

Analyzing germline mutation profile pathways by using QIAGEN Ingenuity Pathway Analysis



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- Introduction to QIAGEN Ingenuity Pathway Analysis
- Search in IPA
- Analyzing germline mutation using IPA
 - Data format
 - Data upload and analysis setup
 - Core analysis
- Querying IPA's Knowledge base
 - Custom network construction
 - Contextualization using public data
- Summary

Account: [XX@gate.sinica.edu.tw](#)
(xx is your email account)
Password: **your password**

Download website: <https://tinyurl.com/GGAIPA>

Dataset: <https://tinyurl.com/IPAslideshare>

Client: (IPA Login)

Training account activation code and link by e-mail, please notices your mailbox

Dear

Reminder: You have 33 days remaining to activate your QIAGEN IPA trial.

Your 14-day trial license activation code is: xUJSRVV2bf2VNIIIV.

Once activated, your QIAGEN IPA trial will be valid for 14 days. Please be aware that this is an evaluation license and there are limitations on the number of uploads, exports and analyses that can be performed during the trial period.

To activate your trial, click [here](#) or copy paste this text to a browser
https://apps.ingenuity.com/isa/account/protected/signup/ipa?coupon=tpd3U2Yyn1NPSXX4Zt1ZIEhWDjAgqiZK63TVMtzy_Mw-FHZhISQKdPVIgaaJjuTR. The trial can be activated to begin anytime within the next 33 days, and after that the code will expire.

We recommend that after you create your account and receive your activation email, you use this [installer](#) or copy paste this text to a browser <https://analysis.ingenuity.com/pa/installer/select> to get started with your QIAGEN IPA trial.

We suggest viewing a few [tutorials](#) to get you started. You may also want to check out our [training videos](#).

If you have any questions, please contact Customer Support at ts-bioinformatics@qiagen.com or by phone at US: +1.650.381.5111 US Toll Free: +1.866.464.3684 or Denmark Toll Free: +45 80 82 0167.

Best regards,
The QIAGEN Digital Insights Team

Create your IPA® Account



Sign-up with Trial Code **Join Institution License**

All fields are required unless otherwise indicated.

Login Information

Email *

Trial sign-up code *
If you do not have a trial sign-up code, fill out this form [here](#).

Password *
Password strength

- The password must be at least 12 characters in length and contain at least 1 uppercase letter, 1 lowercase letter, 1 number, 1 special character and no dollar sign(\$) or empty space.
- Do not use dictionary words, your name, e-mail address, or other personal information that can be easily obtained.
- Do not use the same password for multiple online accounts.

Verify password *

Name and Contact Information

First name *

Last name *

Institution or Company *
Please select an option from autocomplete dropdown

CREATE ACCOUNT



[Home](#) > [Product Log in](#)



Ingenuity Pathway Analysis

[DOWNLOAD CLIENT INSTALLER](#)

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Ingenuity Pathway Analysis for China

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QCI Interpret

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Ingenuity
Pathway
Analysis



Install IPA on your computer

This installer will enable you to access IPA like other desktop applications on your computer (though still requiring an internet connection).

Note: This is all you need to run IPA. It is not necessary to install Java separately from IPA.

Click on the button below to download the installer

IPA for Windows (64-bit)

(Installer recommended for your computer)

Other options:

[IPA for 32-bit Windows](#)

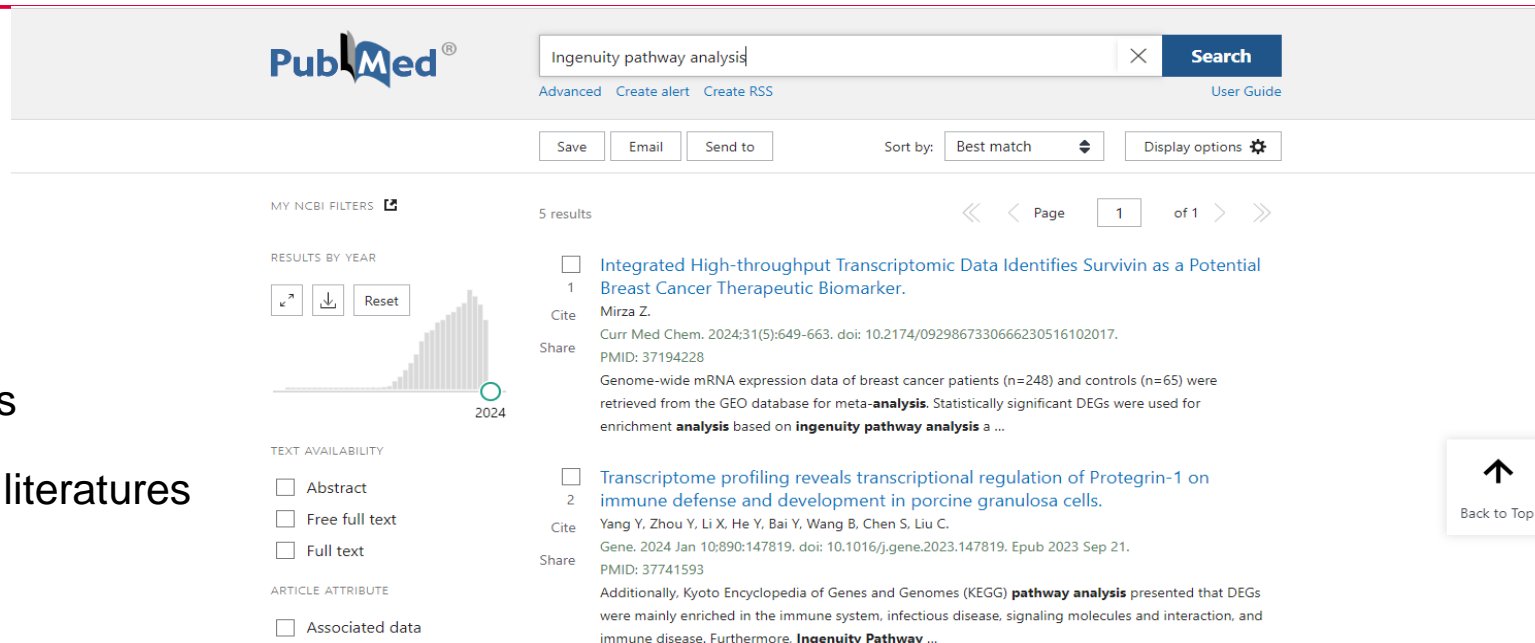
[IPA for macOS](#)

For more information see this [help page](#).

Note: If you have trouble installing or logging in with the IPA client, please click [here](#) for an alternate method to launch IPA.

Looking for more information about IPA? Click [here](#).

4,153 results
In 2023, 303 literatures



Transcriptomics

Genomics

proteomics

metabolomics

► [Comput Mol Biosci.](#) 2023 Jun;13(2):21-34. doi: 10.4236/cmb.2023.132002. Epub 2023 May 19.

Analysis of Differential Gene Expression and Core Canonical Pathways Involved in the Epithelial to Mesenchymal Transition of Triple Negative Breast Cancer Cells by Ingenuity Pathway Analysis

Elizabeth Cagle¹, Brent Lake¹, Anasua Banerjee¹, Jazmine Cuffee¹, Narendra Banerjee¹, Darla Gilmartin¹, Makaiyah Liverman¹, Shennel Brown¹, Erik Armstrong¹, Santanu Bhattacharya^{2,3}, Somiranjana Ghosh⁴, Tanmoy Mandal⁴, Hirendra Banerjee¹

Affiliations + expand
PMID: 37538932 PMCID: PMC10398793 DOI: 10.4236/cmb.2023.132002

► [J Neurosurg.](#) 2018 Mar;128(3):911-922. doi: 10.3171/2016.10.JNS161384. Epub 2017 Apr 14.

Genetic landscape of sporadic vestibular schwannoma

Arl Løge Håvik^{1,2,3}, Ove Bruland², Erling Myrseth⁴, Hrvoje Miletic^{5,6,7}, Mads Aarhus⁸, Per-Morten Knappskog^{2,3}, Morten Lund-Johansen^{1,4,6}

Affiliations + expand
PMID: 28409725 DOI: 10.3171/2016.10.JNS161384

Abstract

OBJECTIVE Vestibular schwannoma (VS) is a benign tumor with associated morbidities and reduced quality of life. Except for mutations in NF2, the genetic landscape of VS remains to be elucidated.

► [Hepatol Int.](#) 2023 Jun;17(3):698-708. doi: 10.1007/s12072-022-10473-x. Epub 2023 Jan 18.

Biomarkers of hepatocellular synthesis in patients with decompensated cirrhosis

Beirvan Gurbuz^{#1}, Nurdan Guldiken¹, Philipp Reuken², Lei Fu^{1,3}, Katharina Remih¹, Christian Preisinger⁴, Radan Brúha⁵, Martin Leniček⁶, Jaromír Petrtýl⁵, Johanna Reissing¹, Mahmoud Aly^{1,7}, Malin Fromme¹, Biaohuan Zhou^{1,8}, Isabel Karkossa⁹, Kristin Schubert⁹, Martin von Bergen^{9,10,11}, Andreas Stallmach², Tony Bruns^{#1,2}, Pavel Strnad^{#1,2}

Affiliations + expand
PMID: 36652164 PMCID: PMC10224844 DOI: 10.1007/s12072-022-10473-x
[Free PMC article](#)

Abstract

Background and aim: Since hepatocytes produce majority of serum proteins, patients with cirrhosis display substantial alterations in the serum proteome. The aim of the current study was to

► [Proteomics.](#) 2021 Jun;21(11-12):e2100037. doi: 10.1002/pmic.202100037. Epub 2021 May 31.

Biomarker identification and pathway analysis of rheumatoid arthritis based on metabolomics in combination with ingenuity pathway analysis

Zhuoru He¹, Zhongqiu Liu¹, Lingzhi Gong¹

Affiliations + expand
PMID: 33969925 DOI: 10.1002/pmic.202100037

Abstract

Rheumatoid arthritis (RA) is a common autoimmune and inflammatory disease worldwide, but understanding its pathogenesis is still limited. In this study, plasma untargeted metabolomics of a discovery cohort and targeted analysis of a verification cohort were performed by gas chromatograph



Ingenuity Literature Findings

Ingenuity Expert Findings – manually curated Findings from the full-text with contextual details from top journals

Ingenuity ExpertAssist Findings – automated text Findings that are manually reviewed from abstracts covering a broader range of publications – comprise a small percentage of IPA's findings



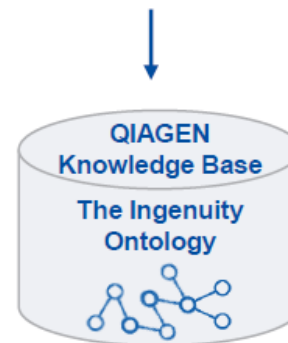
Ingenuity Modeled Knowledge

Ingenuity Expert Knowledge – content we model such as canonical pathways, toxicity lists, etc.

Ingenuity-Supported Third-Party Information – content areas include protein-protein, miRNA, biomarker, clinical trial information and others

Databases such as COSMIC, Clinical Trials, OMIM, TargetScan, BioGRID, MGD and HumanCyc

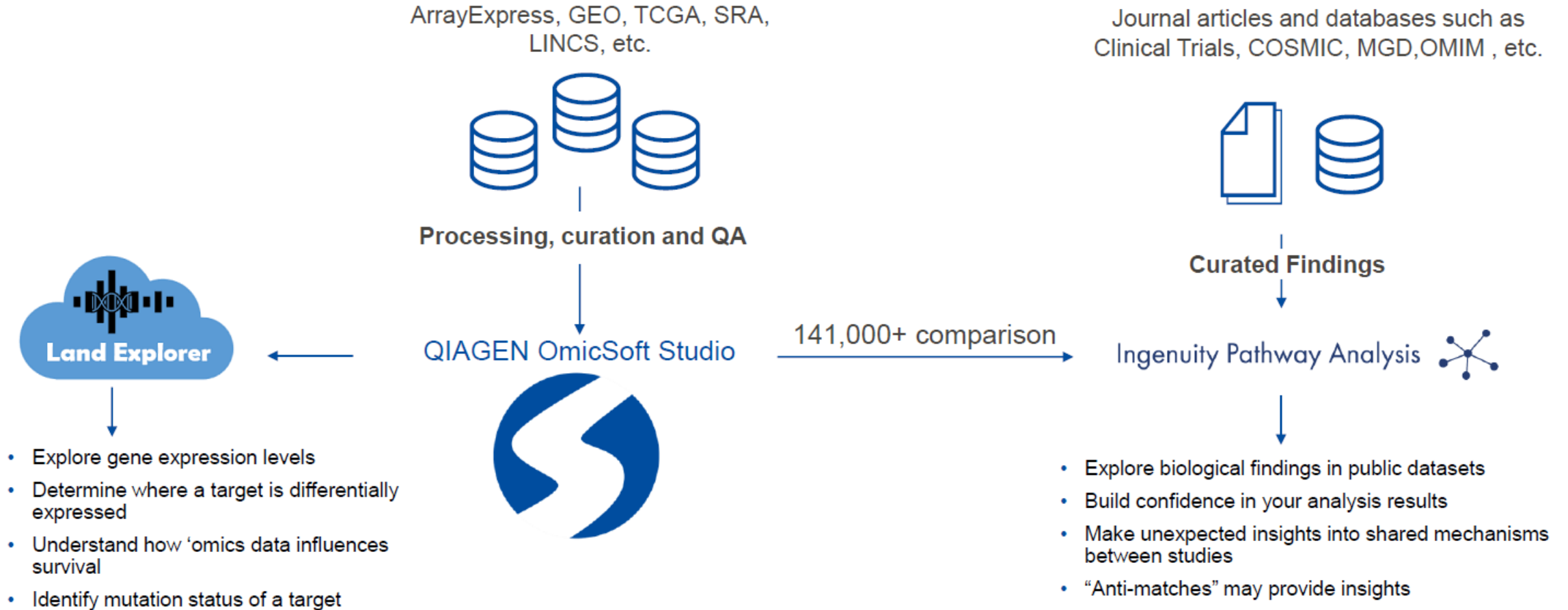
A massive, manually curated Knowledge Base updated weekly for the last ~20 years

