTEMIC LUPUS ERYTHEMATOSUS (red), Ulcerative colitis (green), Inflammatory skin disease (orange), Vitiligo (purple), Inflammatory bowel disease (yellow), and EQTL, Fairfax 2014 (pink).

The main content area shows a table titled "Variants of Interest" with columns for LD Summary, Name, Non-Reference Allele, Location, Gene, Protein Change, Polyphen, RH(FE), and RH(Max). The table lists 17 variants, including rs147278787, rs146721166, rs74162075, rs72450664, rs130317197, rs176080883, rs340977021, rs145520044, rs109300466, and rs147000317.

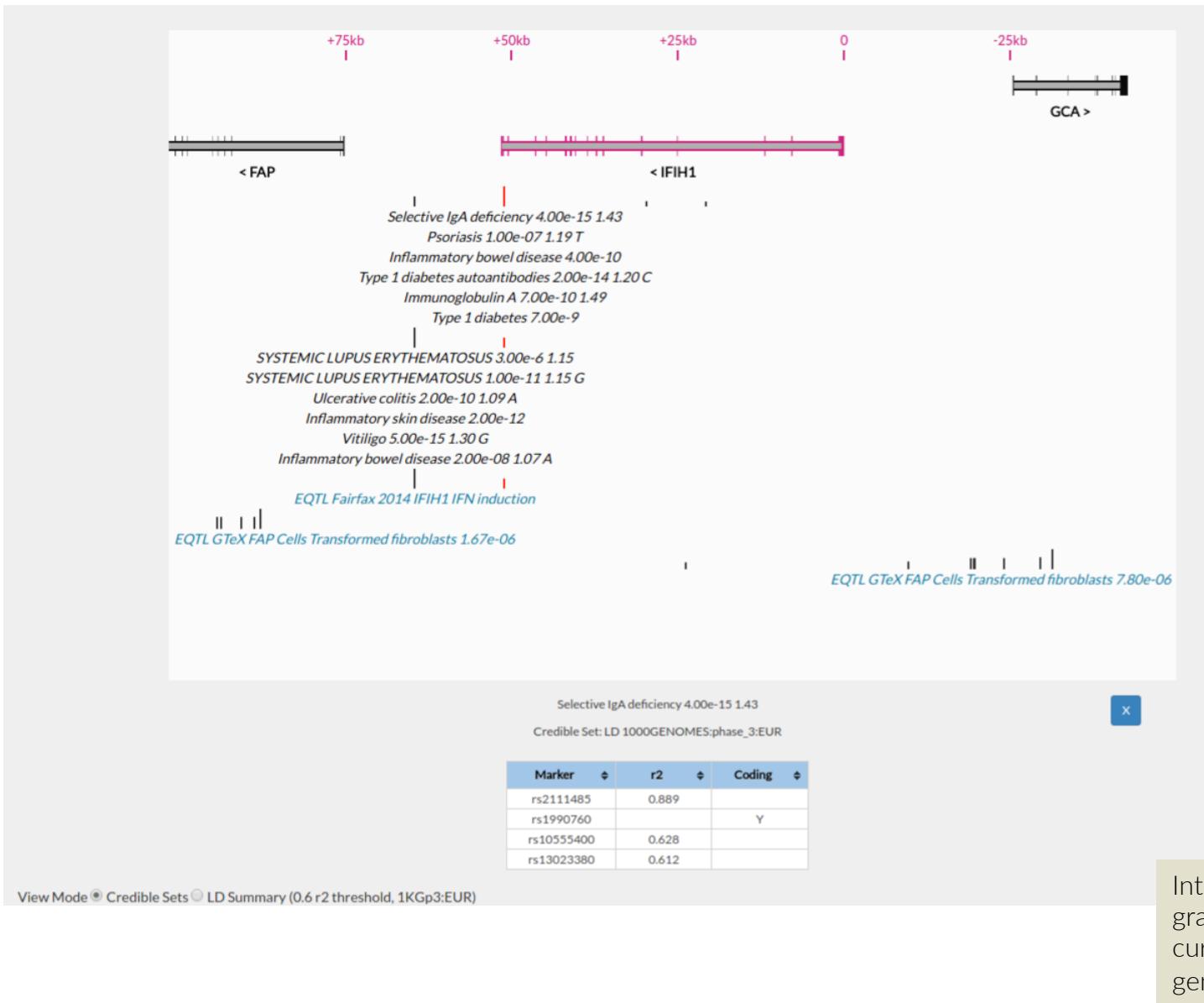
At the bottom, there are tabs for Functional Variants, Expression Data, Protein and Structure Chemistry, Clinical Data, and References. The References section is expanded, showing a table of publications with columns for 1st Author, Year, Journal/Title/File, Title/Description, and Show/Add Details.