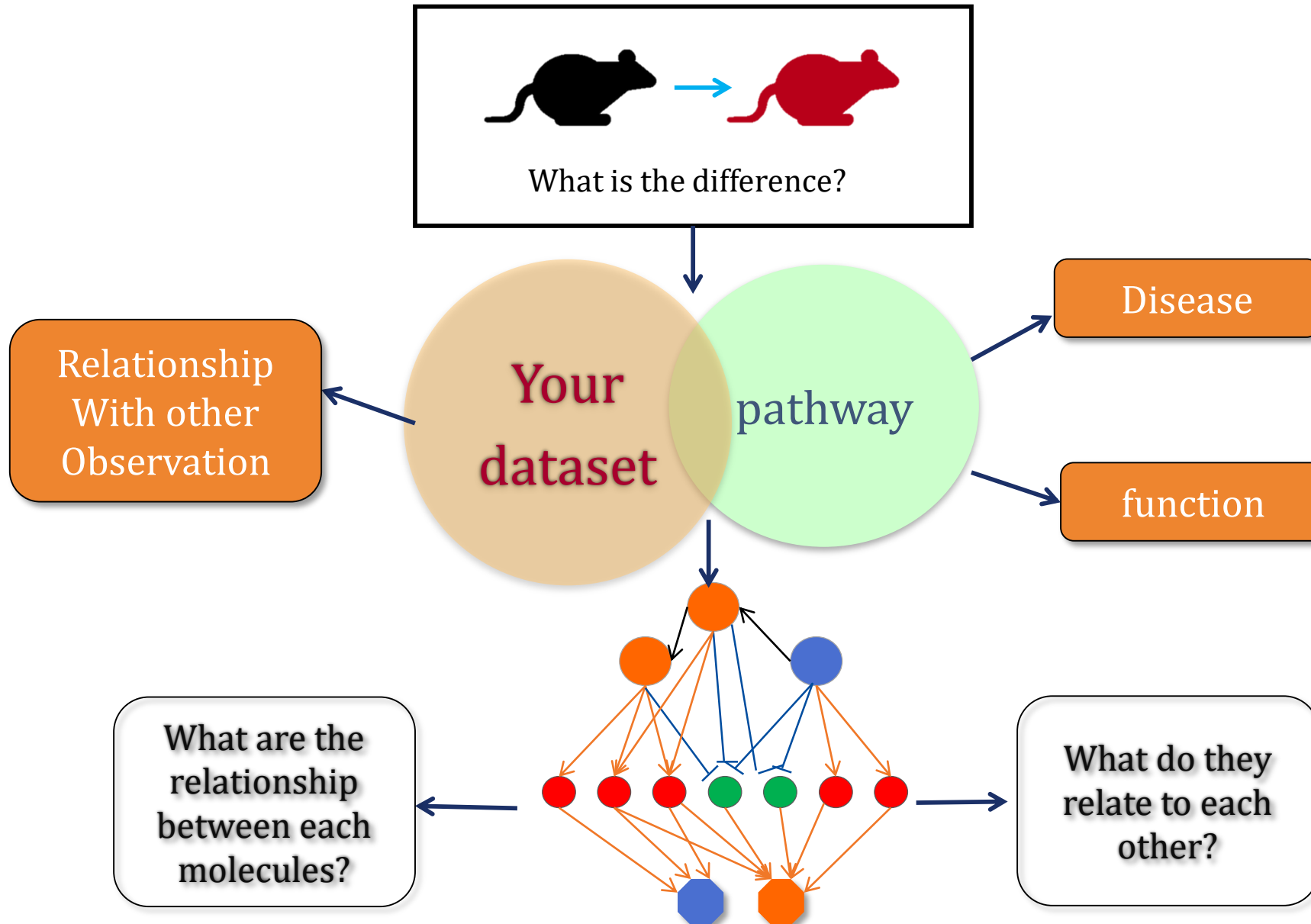


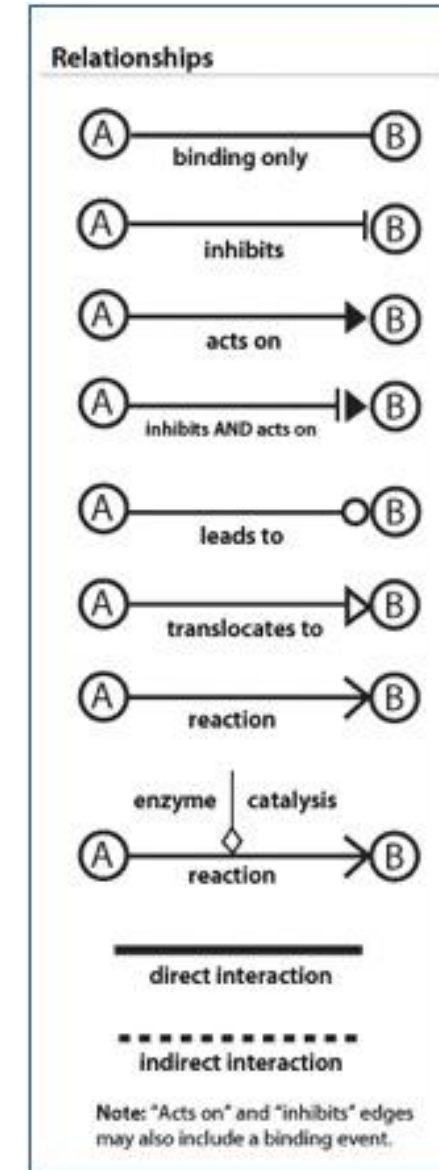
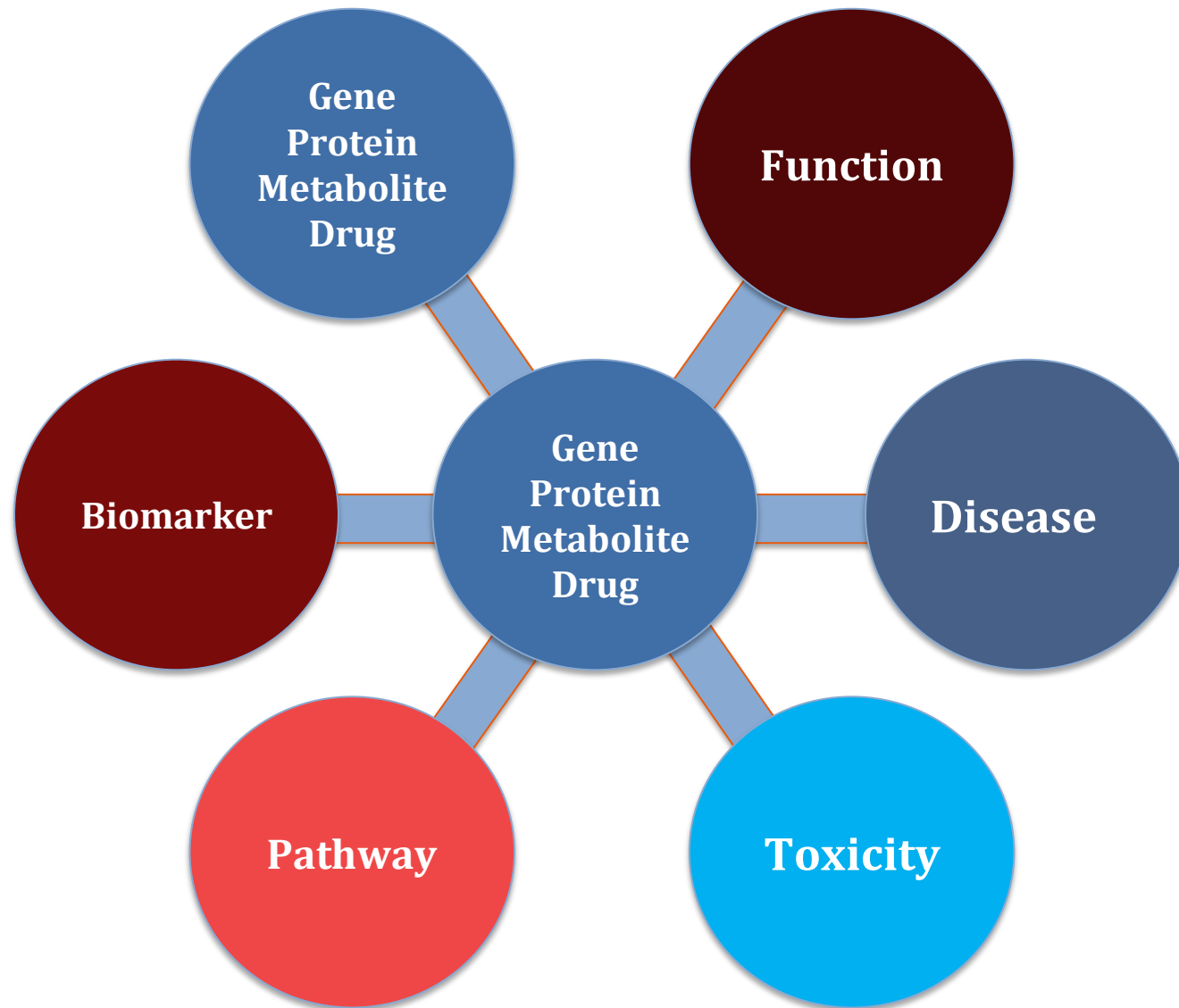
>12.6 million findings

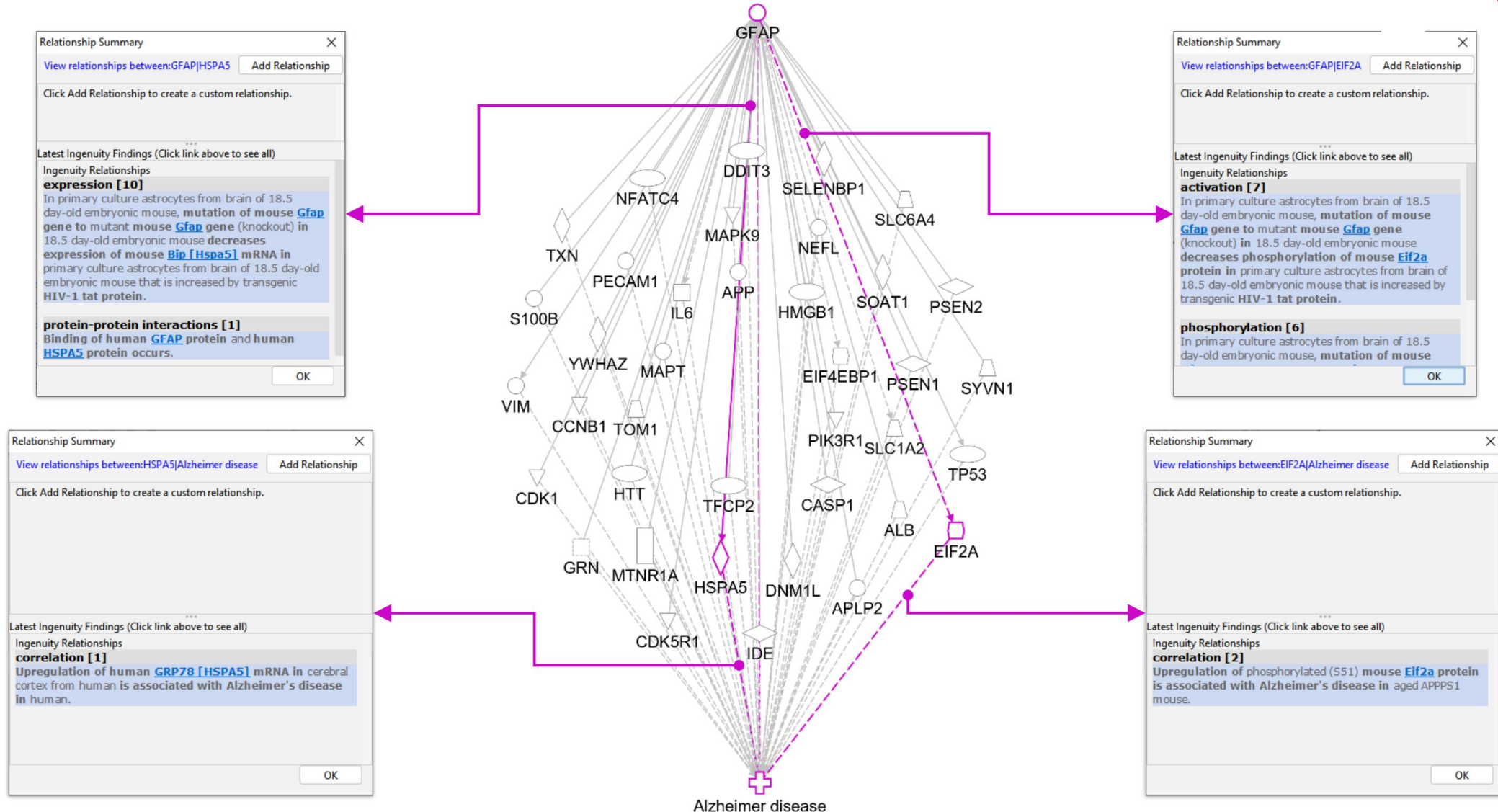
Ingenuity Pathway Analysis











Fully supported:



Human



Mouse



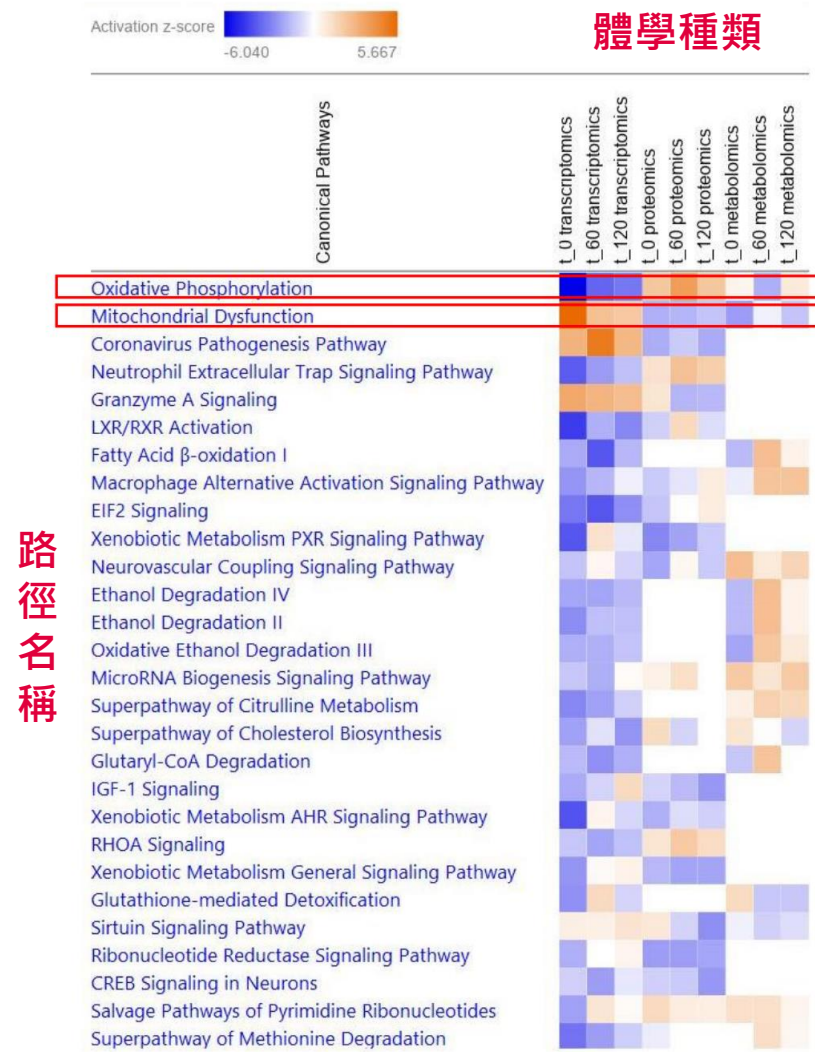
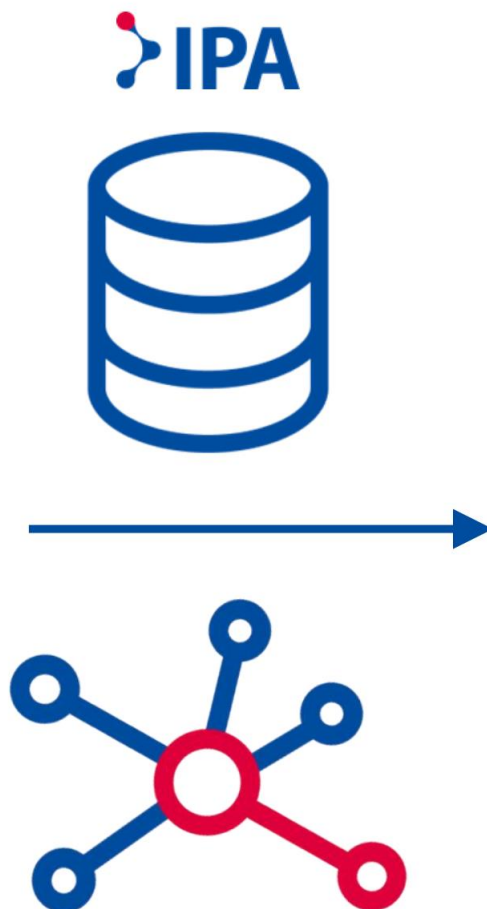
Rat

IPA can remap orthologous identifiers from the following species:

- *Arabidopsis thaliana* (*Thale cress*)
- Bat (*Rhinolophus ferrumequinum*)
- *Caenorhabditis elegans*
- Cat (domestic, *Felis catus*)
- Chicken (*Gallus gallus*)
- Chimpanzee (*Pan troglodytes*)
- Chinese hamster (*Cricetulus griseus*, CHO-K1)
- Cow (*Bos taurus*)
- Crab-eating macaque (*Macaca fascicularis*)
- Dog (*Canis lupus familiaris*)
- Fruit fly (*Drosophila melanogaster*)
- Golden hamster (*Mesocricetus auratus*)
- Guinea pig, domestic (*Cavia porcellus*)
- Horse (*Equus caballus*)
- Human (*Homo sapiens*)
- Mouse (*Mus musculus*)
- Pig (*Sus scrofa*)
- Rabbit (*Oryctolagus cuniculus*)
- Rainbow trout (*Oncorhynchus mykiss*)
- Rat (*Rattus norvegicus*)
- Rhesus Monkey (*Macaca mulatta*)
- *Saccharomyces cerevisiae*
- *Schizosaccharomyces pombe*
- Western clawed frog (*Xenopus tropicalis*)
- Zebrafish (*Danio rerio*)

Omics data type

- RNA-seq
- scRNA-seq
- Microarray
- Nanostring
- qPCR
- ChIP-seq
- Proteomics
- Metabolomics
- RNAi
- CRISPR
- WGS/WES etc.



With dataset

- Find connections in your data
- Identify novel biomarkers
- Uncover key targets and regulators
- Discover novel disease mechanisms
- Compare across experiments

Without dataset

- Search and explore the QIAGEN Knowledge Base
- Test hypothesis in silico
- Identify degree of novelty in a hypothesis

Genes and Chemicals Diseases and Functions Pathways and Lists Datasets and Analyses

Create New... EGFR Search Advanced Search

Project Manager Search Results

My Projects

- smh_miRBA
- CMU_Hung_RNAseq
- QDARDS
- exosome miRNA 2
- exosome miRNA
- 2023-demo
- Isoform
- HTCH_Dr.Liu_2022-12-16
- NDMC1020
- CGU_20221018
- TMU0816
- BIONET
- CMUHuang

Add To My Pathway Add To My List Create Dataset BioProfiler Interaction Network Activity Plot

The search for EGFR matched 158 items.

Symbol	Matched Term	Synonym(s)	Entrez Gene Name	Location
EGFR	EGFR, EGFR vIII, EGFR1, Egfr, HER1 (EGFR)	9030024J15RIK, C-ERBB, EGFR1, EGF receptor, EGFR vIII, EGF-TK, epidermal growth factor receptor, ERBB, ERBB1, ErbB1, ERBP, HER1, HER1 (EGFR), MENA, NISBD2, PIG61, wa-2, Wa5	epidermal growth factor receptor	Plasma Membran

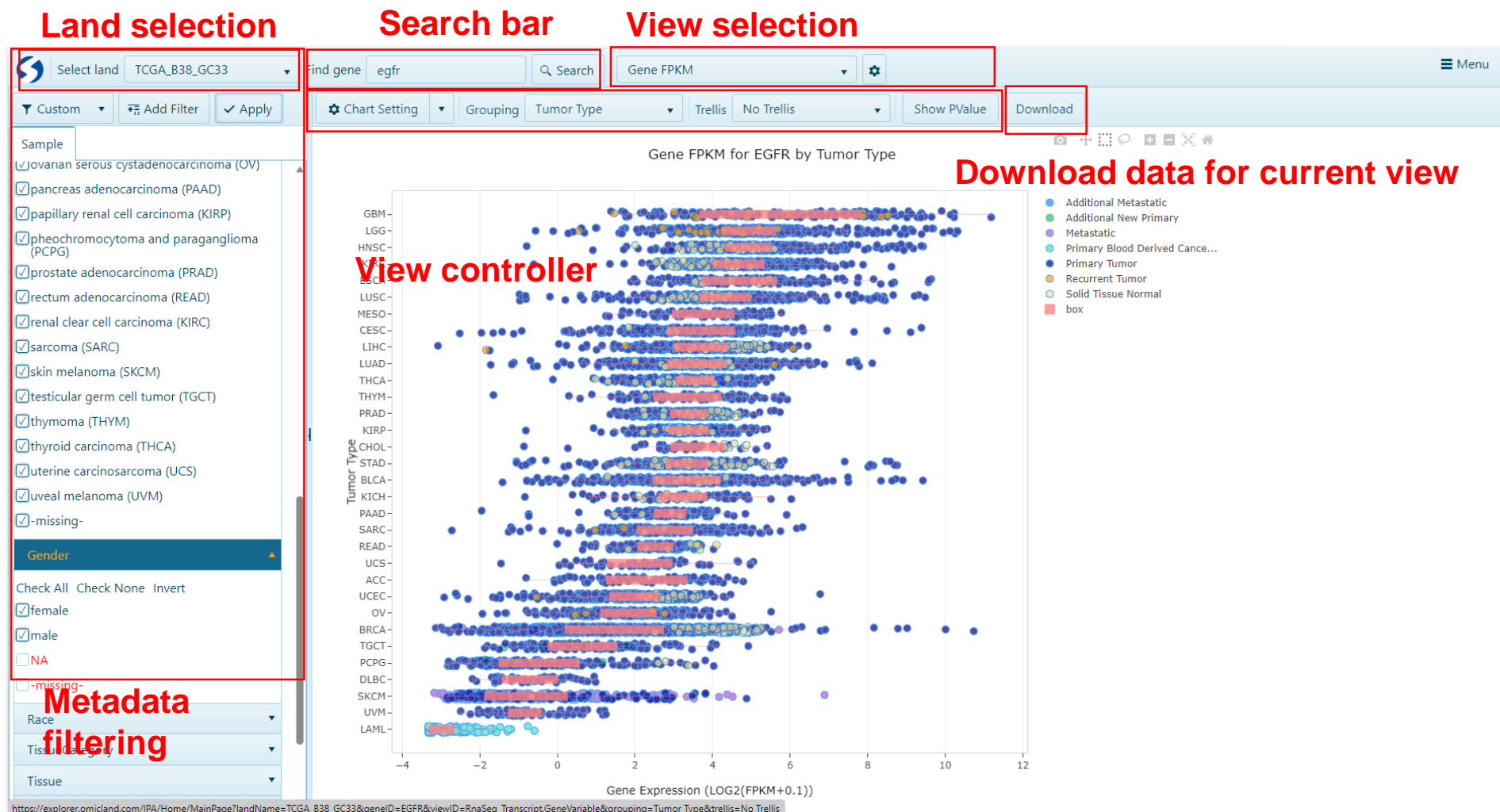
Choose which you want

OmicSoft Land Explorer: Sample-level experimental data

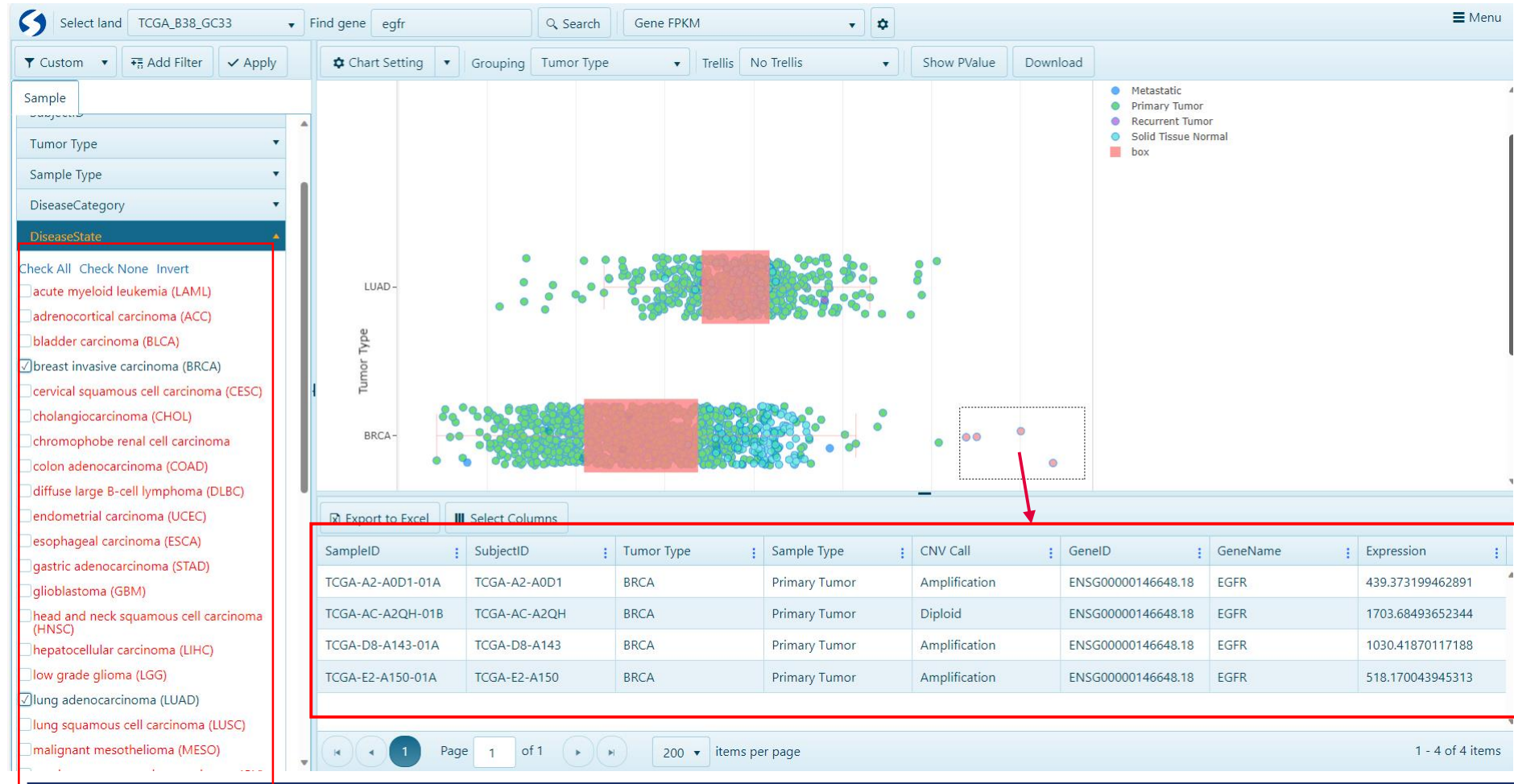
Data Type / Data Source	Normal Tissue	Cell Lines	Oncology Consortia	Oncology Studies	Disease Studies
RNA-seq expression:	Solid tissue (GTEx), Solid tissue (HPA), Blueprint	Cancer cell lines (CCLE)	TCGA, TARGET, BeatAML, ICGC, CGCI, CCLE+GTEx+TCGA, ENCODE RNA-associated gene knockdown	General oncology, Mouse studies	Human disease, Mouse disease, Rat disease
Microarray expression:	Solid tissue (GTEx)	Cancer cell lines (CCLE), Cell lines (Other)	TARGET, expO, METABRIC, CCLE+GTEx	General oncology, Metastasis, Mouse studies	Human disease, Mouse disease, Rat disease
Differential regulation:	Solid tissue (GTEx)	Treated cells (LINCS)	TCGA, TARGET, ENCODE RNA-associated gene knockdown	General oncology, Metastasis, Mouse studies	Human disease, Mouse disease, Rat disease
Alteration frequency:		Cancer cell lines (CCLE), Cell lines (Other)	TCGA, TRACERx, BeatAML, ICGC, TARGET, METABRIC	General oncology, Metastasis	
Survival by expression:			TCGA, BeatAML, TARGET, CGCI	General oncology, Clinical outcomes	
Single Cell differential regulation:	Human Cell Landscape (HCL), Tabula Sapiens			Human Disease (UMI), Human Disease (non-UMI), Mouse Disease (UMI), Mouse Disease (non-UMI)	Human Disease (UMI), Human Disease (non-UMI), Mouse Disease (UMI), Mouse Disease (non-UMI)
Protein expression:	Solid tissue (GTEx)	Cancer cell lines (CCLE)		General oncology	

IPA Gene View :OmicSoft Land Explorer

- TCGA-B38-G33



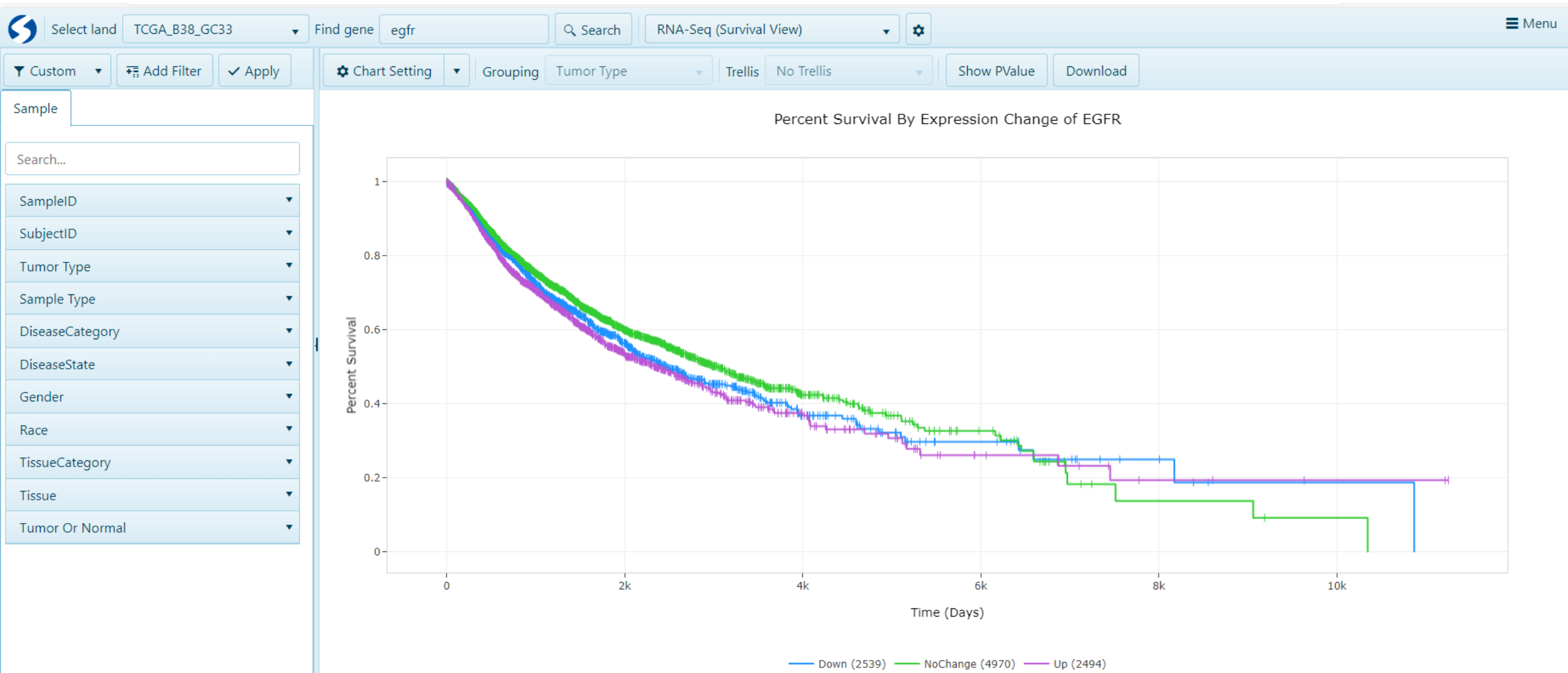
- Through DiseaseState filter, to observe the expression difference of EGFR gene in cancer type in TCGA
- Select a specific experimental group to view more detailed information.



The screenshot shows the OmicSoft Land Explorer interface. The top bar includes a search bar with 'egfr' and a 'Gene FPKM' dropdown. The left sidebar shows a 'DiseaseState' filter with a list of cancer types. The main plot area displays a scatter plot of EGFR expression (log2 FPKM) for LUAD and BRCA cancer types, with a red box highlighting a specific group of samples. A red arrow points from this box to a detailed data table below the plot.

SampleID	SubjectID	Tumor Type	Sample Type	CNV Call	GeneID	GeneName	Expression
TCGA-A2-A0D1-01A	TCGA-A2-A0D1	BRCA	Primary Tumor	Amplification	ENSG00000146648.18	EGFR	439.373199462891
TCGA-AC-A2QH-01B	TCGA-AC-A2QH	BRCA	Primary Tumor	Diploid	ENSG00000146648.18	EGFR	1703.68493652344
TCGA-D8-A143-01A	TCGA-D8-A143	BRCA	Primary Tumor	Amplification	ENSG00000146648.18	EGFR	1030.41870117188
TCGA-E2-A150-01A	TCGA-E2-A150	BRCA	Primary Tumor	Amplification	ENSG00000146648.18	EGFR	518.170043945313

TCGA_B38_GC33



Create New...

Genes and Chemicals

Diseases and Functions

Pathways and Lists

Datasets and Analyses

Sleep Disorders [abnormal sleep pattern,sleep difficulties,...]

Search

Advanced Search

Project Manager

A-Z So

QCIT_mod

selected_skin_re

selected_miRNA

myList

colon cancer-a

Ulcerative Colit

hsa_miRNA filte

hsa_miRNA

cataract

cataract.sig

HCC_CD8

HCC_relapse

HCC_relapse_d

Analyses

Nature_comm

Nature_comm

QCIT_mod - 20

selected_skin_re

selected_miRNA

colon cancer-a

Ulcerative Colit

hsa_miRNA - 20

cataract - 2024-0

Summary

Experiment M

Analysis Set

Top Canonic

Top Upstream

Top Diseases

Search Results

Diseases and Functions

Add To My Pathway

Add To My List

Annotations

Show Findings

Effect on Function

BioProfiler

Activity Plot

Show Functions

Expand Functions

The search for Sleep Disorders matched 1 diseases and functions.

Matching Diseases & Functions

Organismal Injury and Abnormalities

sleep disorder

Sleep Disorders

Neurological Disease

sleep disorder

Sleep Disorders

Psychological Disorders

sleep disorder

Sleep Disorders

Associated Molecule

312

312

312

312

312

312

312

312

Dataset and analysis search

File Edit View Window Help

Genes and Chemicals Diseases and Functions Pathways and Lists **Datasets and Analyses**

Create New...

lung cancer

Search

Advanced Search

Provide Feedback | Support

Gene Chen

Close IPA

QIAGEN Land Explorer

QIAGEN

Search Results

Diseases and Functions

Datasets and Analyses

Search Results

Showing first 5000 results out of 33129 in 18888ms for query [lung cancer]

Folder Types

dataset (17090)

analysis (16031)

VariantLossGain (8)

Open

Add to Comparison

Customize Table

Crea... 2024/... - 2024/... (1/125)

Name

Type

Creation Date

case.diseasestate

colon cancer-association - 2024-03-05 03:36 下午

analysis

2024/03/04 23:36:43

colon cancer-association

dataset

2024/03/04 23:33:24

1294- breast cancer [breast] 1293

analysis

2024/01/12 09:20:15

breast cancer

263- normal control [bladder;bone;bone marrow;brain;embryo...

analysis

2024/01/12 09:19:07

normal control

4631- breast cancer [peripheral blood] 4630

analysis

2024/01/12 09:17:53

breast cancer

4938- breast cancer [breast] 4937

analysis

2024/01/12 09:17:39

breast cancer

5223- breast cancer [breast] 5222

analysis

2024/01/12 09:17:22

breast cancer

1870- lung adenocarcinoma (LUAD);lung squamous cell carcino...

analysis

2024/01/12 09:17:15

lung adenocarci...

2446- normal control;pulmonary fibrosis [lung] 2445

analysis

2024/01/12 09:16:59

normal control;p...

6615- hepatocellular carcinoma (LIHC);intrahepatic cholangiocar...

analysis

2024/01/12 09:16:30

hepatocellular ca...