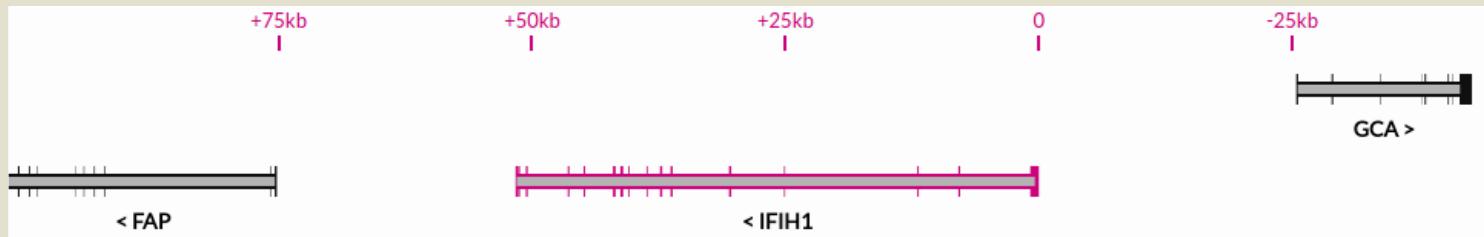
Interactive graphical view of curated data in a genomic context.

Text-based/tabular view of curated data.

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

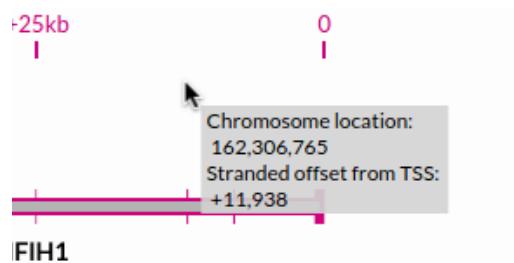
Overview





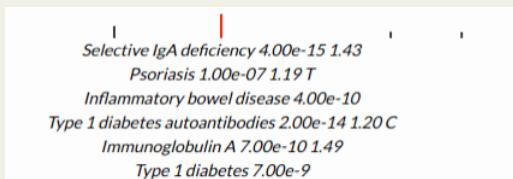
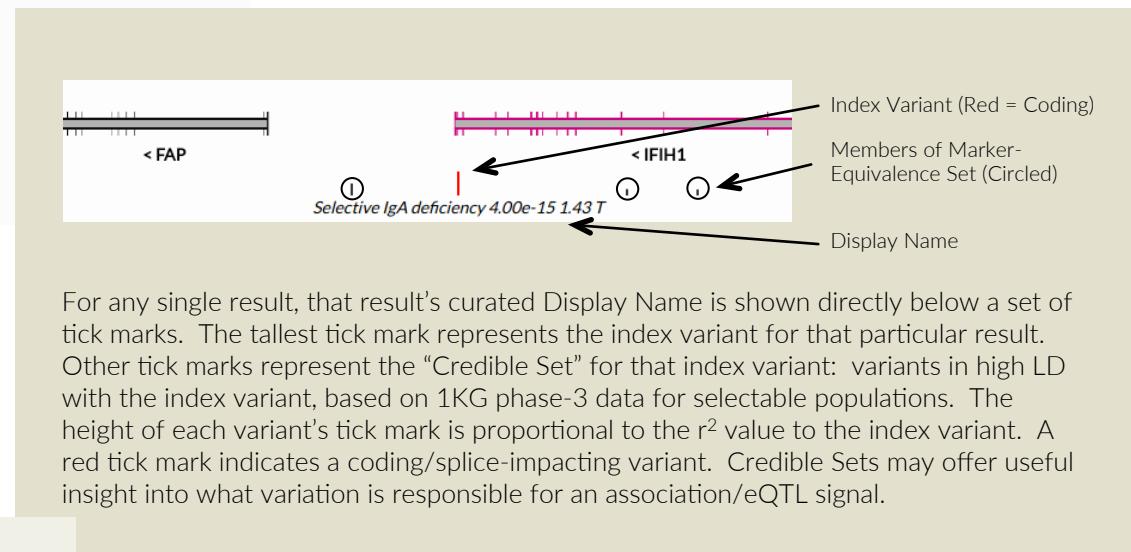
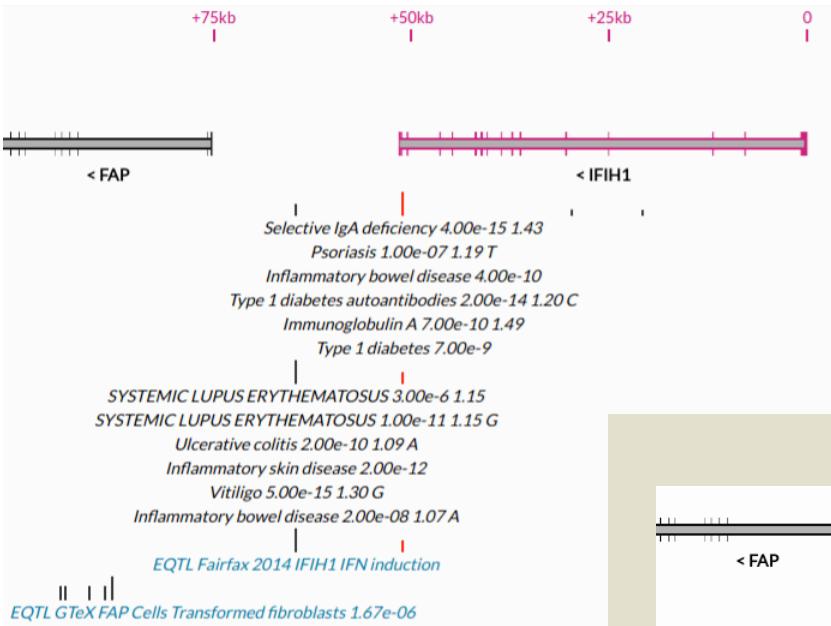
The display window shows a 50-kb neighborhood around the Ensembl canonical transcript for the target gene.