314- normal control [testis] 313

analysis

2024/01/12 09:16:24

normal control

1240- normal control [fetal lung] 1239

analysis

2024/01/12 09:16:13

normal control

3918- breast cancer [breast] 3917

analysis

2024/01/12 09:15:24

breast cancer

4042- chronic obstructive pulmonary disease (COPD);disease co...

analysis

2024/01/12 09:14:00

chronic obstruct

8970- colorectal cancer [colonrectum] 8969

analysis

2024/01/12 08:40:25

colorectal cancer

8975- colorectal cancer [colonrectum] 8974

analysis

2024/01/12 08:40:15

colorectal cancer

1- acute myeloid leukemia (LAML) [bone marrow] NA 168

analysis

2024/01/09 02:17:06

acute myeloid le

1- acute myeloid leukemia (LAML) [bone marrow] NA 213

analysis

2024/01/09 02:16:46

acute myeloid le

1- breast cancer [breast;lymph node;peripheral blood] 0

analysis

2024/01/09 02:13:03

breast cancer

1- breast cancer [breast] 68

analysis

2024/01/09 02:12:49

breast cancer

1- breast cancer [peripheral blood] NA 8

analysis

2024/01/09 02:12:37

breast cancer

1- breast carcinoma [breast] estradiol;ethanol 0

analysis

2024/01/09 02:12:21

breast carcinoma

1- breast carcinoma [breast] estradiol;ethanol 4

analysis

2024/01/09 02:12:05

breast carcinoma

1- germ cell cancer [ovary] NA 4

analysis

2024/01/09 02:09:17

germ cell cancer

1- kidney clear cell sarcoma (CCSK) [kidney] NA 14

analysis

2024/01/09 02:07:58

kidney clear cell

1- kidney rhabdoid cancer [kidney] Transfection_BAF47 442

analysis

2024/01/09 02:07:40

kidney rhabdoid

1- childhood acute lymphocytic leukemia [hematopoietic tissue]...

analysis

2024/01/09 02:02:21

childhood acute

1- endometrial cancer;endometrial squamous cell carcinoma;ova...

analysis

2024/01/09 02:01:04

endometrial can

Libraries > OmicSoft > SingleCellLand > SingleCellHumanUmi > Analyses

1870- lung adenocarcinoma (LUAD);lung squamous cell carcinoma (LUSC) [lung] 1869

Case/Control Differences

Key	Case	Control
cluster	1	0;10;11;12;13;14;15;16;17;18;19;2;20;3;4;5;6;7;8;9
clustercelltype	T cell	alveolar epithelial cell;B cell;cytotoxic T cell;endothelial cell;epithelial cell;fibroblast;macrophage;mast cell;monocyte;myeloid cell;NK cell;T cell;unassigned cell

Comparison Context

cellmarkers	CD235A-
celltype	lung cell
comparisoncategory	Cluster vs Others
comparisoncontrast	T cell (cluster) vs others
diseasestate	lung adenocarcinoma (LUAD);lung squamous cell carcinoma (LUSC)
ethnicity	Caucasian
gender	female;male
organism	human
platformname	NGS.Illumina.NextSeq500
smokingstatus	ex-smoker;NA
tissue	lung
tnmstage	pN0;pT1a;pN0;pT2a;pN1;pT1b;pNX;pT2a

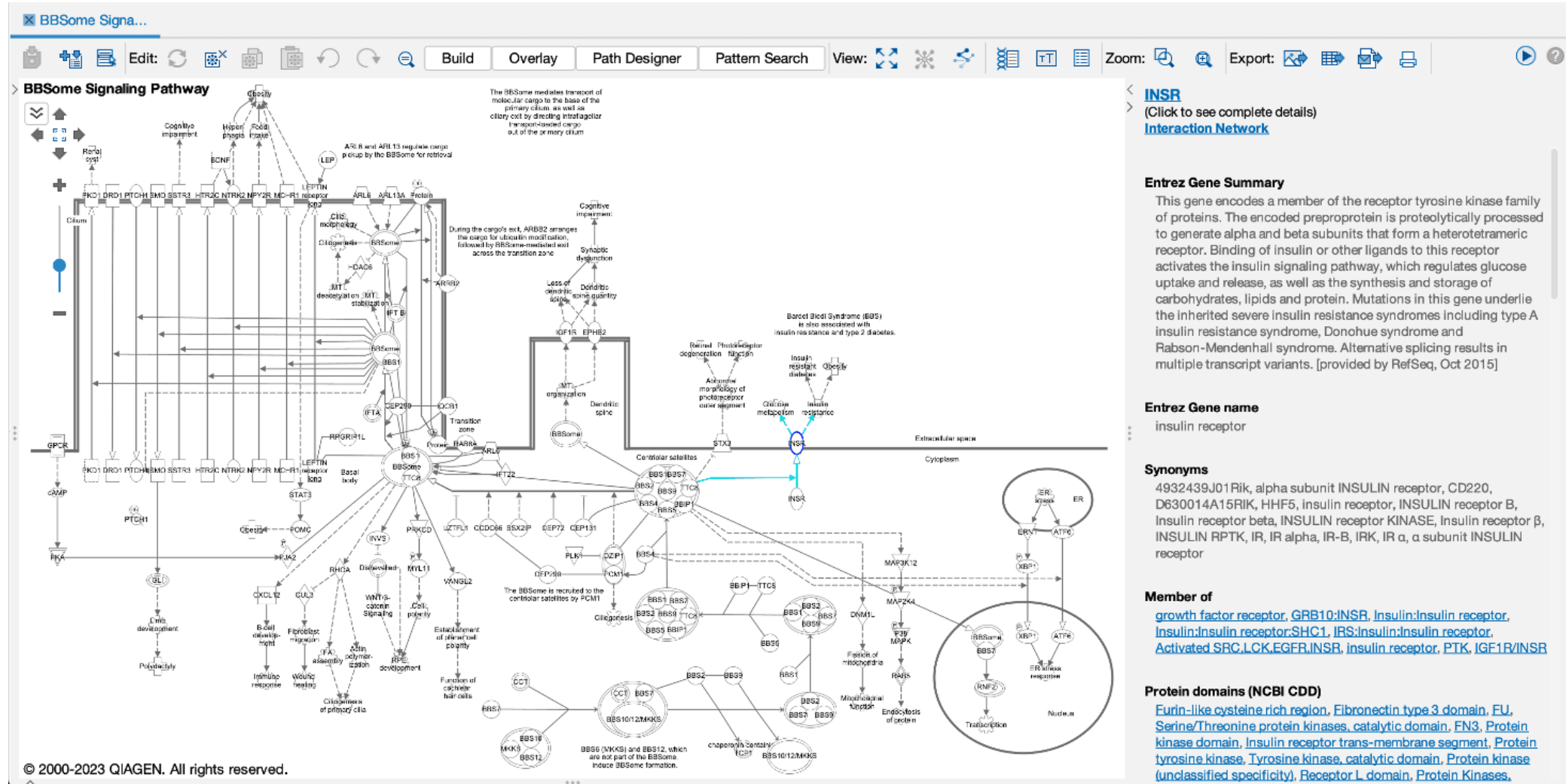
All Experiment Metadata

case.cellmarkers	CD235A-
case.celltype	lung cell
case.cluster	1
case.clustercelltype	T cell
case.diseasestate	lung adenocarcinoma (LUAD);lung squamous cell carcinoma (LUSC)
case.ethnicity	Caucasian
case.gender	female;male
case.samplematerial	cryopreserved cells;MACS depleted cells;surgical resection
case.smokingstatus	ex-smoker;NA

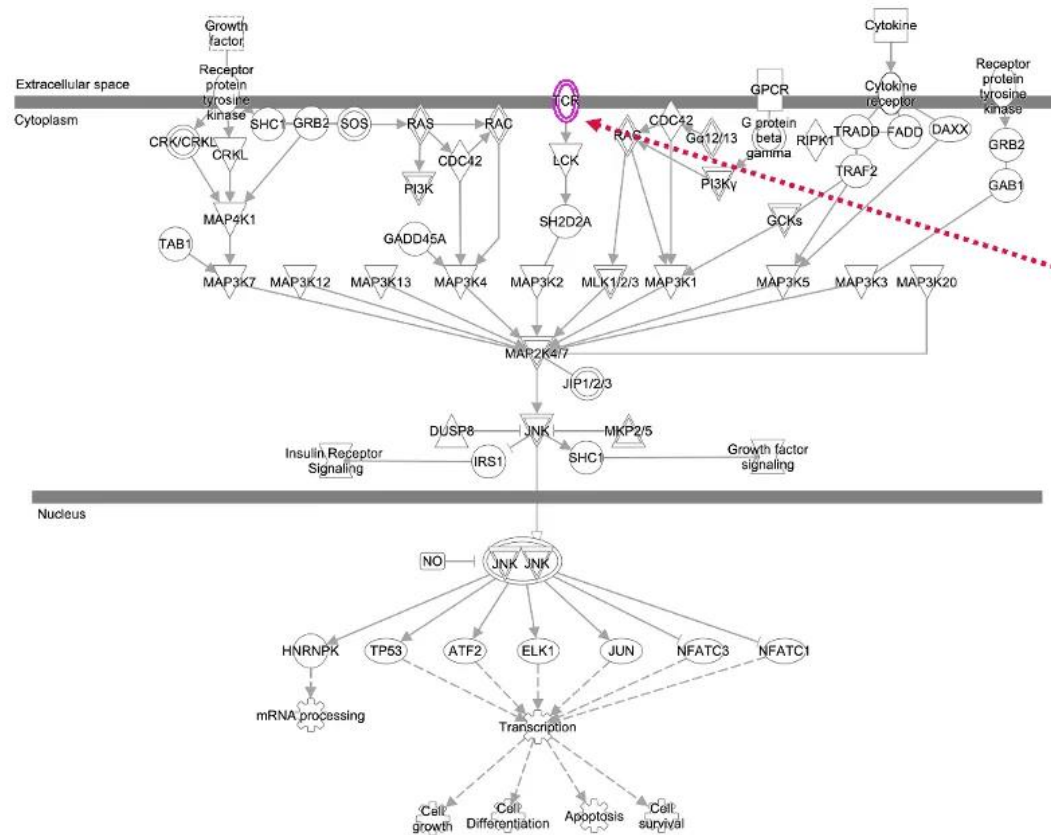
Volcano plot



Easy to find relationship summary



Easy to find relationship summary

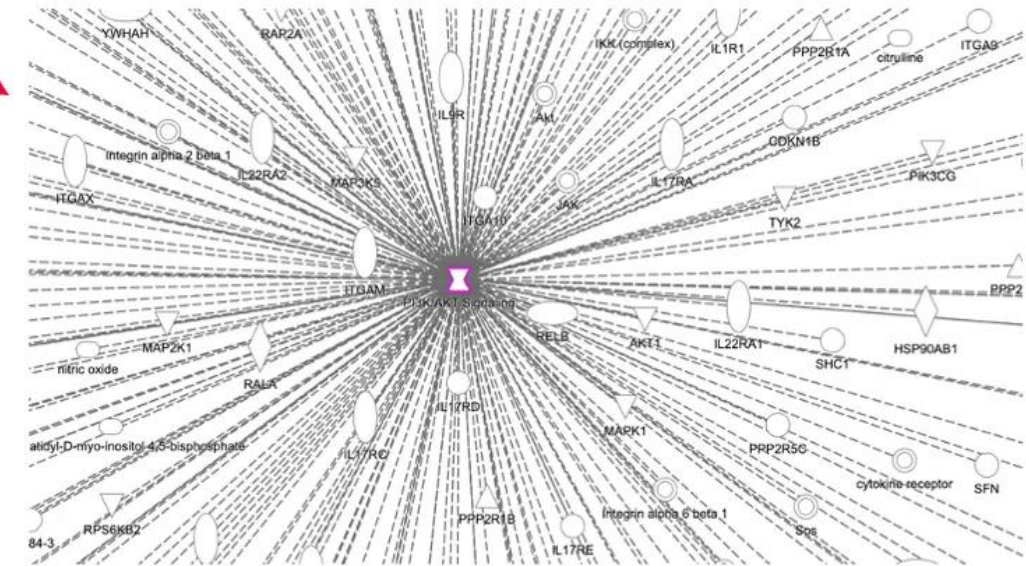
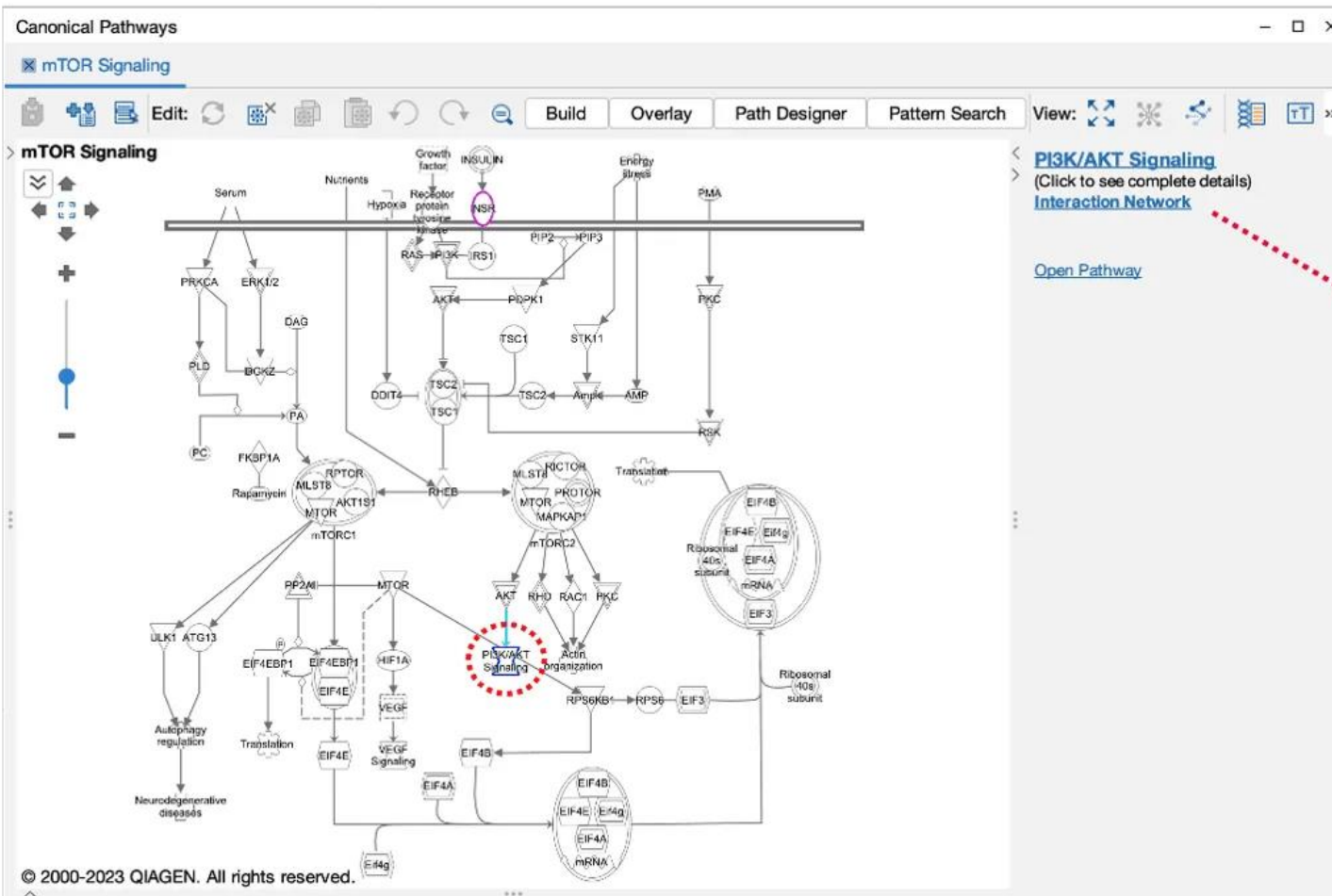


Opens pathway and highlights the node, even if it is a group for which that gene is a member

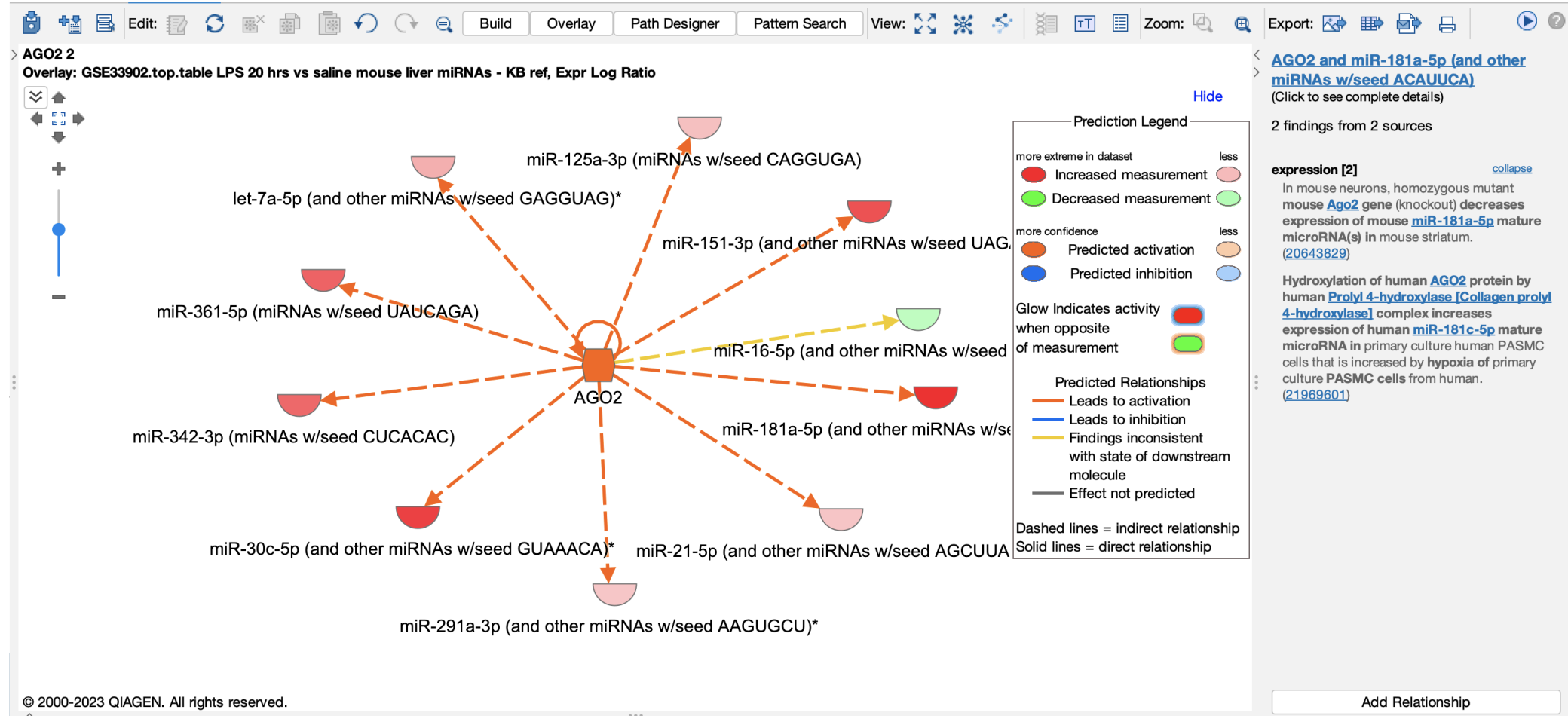
TRB
(Click to see complete details)
[Interaction Network](#)

Canonical Pathway membership
[Hematopoiesis from Pluripotent Stem Cells](#), [TEC Kinase Signaling](#), [CTLA4 Signaling in Cytotoxic T Lymphocytes](#), [T Helper Cell Differentiation](#), [CD28 Signaling in T Helper Cells](#), [Chaperone Mediated Autophagy Signaling Pathway](#), [T Cell Receptor Signaling](#), [Th1 Pathway](#), [Th2 Pathway](#), [Phospholipase C Signaling](#), [Type 1 Diabetes Mellitus Signaling](#), [Altered T Cell and B Cell Signaling in Rheumatoid Arthritis](#), [NUR77 Signaling in T Lymphocytes](#), [PKC8 Signaling in T Lymphocytes](#), [Allograft Rejection Signaling](#), [Autoimmune Thyroid Disease Signaling](#), [Graft-versus-Host Disease Signaling](#), [Regulation of IL-2 Expression in Activated and Anergic T Lymphocytes](#), [Role of NFAT in Regulation of the Immune Response](#), [G Protein Signaling Mediated by Tubby](#), [Systemic Lupus Erythematosus Signaling](#), [PD-1](#), [PD-L1 cancer immunotherapy pathway](#), [SAPK/JNK Signaling](#), [Communication between Innate and Adaptive Immune Cells](#), [NF-κB Signaling](#)
[Show more](#)