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



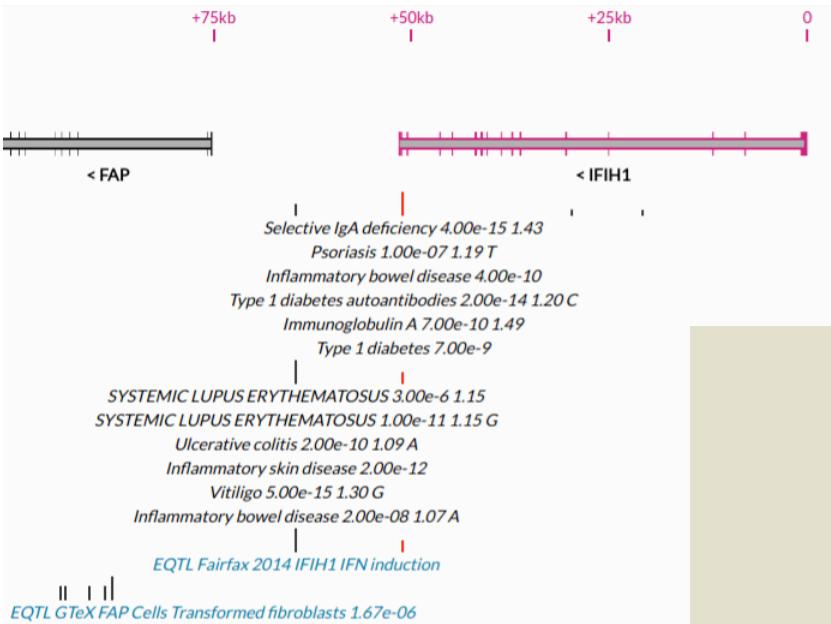
Clicking on the background of the display window will report the click's genomic position and stranded distance from the transcript start site of the target gene.

Interactive graphical view of curated data in a genomic context.

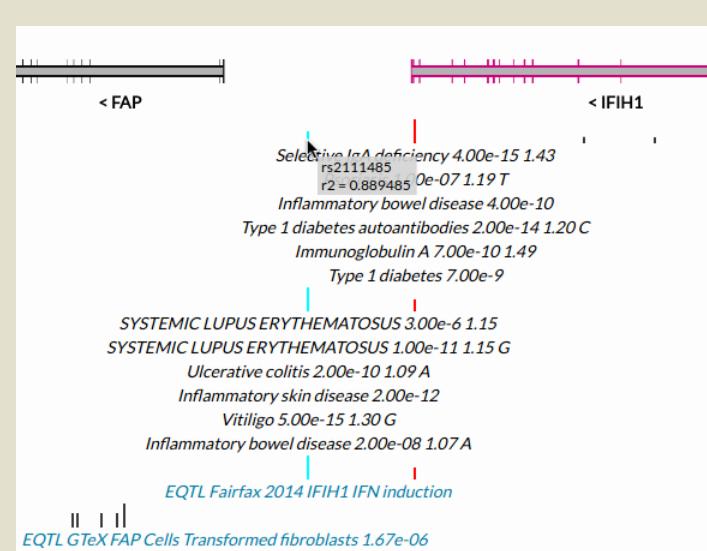


All results with the same index variant and Credible Set have their Display Names grouped together.

Interactive graphical view of curated data in a genomic context.



Curated association and eQTL results are shown below the gene annotations.



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

Selective IgA deficiency 4.00e-15 1.43
Credible Set: LD 1000GENOMES:phase_3:EUR

Marker	r2	Coding
rs2111485	0.889	
rs1990760		Y
rs10555400	0.628	
rs13023380	0.612	

Clicking on the Display Name for a particular association/eQTL result will reveal a dismissible table just below the display window summarizing the index variant and its Credible Set.