Top Findings
Binds:
 PTCRA, DVL2, B2M, CANX, CD247, TRA, CD3E, Pvr, CTCF, RUNX1
Disease:
 neoplasia, sepsis, cancer, adenoma formation, epithelial neoplasia, benign neoplasia, Sézary syndrome, pituitary gland adenoma, head and neck neuroendocrine neoplasia, adult T-cell leukemia/lymphoma
Regulates:
 TNF, IFNG, ERK1/2, NR4A1, BCL2, BAX, P38 MAPK, RELA, IL10, NFKBIA
Role in cell:
 expression in, growth, proliferation, cell death, apoptosis, activation in, cell viability, production in, activation, binding in
Regulated by:
 NOS2, lipopolysaccharide, T lymphocytes, cyclosporin A, ETS1, TRAT1, NOTCH1, 26S proteasome



Summary	Graphical Summary	Pathways	Upstream Analysis	Diseases & Functions	Regulator Effects	Networks	Lists	Analysis Match	Molecules
Upstream Regulators Causal Networks									
<div> Add To My Pathway Add To My List Display as Network Activity Plot Customize Table Mechanistic Networks </div>									
Upstream R...	Expr Log Ratio	Molecule Type	Predicted A...	Activatio...	B-H correct...	Target Molec...	Mechanistic ...		
AGO2		translation regulator	Activated	2.365	2.59E-10	↑let-7a-5p (and...all 10			
SSB		enzyme	Activated	2.000	8.96E-05	↑let-7a-5p (and ...all 4			
PARN		enzyme		1.982	2.73E-07	↑let-7a-5p (and ...all 4			
INSR		kinase		1.475	4.45E-03	↑miR-411-3p (an...all 5			
5-fluorouracil		chemical drug		1.452	7.63E-03	↓miR-16-5p (an...all 4			
IGF1R		transmembrane receptor		1.387	4.98E-04	↑miR-411-3p (an...all 5			
rimonabant		chemical drug		1.265	3.57E-11	↑miR-151-3p (a...all 10			
docetaxel		chemical drug		1.236	2.03E-05	↑let-7a-5p (and ...all 5			
EPHB6		kinase		1.213	8.38E-07	↑let-7a-5p (and ...all 5			
DAZL		translation regulator		1.119	1.98E-06	↑let-7a-5p (and ...all 4			
DICER1		enzyme		1.111	2.98E-06	↑let-7a-5p (and ...all 7			
calcifediol		chemical - endogenous...		0.456	3.91E-10	↑let-7a-5p (and ...all 6			
Gnasas1		other		0.000	5.27E-07	↑let-7a-5p (and ...all 5			
CG		complex		0.000	3.56E-03	↑let-7a-5p (and ...all 5			
hydrogen peroxide		chemical - endogenous...		-0.198	6.23E-05	↑let-7a-5p (and ...all 7			
ALOX5		enzyme		-0.215	1.86E-08	↑let-7a-5p (and ...all 9			
PAX3-FOXO1		fusion gene/product		-0.757	3.50E-06	↑let-7a-5p (and ...all 7			
PPARA		ligand-dependent nucl...		-1.342	2.05E-04	↑let-7a-5p (and ...all 6			
acetaminophen		chemical drug		-1.539	1.08E-09	↑let-7a-5p (and ...all 9			
monocrotaline		chemical toxicant		-1.982	2.40E-05	↑let-7a-5p (and ...all 4			
NORAD		other			7.53E-03	↑miR-125a-3p (...all 2			
DGCR8		enzyme			9.70E-03	↑let-7a-5p (and ...all 2			
Gulo		enzyme			1.49E-05	↑let-7a-5p (and ...all 3			
MTDH		transcription regulator			6.81E-03	↓miR-16-5p (an...all 3			
resolvin D1		chemical - endogenous...			8.93E-03	↑miR-21-5p (an...all 2			
GFI1		transcription regulator			5.43E-03	↑miR-21-5p (an...all 3			
Selected 0 / 35									



Explore eight new and two updated pathways

New Ingenuity signaling pathways

- BBSome Signaling Pathway
- Folate Signaling Pathway
- HEY1 Signaling Pathway
- IL-27 Signaling Pathway
- Sleep NREM Signaling Pathway
- Sleep REM Signaling Pathway
- UFMylation Signaling Pathway
- WNT/SHH Axonal Guidance Signaling Pathway

Existing pathways updated to include an activity pattern

- Eicosanoid Signaling Pathway
- Gap Junction Signaling Pathway

Addition of >168,000 new findings (bringing the total in IPA to over 13.1 million)

- >143,000 Expert findings (from literature curation)
- >12,600 protein-protein interaction findings from BioGrid
- >4,700 cancer mutation findings from ClinVar
- >3,900 protein-protein interaction findings from IntAct
- >1,700 drug-to-disease findings from ClinicalTrials.gov
- >1,200 target-to-disease findings from ClinicalTrials.gov
- >600 gene-to-disease findings from the Online Inheritance in Man (OMIM)
- >300 findings from Clinical Genome Resource (ClinGen)
- >100 Gene Ontology findings
- >50 newly mappable chemicals

149,657 expression datasets (2,436 added)

Land	Repository	Datasets Q3 2023	Datasets Q4 2023	Increase
DiseaseLand	HumanDisease	34,317	35,112	795
	MouseDisease	26,164	26,584	420
	RatDisease	9,287	9,287	
	LINCS	28,234	28,234	
OncoLand	OncoHuman (Formerly OncoGEO)	20,152	21,373	1,221
	OncoMouse	1,054	1,054	
	TCGA	4,438	4,438	
	MetastaticCancer	81	81	
	Hematology	4,267	4,267	
	Pediatrics	444	444	
	ENCODE RNA Binding	486	486	
	ClinicalProteomicTumor	254	254	
Single Cell Land	SingleCellHuman	194	194	
	SingleCellHumanUmi	11,049	11,049	
	SingleCellHumanUmiLite	603	603	
	SingleCellHumanHCL	1,469	1,469	
	SingleCellMouse	81	81	
	SingleCellMouseUmi	3,220	3,220	
	SingleCellMouseUmiLite	115	115	
Normal Cells and Tissues	Human Tissues (GTEx)	1,312	1,312	

Comprehensive germline genomic profiles of children, adolescents and young adults with solid tumors



Hand on Case Study

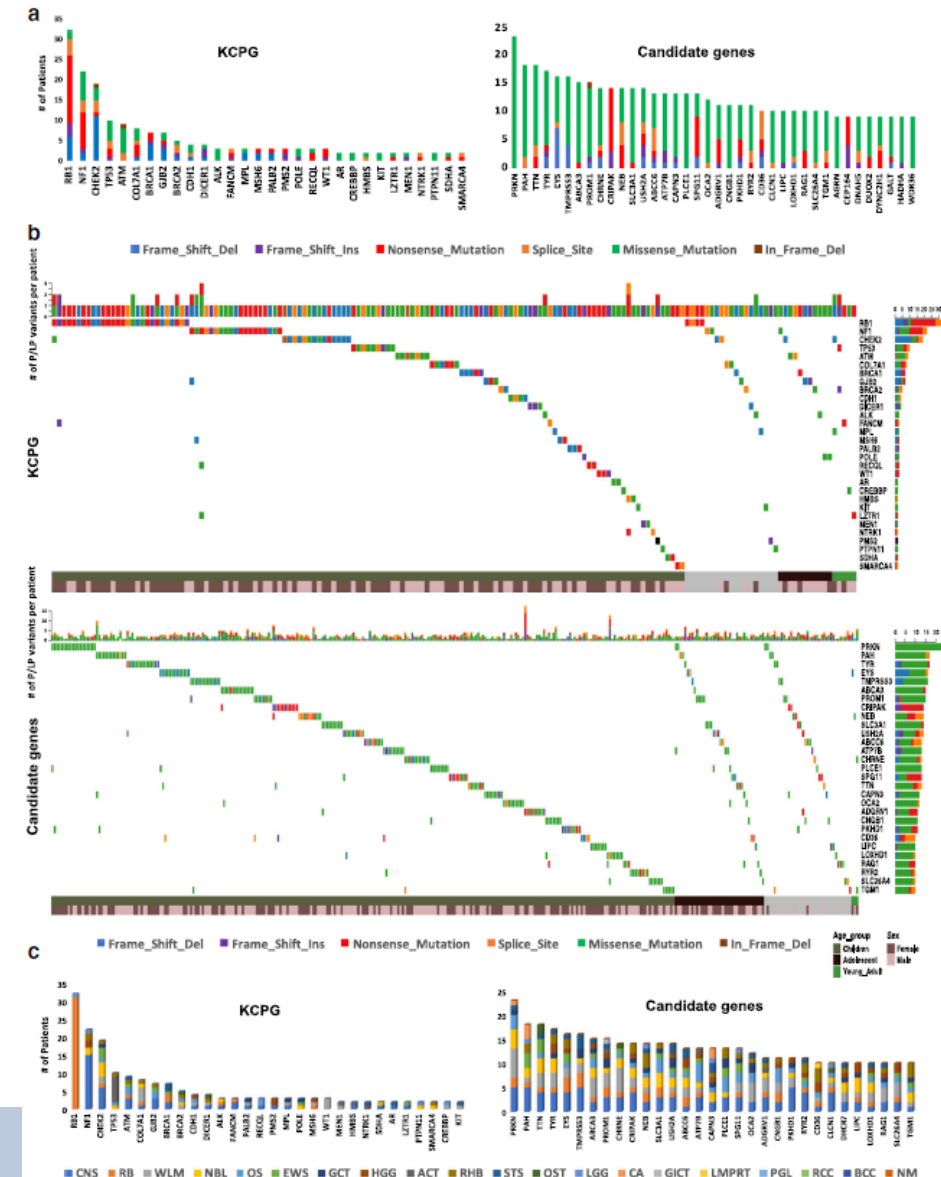
In a study of 1,507 C-AYA patients with solid tumors, 12% had pathogenic variants in known cancer-predisposing genes (KCPG), and 61% had such variants in non-KCPG genes like PRKN, SMARCA1, and SMAD7. Pathway analysis pinpointed p53 as a key network. Drug-target analysis revealed that 1/3 of patients with germline P/LP variants had druggable alterations, with over half from non-KCPG genes, emphasizing the significance of considering candidate genes for targeted therapies.

Akhavanfard, S., Padmanabhan, R., Yehia, L. *et al.* Comprehensive germline genomic profiles of children, adolescents and young adults with solid tumors. *Nat Commun* 11, 2206 (2020).
<https://doi.org/10.1038/s41467-020-16067-1>

Source	CCF/PCGP/SJLIFE				
Age group	Children	Adolescent	Young adult	Unknown	Total
Female/Male	600/582	67/97	5/15	14/13 ^a	686/707
Mean age of onset	6.1 ± 4.5	16.9 ± 1.3	22 ± 2.4	NA	7.6 ± 5.8
All solid tumors (1507)	1182	164	20	141	1507
Central nervous system (323)	266	23	2	32	323
Wilms tumor (207)	189	2	1	15	207
Neuroblastoma (190)	158	2	1	29	190
Rhabdomyosarcoma (134)	114	14	1	5	134
Osteosarcoma (129)	78	42	5	4	129
Retinoblastoma (98)	84			14	98
Ewing's sarcoma (95)	58	27	5	5	95
Soft tissue sarcoma (93)	69	17	3	4	93
High-grade glioma (80)	63	8		9	80
Germ cell tumor (74)	57	11	1	5	74
Low-grade glioma (24)	8	1		15	24
Adrenocortical carcinoma (22)	20	1		1	22
Carcinoma (14)	7	7			14
Giant cell tumor (3)	0	3			3
Renal cell carcinoma (3)	2		1		3
Low malignant potential renal tumors (2)	2				2
Basal cell carcinoma (1)	1				1
Paraganglioma (1)		1			1
Other solid tumor (10)	3	4		3	10
Non-malignant tumor (4)	3	1			4

CCF Cleveland Clinic Foundation, PCGP Pediatric Cancer Genome Project, SJLIFE, St. Jude Life Cohort. ^aGender data were not available for all the patients in this group.

Fig. 2 Distribution of germline pathogenic/likely pathogenic (P/LP) mutations in children, adolescents, and young adults (C-AYA) with solid tumors



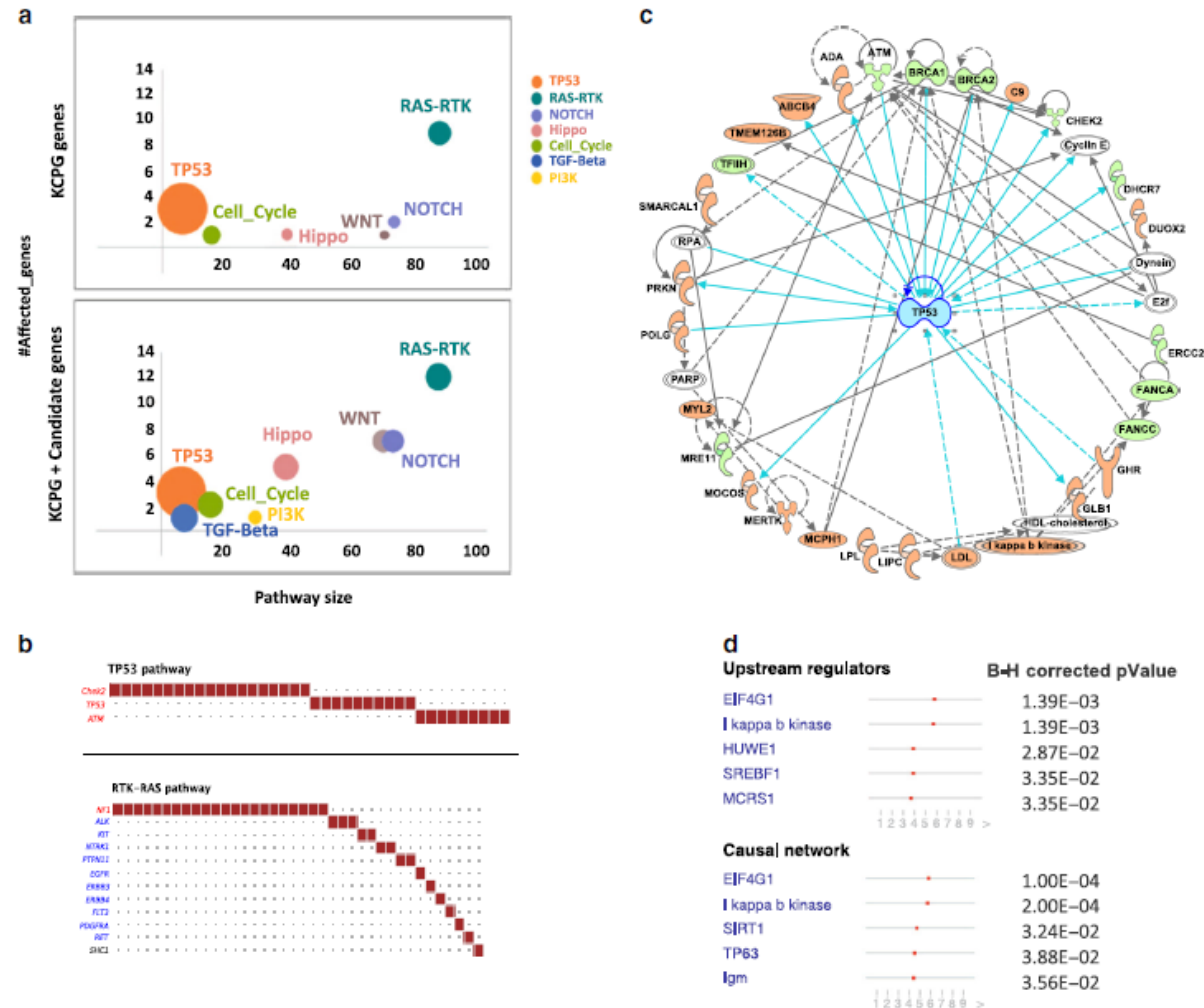


fig. 4 Pathway analysis of altered genes with germline pathogenic/likely pathogenic (P/LP) variants in children, adolescents, and young adults C-AYA) with solid tumors. **a** Affected pathways based on altered genes with P/LP germline variants. Top panel: only known cancer-predisposing genes KCPG, lower panel: a combination of all KCPGs and candidate genes. Size of the circles increases as the fraction affected increases. **b** Genes mutated in P53 (top panel) and RAS-RTK (lower panel) pathways, and the number of patients affected in our cohort. Red font: tumor suppressor genes; blue font: oncogenes. **c** Top network, predicted by Ingenuity Pathway Analysis (IPA), based on all the KCPG (green color) and candidate genes (salmon color) with at least four P/LP variants in our C-AYA patients with solid tumors (right-tailed Fisher's exact test $P = 1 \times 10^{-42}$). **d** Eukaryotic Translation Initiation Factor 4 gamma 1 (EIF4G1, B-H corrected $P = 1.39 \times 10^{-3}$) and I kappa b kinase (Ikb kinase, B-H corrected $P = 1.39 \times 10^{-3}$) predicted to be the top upstream regulators/causal network based on our IPA analysis. Right-tailed Fisher's exact test was used, and Benjamini-Hochberg (B-H) P value correction performed to reduce the false discovery rate (FDR).