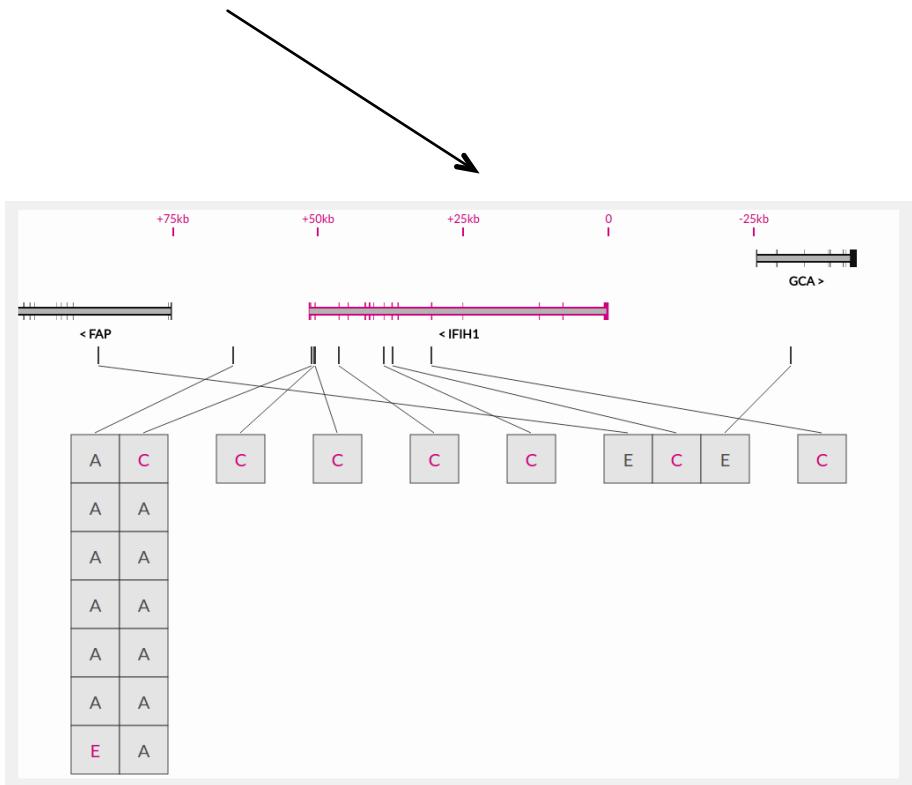
Interactive graphical view of curated data in a genomic context.

Two different graphical perspectives are available

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



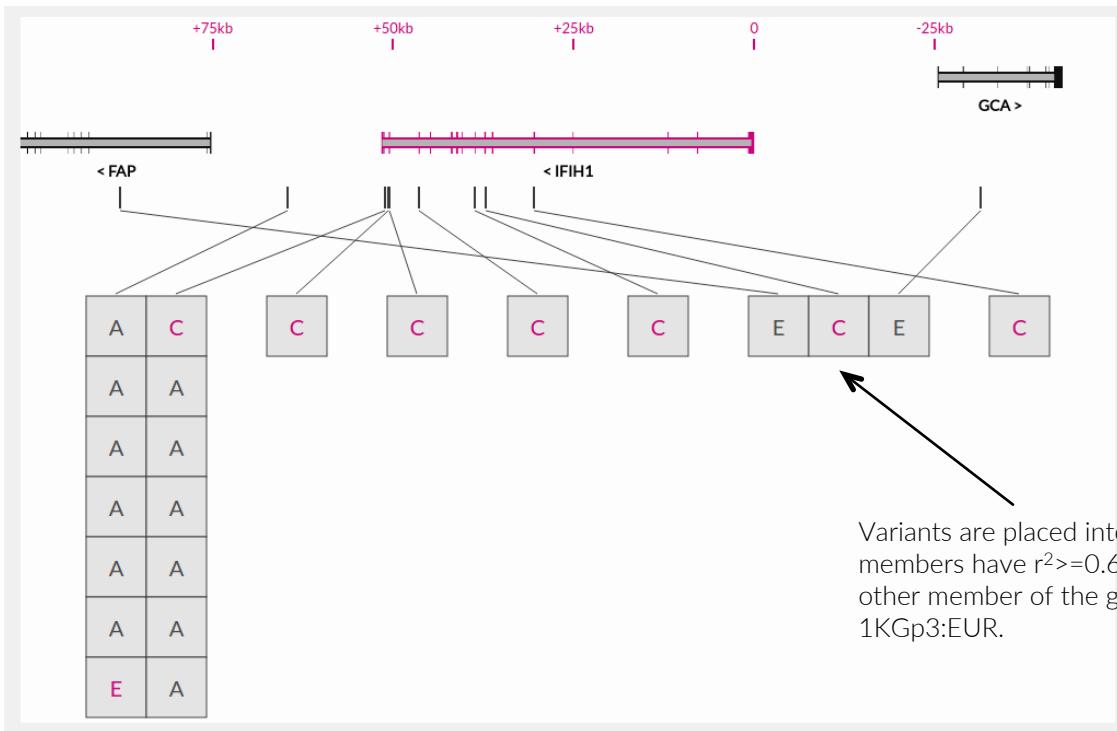
Credible Sets are shown for curated association and eQTL events.



Association/eQTL index variants and other curated variants are clustered into high-LD groups

Interactive graphical view of curated data in a genomic context.

LD-summary graphical perspective



For each variant, boxes indicating particular association results (A), eQTL results (E), or coding impacts (C) are stacked vertically.

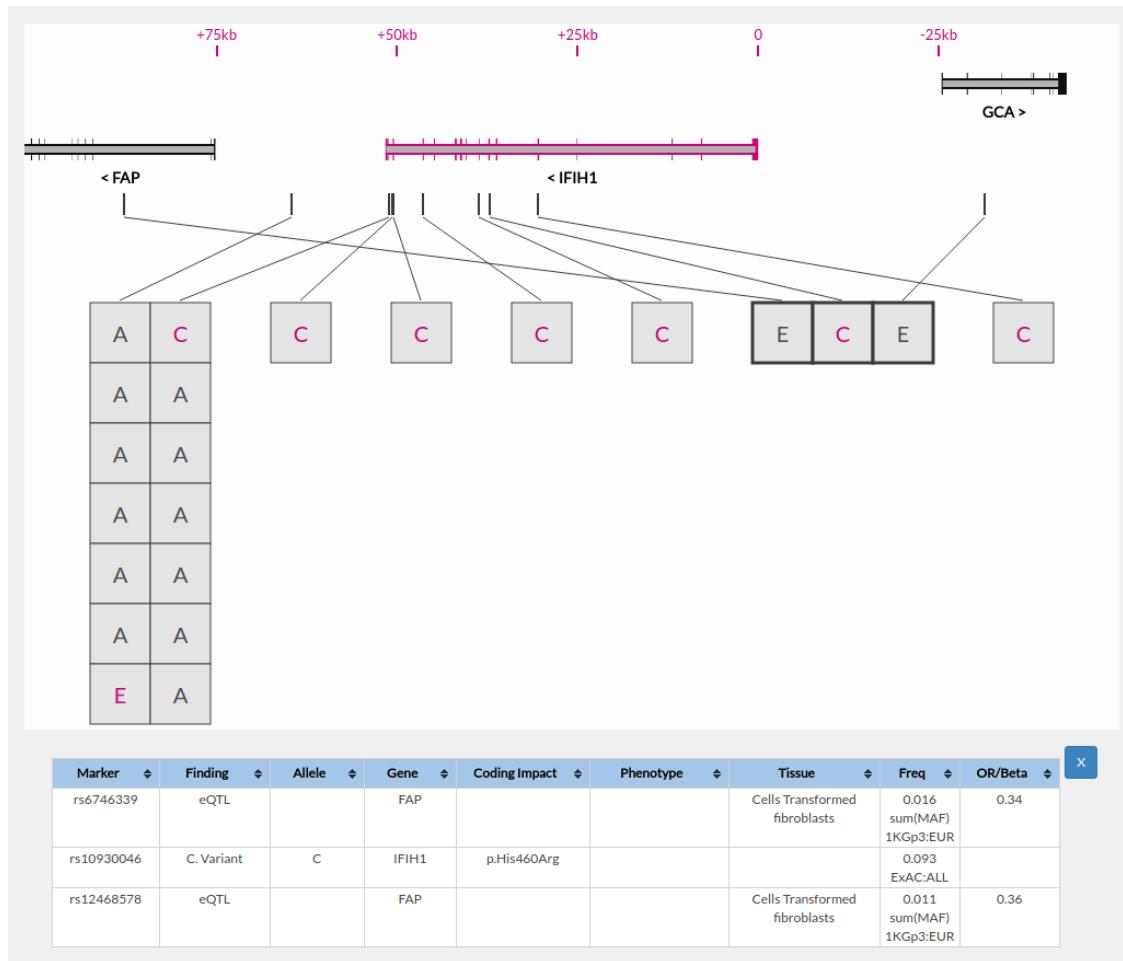
Where the eQTL or coding impact has been assigned to the target gene, that box's label is shaded magenta.

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



Interactive graphic view of curated data in a genomic context.

LD-summary graphical perspective



Clicking on each LD group will reveal a dismissible table below the display window which summarizes the events in that group.

Interactive graphic view of curated data in a genomic context.

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

Overview

Summary (IFIH1 TSS Ensembl 89;chr2:162,318,703)

Association Results

eQTL Results

▼ Variants of Interest

LD Summary	Name	Non-Reference Allele	Location	Gene	Protein Change	Polyphen	f(NFE)	f(Max)
	rs147278787	A	162,318,079	IFIH1	p.Arg77Trp	possibly damaging	0.001	0.003
	rs146721166	C	162,317,872	IFIH1	p.Ile146Val	benign	0.000	0.004
	rs74162075	C	162,310,909	IFIH1	p.Asn160Asp	probably damaging	0.001	0.001
✓	rs72650664	C	162,288,184	IFIH1	p.Lys349Arg	benign	0.001	0.020
	rs150317197	T	162,288,164	IFIH1	p.Pro356Thr	benign	0.001	0.001
	rs117608083	C	162,288,137	IFIH1	p.Lys365Glu	probably damaging	0.000	0.008
	rs140977021	T	162,282,575	IFIH1	p.Val366Glu	probably damaging	0.000	0.004
	rs145520044	T	162,282,551	IFIH1	p.Arg374His	benign	0.001	0.001
✓	rs10930046	C	162,281,473	IFIH1	p.His460Arg	benign	0.015	0.426
	rs147000317	A	162,281,371	IFIH1	p.Gly494Val	probably damaging	0.000	0.008

Page: 1 2 3 4 5 6 7 8 9 10 11 12 13 14 15 16 17 18 19 20 21 22 23 24 25 26 27 rows