	Observation 1		Observation 2	
Molecule Identifier	case group1 vs control Log2FoldChange	case group1 vs control PValue	case group2 vs control Log2FoldChange	case group2 vs control PValue
MARC1	0.0329	0.9119	-0.0845	0.8191
MARCH1	-0.6943	0.1086	0.8869	0.1408
SEPT1	-0.302	0.5747	-0.6987	0.3448
DEC1	-0.0312	0.9897	-2.999	0.4243
A1CF	0.7462	0.408	0.3415	0.7623
A2M	0.5054	0.3388	-1.2587	0.0047
A2MP1	0.709	0.5078	-0.5113	0.7604
A3GALT2	0.9241	0.8273	2.3056	0.4726
AAAS	0.0373	0.8668	0.0646	0.716
AACS	-0.0859	0.7724	-0.5843	0.1295
ABAT	-0.114	0.854	-0.4218	0.4668

IDs (required)

Significance (optional)

Log2, ratio, fold change, etc. (recommended)

Common identifier types

- RNA/scRNA-seq: Ensembl, Gene symbols (Entrez or Hugo)
- Proteomics: UniProt, GenPept (Entrez or HUGO)
- Metabolomics: KEGG, CAS registry, HMDB, PubChem.

Accepted file formats

- .txt (tab-delimited text files)
- .xls, .xlsx, .csv (Excel tables)
- .diff (Cuffdiff output)

IDs are the only **required** column

Change values are needed to calculate activity predictions

Get more complete mapping during dataset upload!

Vendor IDs	Gene	Protein	Transcript	microRNA	SNP	Chemical
Affymetrix (na36)	Entrez Gene (2020/10)	GenPept	Ensembl (101)	miRBase (mature)	Affy SNP IDs	CAS Registry Number
Agilent	GenBank (239)	International Protein Index (IPI)	RefSeq (human 、 mouse)	miRBase (stemloop)	dbSNP	HMDB
Life Tech (ABI)	Symbol-human (HUGO/ HGNC, EG)	UniProt/ Swiss-Prot Accession (2020_03)	UCSC (hg18)			KEGG
Codelink	Symbol- mouse (EG)		UCSC (hg19)			PubChem CID
Illumina	Symbol- rat (EG)		UCSC (hg38)			
Ingenuity	GI Number					
	UniGene					

Gene
symbol

	A	B	C
1	Hugo_SymIVA_Prediction		
2	BRCA2	2	
3	GJB2	2	
4	TP53	2	
5	ABCA3	1	
6	ABCA3	2	
7	ABCC6	1	
8	ACADS	2	
9	ACADS	2	
10	ACSF3	1	
11	AGBL5	2	
12	AGRN	1	
13	AIPL1	2	
14	ALDOB	2	
15	ALG13	1	
16	APOC3	2	
17	AQP5	2	
18	ARMC4	2	
19	ATP8B1	2	
20	BBS1	2	
21	BBS12	2	
22	BLOC1S3	1	
23	C9	2	
24	CAPN1	2	
25	CD36	2	
26	CD96	2	
27	CDHR1	1	
28	CDKAL1	2	
29	CHD7	1	
30	CIB2	2	
31	CLCN1	2	
32	CLN8	1	
33	CLPB	1	
34	CLRN1	2	

ACMG value

Allowable values are:

- 2 benign
- 1 likely benign
- 0 uncertain significance
- 1 likely pathogenic
- 2 pathogenic

GWAS rs id

	A
1	Risk loci
2	rs6012915
3	rs6095946
4	rs6091189
5	rs6014965
6	rs1741640
7	rs9983528
8	rs4616575
9	rs130651
10	rs5751474
11	rs34256596
12	rs9614460
13	rs9330814
14	rs2732875
15	rs174537
16	rs10751097
17	rs11236187
18	rs117042741
19	rs55864876
20	rs2155065
21	rs3087967
22	rs497916
23	rs10774214
24	rs3217810
25	rs3217874
26	rs10849432
27	rs10849434
28	rs77969132
29	rs2730985

Multiple ID columns

	A	B
1	Risk loci	Mapped gene
2	rs6012915	LINC01271
3	rs6095946	COX6CP2
4	rs6091189	RIPOR3
5	rs6014965	BMP7
6	rs1741640	LAMA5
7	rs9983528	PCNT
8	rs4616575	ZNRF3
9	rs130651	PDGFB
10	rs5751474	SCUBE1
11	rs34256596	LINC01639
12	rs9614460	SMC1B
13	rs9330814	WNT7B
14	rs2732875	SHROOM2
15	rs174537	MYRF
16	rs10751097	ANO1
17	rs11236187	POLD3
18	rs117042741	XRRA1

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Enter gene names/symbols/IDs or chemical/drug names here

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Getting started

Exploring large public data resources through IPA

Exploring IPA knowledge

Analyzing mRNA or proteomics data

Analyzing microRNA data

Analyzing phosphoproteomics data

Analyzing genetic gain/loss data

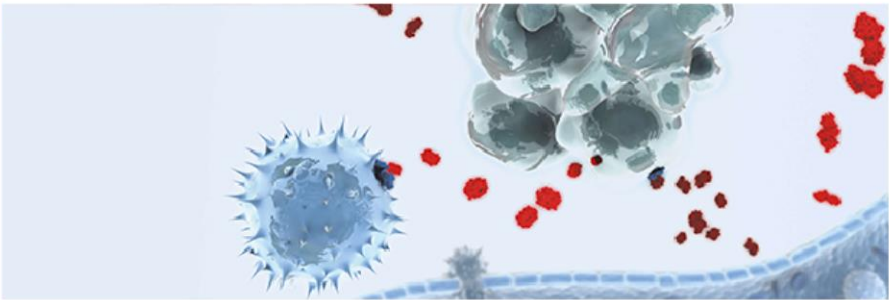
Analyzing metabolomics data

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News

- Starting in Jan 2024, analyses >10 years old will be eligible for [automatic deletion](#).
- Sign up for an [IPA training webinar!](#)
- Check out our NEW [1-minute IPA promo video](#).
- Check out the [single cell expression tutorial](#).
- Read and watch our [gene expression tutorial](#) with embedded videos.
- The IPA downtime for the upcoming **IPA 2023 Winter Release** has been scheduled for:
 - Pacific Standard Time: Friday, December 15, 5 p.m. through Sunday, December 17, 12 p.m. PST (Noon).
 - Central European Time: Saturday, December 16, 02:00 through Sunday, December 17, 21:00.
 - Japan Standard Time: Saturday, December 16, 10:00 through Monday, December 18, 05:00.
 - China CST (Beijing): Saturday, Saturday, December 16, 23:00 through Monday, December 18, 18:00.
- If you are new to IPA or taking a trial please see: [IPA Trial Resources](#).
- Pave your way to greatness using advanced pathway analysis: [Learn more](#).
- Read our [news](#) and sign up for our newsletter.
- Search Google Scholar for [publications that cite IPA](#).
- See how the IPA team is using Machine Learning in a [new paper](#).

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Dataset Upload - Nature_comm_2020_germline_variants_ACMG.xlsx

— □ ×

1. Select File Format: Flexible Format ?
2. Contains Column Header: ☒ Yes ☐ No
3. Select Identifier Type: Please assign at least one column below as "ID", and assign the identifier type(s).
Assign additional columns as ID to improve mapping coverage if desired.
4. Array platform used for experiments: Not specified/applicable Select relevant array platform as a reference set for data analysis.
5. Use the dropdown menus to specify the column names that contain identifiers and observations. For observations, select the appropriate measurement value type.

Raw Data (100) Dataset Summary (99) Metadata

ID/Observation Name	ID	Observatio...
Measurement/Annotation	Gene Symb...	Expr Fold C...
1	Hugo_Symbol	IVA_Prediction
2	BRCA2	2
3	GJB2	2
4	TP53	2
5	ABCA3	1
6	ABCA3	2
7	ABCC6	1
8	ACADS	2
9	ACADS	2
10	ACSF3	1
11	AGBL5	2
12	AGRN	1
13	AIPL1	2
14	ALDOB	2
15	ALG13	1
16	APOC3	2
17	AQP5	2
18	ARMC4	2
19	ATP8B1	2
20	BBS1	2
21	BBS12	2
22	BLOC1S3	1
23	C9	2
24	CAPN1	2
25	CD36	2

Variant ACMG Classification - expected values: [-2, 2]

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Pathways and Lists

Datasets and Analyses

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QIAGEN Land Explorer

QIAGEN

Annotated Dataset: Nature_comm_2020_germline_variants_ACMG

Preview Dataset Nature_comm_2020_germline_variants_ACMG

Mapped IDs (99)

Unmapped IDs (0)

All IDs (99)

Metadata

Add To My Pathway

Add To My List

Create Dataset

Customize Table

Symbol

ABCA3 - WFS1 (1/1)

Variant ACMG Classification	ID	Flags	Symbol	Entrez Gene Name	Location	Type(s)	Drug(s)
1.000	ABCA3	D	ABCA3	ATP-binding cassette subfamily A member 3	Plasma Membrane	transporter	
2.000	ABCA3	D	ABCA3	ATP-binding cassette subfamily A member 3	Plasma Membrane	transporter	
1.000	ABCC6		ABCC6	ATP-binding cassette subfamily C member 6	Plasma Membrane	transporter	
2.000	ACADS	D	ACADS	acetyl-CoA dehydrogenase	Cytoplasm	enzyme	
2.000	ACADS	D	ACADS	acetyl-CoA dehydrogenase	Cytoplasm	enzyme	
1.000	ACSF3		ACSF3	acyl-CoA dehydrogenase family class III member 3	Cytoplasm	enzyme	
2.000	AGBL5		AGBL5	acylglycerol-binding protein-like 5	Cytoplasm	peptidase	
1.000	AGRN		AGRN	agmatase	Cytoplasm	other	
2.000	AIPL1		AIPL1	aryl glycerophosphorylase-like 1	Cytoplasm	other	
2.000	ALDOB		ALDOB	aldose B dehydrogenase	Cytoplasm	enzyme	
1.000	ALG13		ALG13	alpha-mannosidase 13	Cytoplasm	enzyme	
2.000	APOC3		APOC3	apolipoprotein C-III	Plasma Membrane	transporter	volanesorsen
2.000	AQP5		AQP5	aquaporin 5	Plasma Membrane	transporter	
2.000	ATP8B1		ATP8B1	ATPase 8B family class 1 member 1	Plasma Membrane	transporter	
2.000	BBS1		BBS1	blindness 1	Cytoplasm	other	
2.000	BBS12		BBS12	blindness 12	Cytoplasm	other	
1.000	BLOC1S3		BLOC1S3	brain lipid oxo-L-lysine decarboxylase 1 subunit 3	Cytoplasm	transporter	
2.000	BRCA2		BRCA2	breast cancer 2	Nucleus	transcription regulator	
2.000	C9		C9	complement component 9	Plasma Membrane	other	
2.000	CAPN1		CAPN1	calpain 1	Cytoplasm	peptidase	alicapostat, BLD-2660
2.000	CD36		CD36	cluster of differentiation 36	Plasma Membrane	transmembrane receptor	VT1021, zeaxanthin
2.000	CD96		CD96	cluster of differentiation 96	Plasma Membrane	other	GSK6097608
1.000	CDHR1		CDHR1	conduction hearing related 1	Cytoplasm	other	
2.000	CDKAL1		CDKAL1	cyclin dependent kinase associated like 1	Cytoplasm	enzyme	
1.000	CHD7		CHD7	chromatin domain 7	Cytoplasm	enzyme	
2.000	CIB2		CIB2	calcium and integrin binding family member 2	Cytoplasm	kinase	
2.000	ICK		ICK	iliogenesis associated kinase 1	Cytoplasm	kinase	
2.000	CLCN1		CLCN1	chloride voltage-gated channel 1	Plasma Membrane	ion channel	
1.000	CLN8		CLN8	CLN8 transmembrane ER and ERGIC protein	Cytoplasm	other	
1.000	CLPB		CLPB	ClpB family mitochondrial disaggregase	Nucleus	transcription regulator	
2.000	CLRN1		CLRN1	clarin 1	Plasma Membrane	other	

0 / 99

Flags:

"D" - Duplicates. Gene/Protein/Chemical identifiers marked with an asterisk indicate that multiple identifiers in the dataset file map to a single gene/chemical in the Global Molecular Network.

"O" - Override molecules. Gene/Protein/Chemical identifiers marked as "Override" are displayed with italic text.

"A" - Gene/Protein/Chemical ID marked as Absent. The gene/protein/chemical will not be used as a focus molecule or appear in networks unless you also explicitly override this flag with the Override column.

Create Core Analysis

Selected Dataset: Nature_comm_2020_germline_variants_ACMG

Based on this dataset, which Core Analysis type would you like to run?

Variant Effects Analysis

On which measurement type would you like to base the analysis?

Variant ACMG Classification

This measurement will be used to calculate directionality (z-scores) in the analysis and will be displayed in color on pathways and networks. If you choose a non-directional measurement (e.g. p-value) then z-scores will not be calculated.

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Dataset Upload - Nature_comm_2020_germline_variants_ACMG.xlsx


1. Select File Format: Flexible Format
2. Contains Column Header: ☒ Yes ☐ No
3. Select Identifier Type: Please assign at least one column below as "ID", and assign the identifier type(s).
Assign additional columns as ID to improve mapping coverage if desired.
4. Array platform used for experiments: Not specified/applicable Select relevant array platform as a reference set for data analysis.
5. Use the dropdown menus to specify the column names that contain identifiers and observations. For observations, select the appropriate measurement value type.

Raw Data (100) Dataset Summary (99) Metadata

ID/Observation Name	ID	Observatio...
Measurement/Annotation	Gene Symb...	Expr Fold C...
1	Hugo_Symbol	IVA_Prediction
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9	ACADS	2
10	ACSF3	1
11	AGBL5	2
12	AGRN	1
13	AIPL1	2
14	ALDOB	2
15	ALG13	1
16	APOC3	2
17	AQP5	2
18	ARMC4	2
19	ATP8B1	2
20	BBS1	2
21	BBS12	2
22	BLOC1S3	1
23	C9	2
24	CAPN1	2
25	CD36	2

Warning

Your dataset does not contain any metadata.
Metadata is useful when you search for datasets or subsequent analyses.

 Please click on the Metadata tab to add keywords that describe the data.

Click OK to continue to save the dataset without metadata.

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Biomarker Comparison Analysis...

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Datasets and Analyses

Search

Advanced Search

germline_variants_ACMG

germline_variants_ACMG

All IDs (99)

Metadata

Table

Create Dataset

Customize Table

Symbol

ABCA3 - WFS1 (1/1)

ID	Flags	Symbol	Entrez Gene Name	Location	Type(s)	Drug(s)
ABCA3	D	ABCA3*	ATP binding cassette subfamily A member 3	Plasma Membrane	transporter	
ABCA3	D	ABCA3*	ATP binding cassette subfamily A member 3	Plasma Membrane	transporter	
ABCC6		ABCC6	ATP binding cassette subfamily C member 6	Plasma Membrane	transporter	
ACADS	D	ACADS*	acyl-CoA dehydrogenase short chain	Cytoplasm	enzyme	
ACADS	D	ACADS*	acyl-CoA dehydrogenase short chain	Cytoplasm	enzyme	
ACSF3		ACSF3	acyl-CoA synthetase family member 3	Cytoplasm	enzyme	
AGBL5		AGBL5	AGBL carboxypeptidase 5	Cytoplasm	peptidase	
AGRN		AGRN	agrin	Plasma Membrane	other	
AIPL1		AIPL1	aryl hydrocarbon receptor interacting prot...	Nucleus	other	
ALDOB		ALDOB	aldolase, fructose-bisphosphate B	Cytoplasm	enzyme	
ALG13		ALG13	ALG13 UDP-N-acetylglucosaminyltransfer...	Cytoplasm	enzyme	
APOC3		APOC3	apolipoprotein C3	Extracellular Space	transporter	volanesorsen
AQP5		AQP5	aquaporin 5	Plasma Membrane	transporter	
ATP8B1		ATP8B1	ATPase phospholipid transporting 8B1	Plasma Membrane	transporter	
BBS1		BBS1	Bardet-Biedl syndrome 1	Extracellular Space	other	
BBS12		BBS12	Bardet-Biedl syndrome 12	Extracellular Space	other	
BLOC1S3		BLOC1S3	biogenesis of lysosomal organelles compl...	Cytoplasm	transporter	
BRCA2		BRCA2	BRCA2 DNA repair associated	Nucleus	transcription regulator	
C9		C9	complement C9	Extracellular Space	other	
CAPN1		CAPN1	calpain 1	Cytoplasm	peptidase	alicapistat, BLD-2660
CD36		CD36	CD36 molecule	Plasma Membrane	transmembrane receptor	VT1021, zeaxanthin
CD96		CD96	CD96 molecule	Plasma Membrane	other	GSK6097608
CDHR1		CDHR1	cadherin related family member 1	Plasma Membrane	other	
CDKAL1		CDKAL1	CDK5 regulatory subunit associated protei...	Cytoplasm	enzyme	
CHD7		CHD7	chromodomain helicase DNA binding prot...	Nucleus	enzyme	
CIB2		CIB2	calcium and integrin binding family memb...	Cytoplasm	kinase	
ICK		ICK	ciliogenesis associated kinase 1	Cytoplasm	kinase	
CLCN1		CLCN1	chloride voltage-gated channel 1	Plasma Membrane	ion channel	
CLN8		CLN8	CLN8 transmembrane ER and ERGIC protein	Cytoplasm	other	

0 / 99

Flags:

"D" - Duplicates. Gene/Protein/Chemical identifiers marked with an asterisk indicate that multiple identifiers in the dataset file map to a single gene/chemical in the Global Molecular Network.

"O" - Override molecules. Gene/Protein/Chemical identifiers marked as "Override" are displayed with italic text.

"A" - Gene/Protein/Chemical ID marked as Absent. The gene/protein/chemical will not be used as a focus molecule or appear in networks unless you also explicitly override this flag with the Override column.

Edit Dataset Settings

Analyze/Filter Dataset

Close

—

44

Variant Effects Analysis - Nature_comm_2020_germline_variants_ACMG - 2024-03-20 10:26 下午



Summary Graphical Summary Pathways Upstream Analysis Diseases & Functions Regulator Effects Networks Lists Analysis Match Molecules

Export: 

> Experiment Metadata

> Analysis Settings

> Top Canonical Pathways

Name		p-value	Overlap
Retinoid metabolism and transport		3.73E-05	9.1 % 4/44
Plasma lipoprotein assembly, remodeling, and clearance		3.18E-04	5.3 % 4/76
HDR through MMEJ (alt-NHEJ)		1.16E-03	16.7 % 2/12
Sirtuin Signaling Pathway		1.41E-03	2.1 % 6/285
Sensory processing of sound by outer hair cells of the cochlea		1.69E-03	5.5 % 3/55

1 2 3 4 5 6 7 8 9 >






> Top Upstream Regulators

> Upstream Regulators

Name		p-value	Predicted Activation
SREBF1		2.26E-05	
ASGR1		5.22E-05	
PPARGC1A		1.82E-04	
SIRT1		2.82E-04	
OSM		2.96E-04	

1 2 3 4 5 6 7 8 9 >

> Causal Network

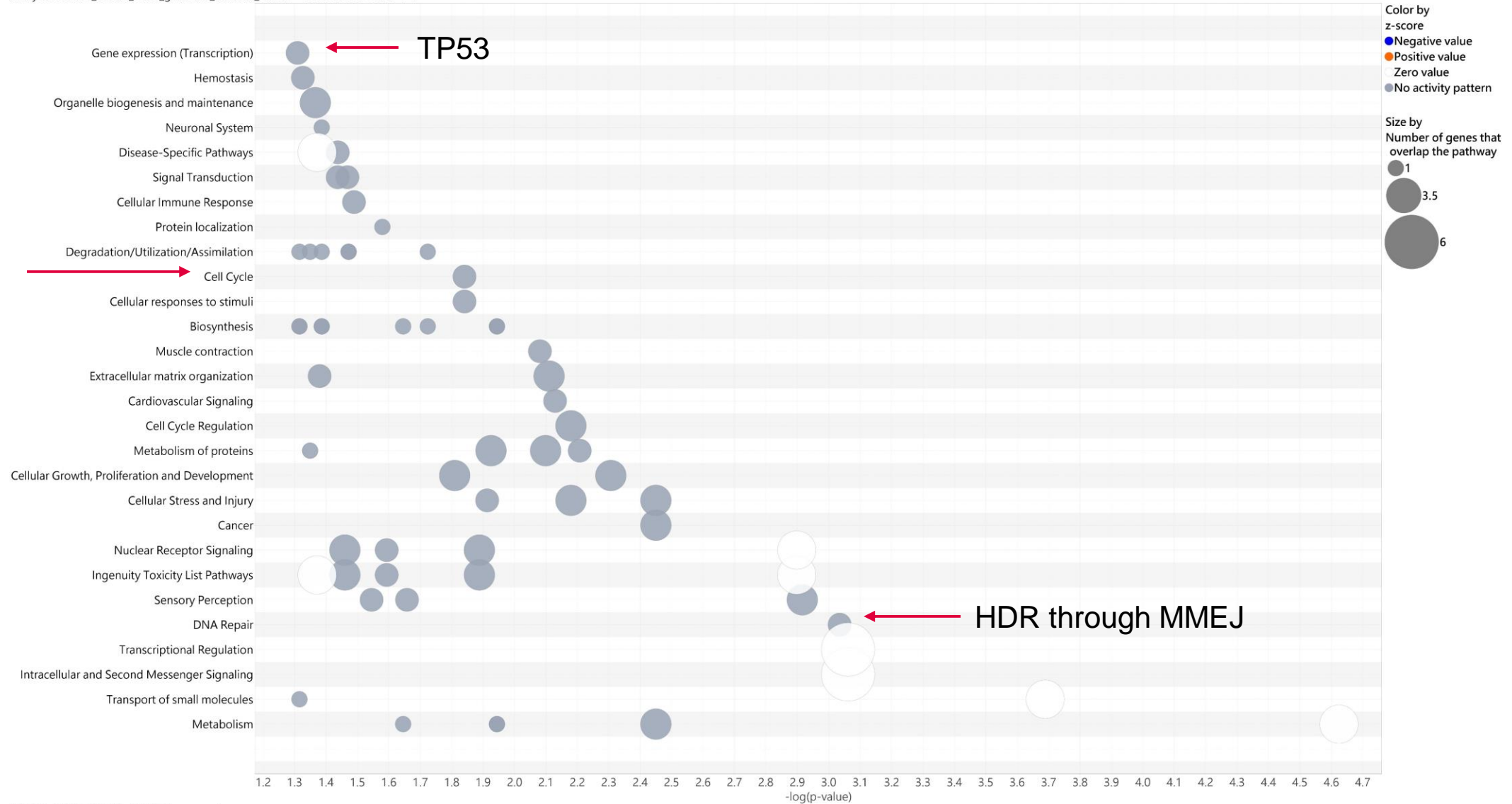
Name		p-value	Predicted Activation
PRKCE		2.54E-06	
GAB2		6.99E-06	
BCR (complex)		7.63E-06	
AGER		8.99E-06	
VEGFA		1.16E-05	

1 2 3 4 5 6 7 8 9 >

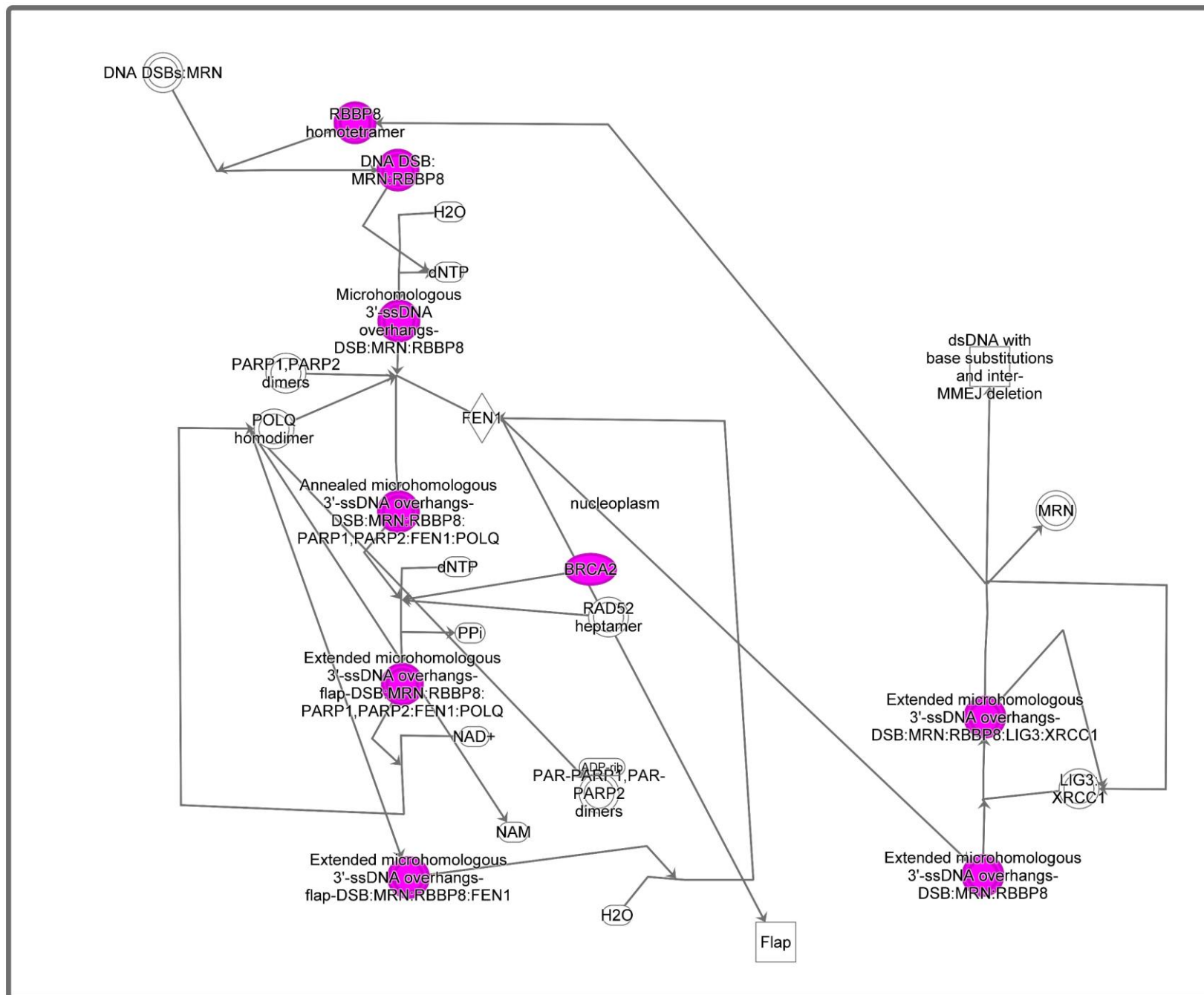
> Top Diseases and Bio Functions

This is a logarithmic scale showing values from 1 to 1E-10.

Analysis: Nature_comm_2020_germline_variants_ACMG - 2024-03-20 05:07 下午



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Variant Effects Analysis - Nature_comm_2020_germline_variants_ACMG - 2024-03-20 05:07 下午

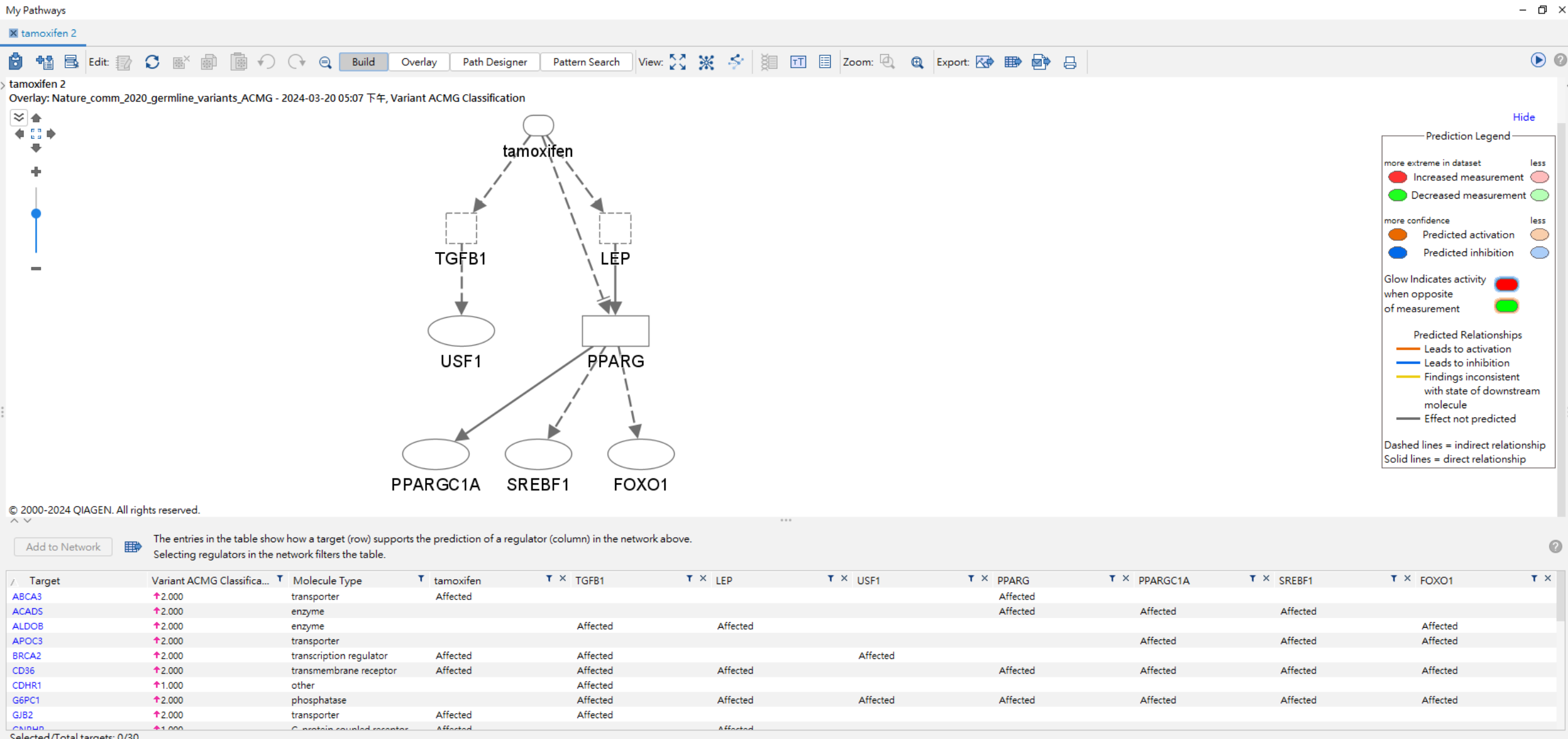
Summary Graphical Summary Pathways **Upstream Analysis** Diseases & Functions Regulator Effects Networks Lists Analysis Match Molecules

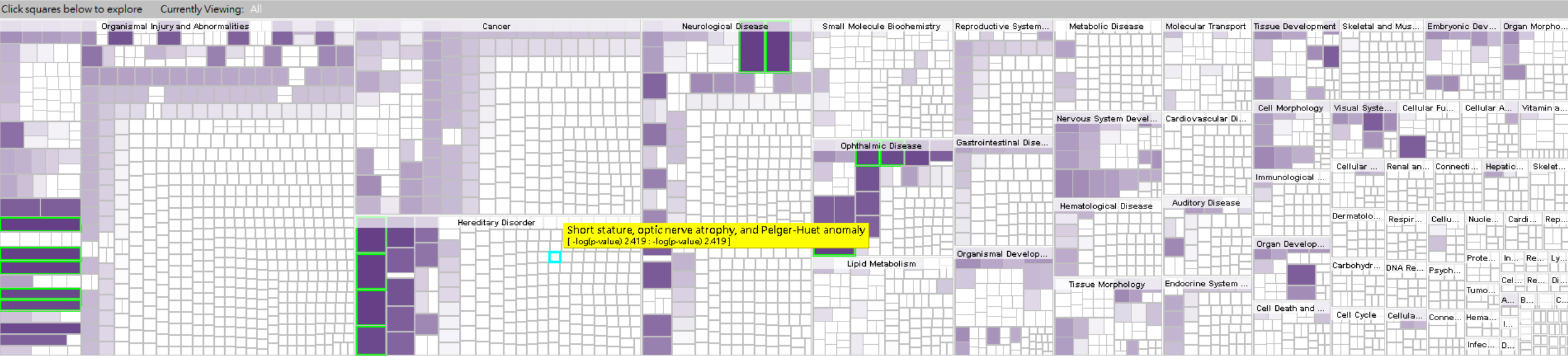
Upstream Regulators Causal Networks

Add To My Pathway Add To My List Display as Network Activity Plot Customize Table Mechanistic Networks p-value... 7.73E-08 - 6.70E-04 (1/22)