Functional Variants

Expression Data

Protein and Structure Chemistry

Clinical Data

▼ References

1st Author	Year	Journal/Site/File	Title/Description	Show/Add Details
Fairfax BP	2014	Science	Innate immune activity conditions the effect of regulatory variants upon monocyte gene expression.	Show/Add Details
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.	Show/Add Details (1)
Pettersson M	2017	Am. J. Med. Genet. A	Further evidence for specific IFIH1 mutation as a cause of Singleton-Merten syndrome with phenotypic heterogeneity.	Show/Add Details
Wu B	2013	Cell	Structural basis for dsRNA recognition, filament formation, and antiviral signal activation by MDA5.	Show/Add Details (3)
Barrett JC	2009	Nat. Genet.	Genome-wide association study and meta-analysis find that over 40 loci affect risk of type 1 diabetes.	Show/Add Details
Baurecht H	2015	Am. J. Hum. Genet.	Genome-wide comparative analysis of atopic dermatitis and psoriasis gives insight into opposing genetic mechanisms.	Show/Add Details
Bentham J	2015	Nat. Genet.	Genetic association analyses implicate aberrant regulation of innate and adaptive immunity genes in the pathogenesis of systemic lupus erythematosus.	Show/Add Details
Bronson PG	2016	Nat. Genet.	Common variants at PVT1, ATG13-AMBR1, AH1 and CLEC16A are associated with selective IgA deficiency.	Show/Add Details
Ferreira RC	2010	Nat. Genet.	Association of IFIH1 and other autoimmunity risk alleles with selective IgA deficiency.	Show/Add Details
GTEx Consortium	2015	Science	Human genomics. The Genotype-Tissue Expression (GTEx) pilot analysis: multitissue gene regulation in humans.	Show/Add Details (1)

Page: 1 2 3 4 5 6 7 8 9 10 11 12 13 14 15 16 17 18 19 20 21 22 23 24 25 26 27 rows

Add Pubmed Reference Add File Reference Add Web Reference Add bioRxiv Reference

Text-based/
tabular view of
curated data.

There are nine sections containing different curated data

- ▶ Summary (IFIH1 TSS Ensembl 89:chr2:162,318,703)
- ▶ Association Results
- ▶ eQTL Results
- ▶ Variants of Interest
- ▶ Functional Variants
- ▶ Expression Data
- ▶ Protein and Structure Chemistry
- ▶ Clinical Data
- ▶ References

Text-based/
tabular view of
curated data.

Summary section

The Ensembl version used for automated data curation is shown here, along with the start position of the canonical transcript for the target gene.

A screenshot of a gene summary interface. At the top, a blue header bar displays the text "▼ Summary (IFIH1 TSS Ensembl 89:chr2:162,318,703)". Below this is a white input field with the placeholder "Enter summary". A black arrow points from the explanatory text above to the "Enter summary" field.

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	
Immunoglobulin A 7.00e-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: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:EUR (1)	<input checked="" type="checkbox"/>	Enter comment	
Inflammatory bowel	Inflammatory bowel	Liu JZ; Association analyses	2015	rs1990760		4.00e-10		LD 1KGp3:EUR (3)	<input checked="" type="checkbox"/>	Enter comment	
Editable Display Name by which to label the association result in the display window.				Note that reported alleles are not validated against the genome assembly. Alleles followed by an asterisk are those which do not match any forward-strand allele for the index variant.				Dropdown menu to select Credible Set. 'None' and 'Unset' are also options.			
SYSTEMIC LUPUS ERYTHEMATOSUS 1.00e-11 1.15 G	SYSTEMIC LUPUS ERYTHEMATOSUS	and CLEC16A are associated with selective IgA deficiency.									
		Bentham J; Genetic association analyses implicate aberrant regulation of innate and adaptive immunity genes in the pathogenesis of systemic lupus erythematosus.	2015	rs2111485	G	1.00e-11	1.15	LD 1KGp3:EUR (1)	<input checked="" type="checkbox"/>	Enter comment	
Page: 1 ▼ (H) (H) 1 to 10 of 17 rows (H) (H) 10 ▼				Add Association				Text-based/ tabular view of curated data.			

Button for manual addition of association results.

Text-based/ tabular view of curated data.

Association Results section

Adding an association result

Page: 1 10 1 to 10 of 18 rows 10 Click button

Add Association

↓

Association Results											
Display Name	Phenotype	Source	Year	Index Variant	Allele	Pvalue	OR/Beta	Credible Set	Show	Curator Comment	
	Eg. Schizophrenia	Yin X; Genome-wide meta-analysis identifies multiple novel associations and ethnic heterogeneity of psoriasis susceptibility.		Eg. rs123	Eg. T	Eg. 1.00e-8	Eg. 1.17			Enter comment	X S

Report phenotype

Select appropriate reference from those known to the target-gene database

Report index variant

Report allele

Report Pvalue

Report O.R./Beta

Provide curator comment, if desired

X button will abandon the manual entry of an association result

S button will save the result to the target-gene database

↓

Association Results											
Display Name	Phenotype	Source	Year	Index Variant	Allele	Pvalue	OR/Beta	Credible Set	Show	Curator Comment	
Psoriasis 1.0E-7 1.19 T	Psoriasis	Yin X; Genome-wide meta-analysis identifies multiple novel associations and ethnic heterogeneity of psoriasis susceptibility.	2015	rs1990760	T	1.0E-7	1.19	Unset		Enter comment	X

After the association has been saved to the target-gene database, Display Name, Credible Set, Display Toggle, and Curator Comment can be further modified, with changes automatically preserved in the target-gene database.

X button can be used to remove an association result from the target-gene database

Text-based/ tabular view of curated data.

eQTL Results section

eQTL Results													
Display Name	Tissue	Gene	Source	Year	Index Variant	Pvalue	Beta	Credible Set	Show	Curator Comment			
EQTL Fairfax 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	<input type="button" value="X"/>		
EQTL GTEx FAP Artery Tibial 5.19e-07	Artery Tibial	FAP	GTEx Consortium	2015	rs3827491	5.19e-07	0.43	LD 1KGp3:EUR (31)	<input type="checkbox"/>	Tissue pval threshold = 3.54e-05			
EQTL GTEx FAP Cells Transformed fibroblasts	Cells Transformed fibroblasts	FAP	GTEx Consortium	2015	rs6746339	1.67e-06	0.34	LD 1KGp3:EUR (4)	<input checked="" type="checkbox"/>	Tissue pval threshold = 3.54e-05			
EQTL GTEx FAP Cells Transformed fibroblasts	Cells Transformed fibroblasts	FAP	GTEx Consortium	2015	rs12468578	7.80e-06	0.36	LD 1KGp3:EUR (7)	<input checked="" type="checkbox"/>	Tissue pval threshold = 3.54e-05			
EQTL GTEx FAP Muscle Skeletal 1.58e-05	Muscle Skeletal	FAP	GTEx Consortium	2015	rs35544136	1.58e-05	0.67	LD 1KGp3:EUR (1)	<input type="checkbox"/>	Tissue pval threshold = 1.85e-05			
EQTL GTEx FAP Nerve Tibial 2.55e-05	Nerve Tibial	FAP	GTEx Consortium	2015	rs3788967	2.55e-05	0.56	LD 1KGp3:EUR (1)	<input type="checkbox"/>	Tissue pval threshold = 4.11e-05			
EQTL GTEx FAP Thyroid 2.67e-05	Thyroid	FAP	GTEx Consortium	2015	rs2389683	2.67e-05	0.45	LD 1KGp3:EUR (0)	<input type="checkbox"/>	Tissue pval threshold = 3.29e-05			
EQTL GTEx FAP Thyroid 2.91e-05	Thyroid	FAP	GTEx Consortium	2015	rs11455810	2.91e-05	0.44	LD 1KGp3:EUR (0)	<input type="checkbox"/>	Tissue pval threshold = 3.29e-05			
EQTL GTEx FAP Thyroid 4.53e-06	Thyroid	FAP	GTEx Consortium	2015	rs16846600	4.53e-06	0.51	LD 1KGp3:EUR (56)	<input type="checkbox"/>	Tissue pval threshold = 3.29e-05			
EQTL GTEx GCA Adrenal Gland 6.34e-06	Adrenal Gland	GCA	GTEx Consortium	2015	rs34977319	6.34e-06	-0.77	LD 1KGp3:EUR (0)	<input type="checkbox"/>	Tissue pval threshold = 1.75e-05			

Page: [1](#) [2](#) [3](#) [4](#) 1 to 10 of 10 rows [11](#) [12](#) [13](#) [14](#) [15](#) [16](#) [17](#) [18](#) [19](#) [20](#)

Add eQTL

This section is very similar to the Association Results section. One important difference is that Credible Sets are built using only variants showing significant eQTL results relative to tissue-specific thresholds.

Text-based/
tabular view of
curated data.

Variants of Interest section

▼ Variants of Interest

LD Summary	Name	Non-Reference Allele	Location	Gene	Protein Change	Polyphen	f(NFE)	f(Max)
<input type="checkbox"/>	rs147278787	A	162,318,079	IFIH1	p.Arg77Trp	possibly damaging	0.001	0.003
<input type="checkbox"/>	rs146721166	C	162,317,872	IFIH1	p.Ile146Val	benign	0.000	0.004
<input type="checkbox"/>	rs74162075	C	162,310,909	IFIH1	p.Asn160Asp	probably damaging	0.001	0.001
<input checked="" type="checkbox"/>	rs72650664	C	162,288,184	IFIH1	p.Lys349Arg	benign	0.001	0.020
<input type="checkbox"/>	rs150317197	T	162,288,164	IFIH1	p.Pro356Thr	benign	0.001	0.001
<input type="checkbox"/>	rs117608083	C	162,288,137	IFIH1	p.Lys365Glu	probably damaging	0.000	0.008
<input type="checkbox"/>	rs140977021	T	162,282,575	IFIH1	p.Val366Glu	probably damaging	0.000	0.004
<input type="checkbox"/>	rs145520044	T	162,282,551	IFIH1	p.Arg374His	benign	0.001	0.001
<input checked="" type="checkbox"/>	rs10930046	C	162,281,473	IFIH1	p.His460Arg	benign	0.015	0.426
<input type="checkbox"/>	rs147000317	A	162,281,371	IFIH1	p.Gly494Val	probably damaging	0.000	0.008

Page: 1 ▼ 1 to 10 of 27 rows 10 ▼



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

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.

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.

Functional Variants, Expression Data, Protein and Structure Chemistry, and Clinical Data sections

Functional Variants

Expression Data

1st Author	Year	Journal/Site/File	Title/Description
GTEX Consortium	2015	Science	Human genomics. The Genotype-Tissue Expression (GTEX) pilot analysis: multitissue gene regulation in humans.

Highest expression in EBV-transformed lymphocytes, spleen and lung

Page: 1 10

Editable description that is stored with the reference Detail

Protein and Structure Chemistry

Clinical Data



Each of these sections contains those reference Details which have been assigned to these particular sections.



Clicking on a Detail's thumbnail will overlay the full-sized version

Text-based/ tabular view of curated data.

Reference section

References

1st Author	Year	Journal/Site/File	Title/Description	Show/Add Details	X
Fairfax BP	2014	Science	Innate immune activity conditions the effect of regulatory variants upon monocyte gene expression.	Show/Add Details	X
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.	Show/Add Details (1)	X
<div style="display: flex; align-items: center;"> <div style="flex: 1;"> <p>Section Assignment</p> <div style="border: 1px solid #ccc; padding: 2px; width: fit-content;">Functional</div> </div> <div style="flex: 1; margin-left: 20px;"> <p>Functional data linking rs1990760 (A946T) and protection from infection challenge in mice.</p> </div> <div style="flex: 1; margin-left: 20px;"> <p>Figure 4: <i>Ifih1^{fl/fl}</i> mice exhibit protection from EMCV challenge.</p> <p>Survival (%)</p> <p>Time after EMCV infection (d)</p> </div> <div style="flex: 1; margin-left: 20px;"> <p>Button to remove Detail elements</p> </div> </div>					
<div style="display: flex; align-items: center;"> <div style="flex: 1;"> <p>Dropdown menu to assign Detail to a particular Target Gene Notebook section</p> </div> <div style="flex: 1; margin-left: 20px;"> <p>Button to add a new Detail to this reference</p> </div> <div style="flex: 1; margin-left: 20px;"> <p>Add Detail</p> </div> <div style="flex: 1; margin-left: 20px;"> <p>Button to remove manually-added references from the target-gene database</p> </div> </div>					
Pettersson M	2017	Am. J. Med. Genet. A	Further evidence for specific IFIH1 mutation as a cause of Singleton-Merten syndrome with phenotypic heterogeneity.	Show/Add Details	X
Wu B	2013	Cell	Structural basis for dsRNA recognition, filament formation, and antiviral signal activation by MDA5.	Show/Add Details (3)	X
Barrett JC	2009	Nat. Genet.	Genome-wide association study and meta-analysis find that over 40 loci affect risk of type 1 diabetes.	Show/Add Details	
Baurecht H	2015	Am. J. Hum. Genet.	Genome-wide comparative analysis of atopic dermatitis and psoriasis gives insight into opposing genetic mechanisms.	Show/Add Details	
Bentham J	2015	Nat. Genet.	Genetic association analyses implicate aberrant regulation of innate and adaptive immunity genes in the pathogenesis of systemic lupus erythematosus.	Show/Add Details	
Bronson PG	2016	Nat. Genet.	Common variants at PVT1, ATG13-AMBRA1, AH11 and CLEC16A are associated with selective IgA deficiency.	Show/Add Details	
Ferreira RC	2010	Nat. Genet.	Association of IFIH1 and other autoimmunity risk alleles with selective IgA deficiency.	Show/Add Details	
GTEx Consortium	2015	Science	Human genomics. The Genotype-Tissue Expression (GTEx) pilot analysis: multitissue gene regulation in humans.	Show/Add Details (1)	

Page: 1 ▾ (H) (H) 1 to 10 of 17 rows (H) (H) 10 ▾

Buttons to add new references of different types

Toggles to show/hide Detail elements assigned to each reference

Text-based/ tabular view of curated data.

Reference section

Adding new references is similar to adding new Association results

Add new Pubmed reference

1st Author	Year	Journal/Site/File	Title/Description	X	S
			Pubmed ID: Eg. 23817569	X	S

Add new file-based reference

1st Author	Year	Journal/Site/File	Title/Description	X	S
	Eg. 2017	Choose File	No file chosen	X	S
			Eg. Some description	X	S

Add new web reference

1st Author	Year	Journal/Site/File	Title/Description	X	S
	Eg. 2017	https://www.google.com	Eg. Some description	X	S

Add new bioRxiv reference

1st Author	Year	Journal/Site/File	Title/Description	X	S
			doi: Eg. 10.1101/045831	X	S

Button to abandon
new reference
addition

Button to complete
new reference
addition

Text-based/
tabular view of
curated data.

Reference section

Adding a new Detail is also similar to adding new Association results

1st Author	Year	Journal/Site/File	Title/Description	
Fairfax BP	2014	Science	Innate immune activity conditions the effect of regulatory variants upon monocyte gene expression.	<button>Hide Details</button>

Eg. Some description

Editable text
description of the
Detail

Detail graphic can
be dragged here or
pasted from the
system clipboard

Button to abandon
new Detail
addition

Button to complete
new Detail addition

Text-based/
tabular view of
curated data.