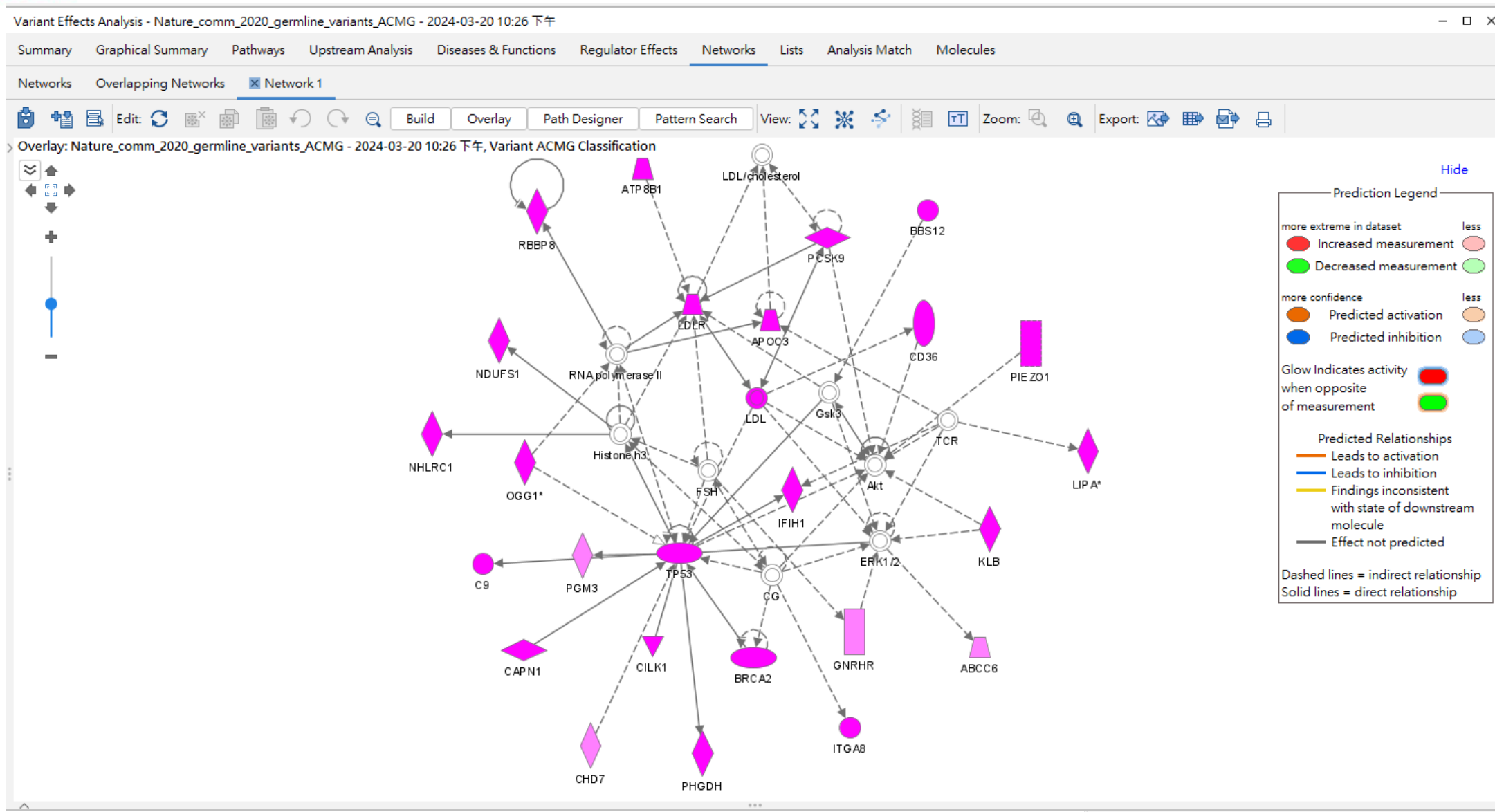
Upstream Regulator	Variant ACMG Classification	Molecule Type	p-value of overlap	Target Molecules in Dataset	Mechanistic Network
SREBF1		transcription regulator		ACADS, APOC3, CD36, G6PC1, ...all 10	36 (12)
MED13		transcription regulator		CD36, CIB2, G6PC1, ODAD2, ...all 5	
CTNNB1		transcription regulator		ABCA3, ACADS, AGRN, AQP5, ...all 16	
PPARGC1A		transcription regulator		ACADS, APOC3, CD36, G6PC1, ...all 11	26 (11)
pirinixic acid		chemical toxicant		ACADS, C9, CD36, G6PC1, ...all 10	37 (13)
fenofibrate		chemical drug		APOC3, CD36, G6PC1, LDLR, ...all 8	37 (14)
L-serine		chemical - endogenous mammalian		PCK2, PHGDH, TP53, ...all 3	
NR4A1		ligand-dependent nuclear receptor		ALDOB, CD36, G6PC1, GNRHR, ...all 8	27 (11)
NR1D1		ligand-dependent nuclear receptor		APOC3, G6PC1, KLB, TSHB, ...all 4	
LEP		growth factor		ALDOB, CD36, G6PC1, GNRHR, ...all 10	32 (14)
8-bromo-cAMP		chemical reagent		BRCA2, G6PC1, GNRHR, ITGA8, ...all 10	31 (11)
artepillin C		chemical reagent		G6PC1, LDLR, PCSK9, ...all 3	27 (7)
FBXW7		enzyme		CD36, IVD, LDLR, LIPA, TP53, ...all 5	28 (7)
MTDH		transcription regulator		ALDOB, C9, CD36, LDLR, PCSK9, ...all 6	27 (7)
NU 1025		chemical toxicant		BRCA2, TP53, ...all 2	
PRKAA		group		CD36, G6PC1, OGG1, TP53, ...all 4	35 (13)
tamoxifen		chemical drug	5.05E-05	ABCA3, BRCA2, CD36, GJB2, ...all 8	30 (8)
MAP4K4		kinase	6.62E-05	ACADS, IVD, NDUFS1, PHYH, ...all 5	
PPARA		ligand-dependent nuclear receptor	6.88E-05	ACADS, ALDOB, APOC3, C9, ...all 9	23 (11)
C1QL3		other	7.40E-05	CD36, G6PC1, ...all 2	17 (6)
cholesterol ester		chemical - endogenous mammalian	7.40E-05	CD36, LDLR, ...all 2	
Insulin		group	7.50E-05	ALDOB, APOC3, CD36, G6PC1, ...all 11	37 (14)
PPARG		ligand-dependent nuclear receptor	1.09E-04	ABCA3, ACADS, CD36, G6PC1, ...all 9	38 (13)
THRB		ligand-dependent nuclear receptor	1.18E-04	APOC3, G6PC1, LDLR, PHYH, ...all 7	31 (8)
OGA		enzyme	1.37E-04	CAPN1, CD36, CDKAL1, CILK1, ...all 8	
FGF21		growth factor	1.89E-04	G6PC1, KLB, LDLR, PHGDH, ...all 4	28 (7)
sterol		chemical - endogenous mammalian	1.98E-04	LDLR, LIPA, PCSK9, ...all 3	4 (3)
EIF2AK3		kinase	2.16E-04	CAPN1, COG8, PCK2, TP53, ...all 5	
vitamin A		chemical - endogenous mammalian	2.17E-04	LRAT, STRA6, TSHB, ...all 3	
olaparib		chemical drug	2.39E-04	BRCA2, IFIH1, RBBP8, TP53, ...all 4	
daunorubicin		chemical drug	2.82E-04	ABCA3, ABCC6, TP53, ...all 3	
cyclopropanecarboxylic acid		chemical reagent	3.06E-04	ACADS, G6PC1, TP53, ...all 3	
dexamethasone		chemical drug	3.16E-04	ABCC6, ACADS, ALDOB, AQP5, ...all 20	30 (10)
USF1		transcription regulator	3.37E-04	BRCA2, G6PC1, LDLR, TP53, ...all 4	
elaidic acid		chemical - endogenous mammalian	3.37E-04	LDLR, LIPA, PCSK9, PHGDH, ...all 4	

Selected 0 / 1090





Categories	Diseases or Functions Annotation	p-value	Predicted Activation State	Activation z-score	Molecules	# Molecules
Hereditary Disorder,Neurological Disease,Organis...	Familial neurological disorder	4.84E-21			↑AGRN, ↑ALG13, ↑BBS1, ↑BBS12, ↑BRC...all 53	53
Hereditary Disorder,Neurological Disease,Organis...	Autosomal recessive neurological disorder	9.95E-21			↑AGRN, ↑BBS1, ↑BBS12, ↑BRCA2, ↑CA...all 35	35
Hereditary Disorder,Ophthalmic Disease,Organis...	Hereditary Eye Disease	2.19E-20			↑AGBL5, ↑AIPL1, ↑BBS1, ↑BBS12, ↑BLO...all 28	28
Ophthalmic Disease,Organismal Injury and Abno...	Retinitis pigmentosa	3.76E-17			↑AGBL5, ↑AIPL1, ↑BBS1, ↑BBS12, ↑CD...all 16	16
Hereditary Disorder,Ophthalmic Disease,Organis...	Hereditary retinitis pigmentosa	5.38E-17			↑AGBL5, ↑AIPL1, ↑BBS1, ↑CDHR1, ↑CIB2,...all 15	15
Hereditary Disorder,Ophthalmic Disease,Organis...	Hereditary retinal degeneration	1.07E-16			↑AGBL5, ↑AIPL1, ↑BBS1, ↑BBS12, ↑C9,...all 18	18
Hereditary Disorder,Ophthalmic Disease,Organis...	Autosomal recessive retinitis pigmentosa	2.74E-15			↑AGBL5, ↑AIPL1, ↑CDHR1, ↑CIB2, ↑CLR...all 12	12
Developmental Disorder,Ophthalmic Disease,Org...	Developmental disorder of retina	2.61E-14			↑AGBL5, ↑AIPL1, ↑BBS1, ↑BBS12, ↑CD...all 15	15
Cellular Function and Maintenance,Organ Develo...	Maintenance of photoreceptors	8.90E-14			↑BBS1, ↑BBS12, ↑CDHR1, ↑CIB2, ↑CLN8,...all 9	9
Ophthalmic Disease,Organismal Injury and Abno...	Retinal degeneration	9.31E-14		0.000	↑AGBL5, ↑AIPL1, ↑BBS1, ↑BBS12, ↑C9,...all 19	19
Hereditary Disorder,Ophthalmic Disease,Organis...	Autosomal recessive retinal degeneration	1.04E-13			↑AGBL5, ↑AIPL1, ↑CDHR1, ↑CIB2, ↑CLR...all 13	13
Developmental Disorder,Hereditary Disorder,Oph...	Retinal dystrophy	1.35E-13			↑AGBL5, ↑AIPL1, ↑BBS1, ↑BBS12, ↑CD...all 14	14
Ophthalmic Disease,Organismal Injury and Abno...	Disease of retina	2.01E-13		0.000	↑ABCC6, ↑AGBL5, ↑AIPL1, ↑BBS1, ↑BBS...all 25	25
Hereditary Disorder,Neurological Disease,Organis...	Familial central nervous system disease	4.45E-13			↑ALG13, ↑BBS1, ↑BBS12, ↑BRCA2, ↑C9,...all 39	39



Summary Graphical Summary Pathways Upstream Analysis Diseases & Functions Regulator Effects Networks Lists Analysis Match Molecules							
<div> Add To My Pathway Add To My List Create Dataset Customize Table </div>							
Symbol	Entrez Gene Name	Identifier	Variant ACMG Classific...	Location	Type(s)	Biomarker Application(s)	Drug(s)
ABCA3*	ATP binding cassette subfamily ...	ABCA3*	Pathogenic	Plasma Membrane	transporter		
ABCC6	ATP binding cassette subfamily ...	ABCC6	Likely Pathogenic	Plasma Membrane	transporter		
ACADS*	acyl-CoA dehydrogenase short ...	ACADS*	Pathogenic	Cytoplasm	enzyme	unspecified application	
ACSF3	acyl-CoA synthetase family me...	ACSF3	Likely Pathogenic	Cytoplasm	enzyme		
AGBL5	AGBL carboxypeptidase 5	AGBL5	Pathogenic	Cytoplasm	peptidase		
AGRN	agrin	AGRN	Likely Pathogenic	Plasma Membrane	other		
AIPL1	aryl hydrocarbon receptor inter...	AIPL1	Pathogenic	Nucleus	other		
ALDOB	aldolase, fructose-bisphosphate...	ALDOB	Pathogenic	Cytoplasm	enzyme		
ALG13	ALG13 UDP-N-acetylglucosami...	ALG13	Likely Pathogenic	Cytoplasm	enzyme		
APOC3	apolipoprotein C3	APOC3	Pathogenic	Extracellular Space	transporter	diagnosis, efficacy	volanesorsen
AQP5	aquaporin 5	AQP5	Pathogenic	Plasma Membrane	transporter		
ATP8B1	ATPase phospholipid transporti...	ATP8B1	Pathogenic	Plasma Membrane	transporter		
BBS1	Bardet-Biedl syndrome 1	BBS1	Pathogenic	Extracellular Space	other		
BBS12	Bardet-Biedl syndrome 12	BBS12	Pathogenic	Extracellular Space	other		
BLOC1S3	biogenesis of lysosomal organe...	BLOC1S3	Likely Pathogenic	Cytoplasm	transporter	diagnosis	
BRCA2	BRCA2 DNA repair associated	BRCA2	Pathogenic	Nucleus	transcription regulator	diagnosis, efficacy, unspecified application	
C9	complement C9	C9	Pathogenic	Extracellular Space	other		
CAPN1	calpain 1	CAPN1	Pathogenic	Cytoplasm	peptidase		BLD-2660, alicapostat
CD36	CD36 molecule	CD36	Pathogenic	Plasma Membrane	transmembrane receptor	efficacy, unspecified application	VT1021, zeaxanthin
CD96	CD96 molecule	CD96	Pathogenic	Plasma Membrane	other		GSK6097608
CDHR1	cadherin related family member 1	CDHR1	Likely Pathogenic	Plasma Membrane	other		
CDKAL1	CDK5 regulatory subunit associ...	CDKAL1	Pathogenic	Cytoplasm	enzyme		
CHD7	chromodomain helicase DNA bi...	CHD7	Likely Pathogenic	Nucleus	enzyme		
CIB2	calcium and integrin binding fa...	CIB2	Pathogenic	Cytoplasm	kinase		
CILK1	ciliogenesis associated kinase 1	ICK	Pathogenic	Cytoplasm	kinase		
CLCN1	chloride voltage-gated channel 1	CLCN1	Pathogenic	Plasma Membrane	ion channel		
CLN8	CLN8 transmembrane ER and E...	CLN8	Likely Pathogenic	Cytoplasm	other		
CLPB	ClpB family mitochondrial disag...	CLPB	Likely Pathogenic	Nucleus	transcription regulator		
CLRN1	clarin 1	CLRN1	Pathogenic	Plasma Membrane	other		
CNGB1	cyclic nucleotide gated channel ...	CNGB1	Likely Pathogenic	Plasma Membrane	ion channel		
COG8	component of oligomeric golgi ...	COG8	Pathogenic	Cytoplasm	transporter		
DNAAF4	dynein axonemal assembly fact...	DYX1C1	Likely Pathogenic	Nucleus	other		
DOCK7	dedicator of cytokinesis 7	DOCK7	Likely Pathogenic	Plasma Membrane	other		
ELMOD3	ELMO domain containing 3	ELMOD3	Pathogenic	Other	other		

Selected/Total molecules: 0 / 92



Draw customer pathway



Create New...

Search

Advanced Search



— □ ×

Annotated Dataset: myList

Preview Dataset myList

Mapped IDs (16) Unmapped IDs (0) All IDs (16) Metadata

Add To My Pathway

New My Pathway

Create Dataset

Customize Table



ID	Symbol	Entrez Gene Name	Location	Type(s)	Drug(s)
ERGIC1	ERGIC1	endoplasmic reticulum-golgi intermediate c...	Cytoplasm	other	
GNA12	GNA12	G protein subunit alpha 12	Plasma Membrane	enzyme	
HLA-DRB1	HLA-DRB1	major histocompatibility complex, class II, D...	Plasma Membrane	transmembrane receptor	apolizumab
LINC01271	LINC01271		Other	other	
LOC105373831	LOC105373831	uncharacterized LOC105373831	Other	other	
PDGFB	PDGFB	platelet derived growth factor subunit B	Extracellular Space	growth factor	dasatinib, sunitinib, pegpleranib, imatinib/...
PITX1-AS1	PITX1-AS1	PITX1 antisense RNA 1	Other	other	
PLCL1	PLCL1	phospholipase C like 1 (inactive)	Cytoplasm	enzyme	quinacrine
PNKD	PNKD	PNKD metallo-beta-lactamase domain conta...	Nucleus	other	
PRDM1	PRDM1	PR/SET domain 1	Nucleus	transcription regulator	
RPS21P8	RPS21P8	ribosomal protein S21 pseudogene 8	Other	other	
SFMBT1	SFMBT1	Scm like with four mbt domains 1	Nucleus	transcription regulator	
SMAD3	SMAD3	SMAD family member 3	Nucleus	transcription regulator	
SMAD7	SMAD7	SMAD family member 7	Nucleus	transcription regulator	
TET2	TET2	tet methylcytosine dioxygenase 2	Nucleus	enzyme	
ZBTB40	ZBTB40	zinc finger and BTB domain containing 40	Nucleus	transcription regulator	

16 / 16

Flags:

"D" - Duplicates. Gene/Protein/Chemical identifiers marked with an asterisk indicate that multiple identifiers in the dataset file map to a single gene/chemical in the Global Molecular Network.

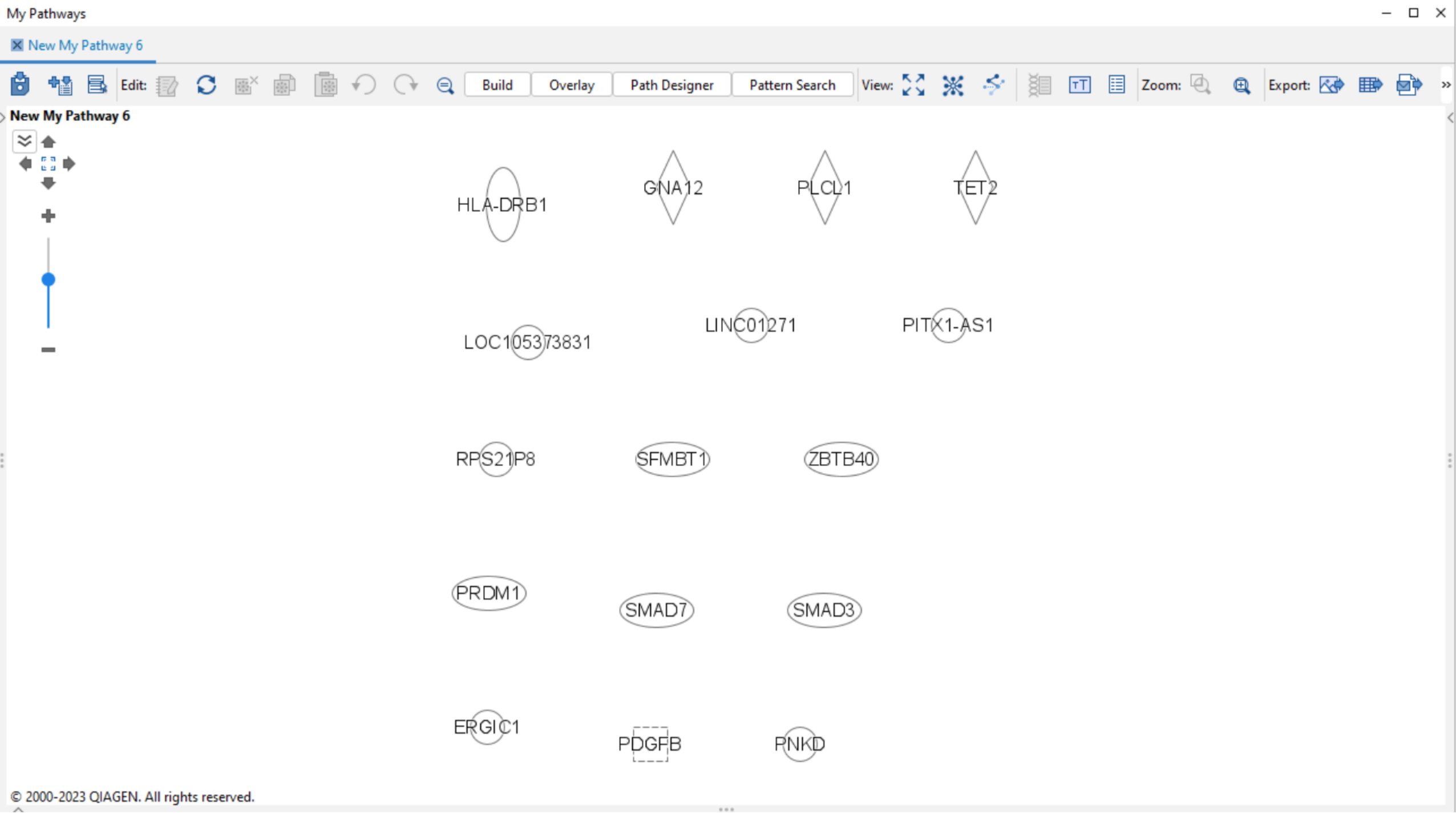
"O" - Override molecules. Gene/Protein/Chemical identifiers marked as "Override" are displayed with italic text.

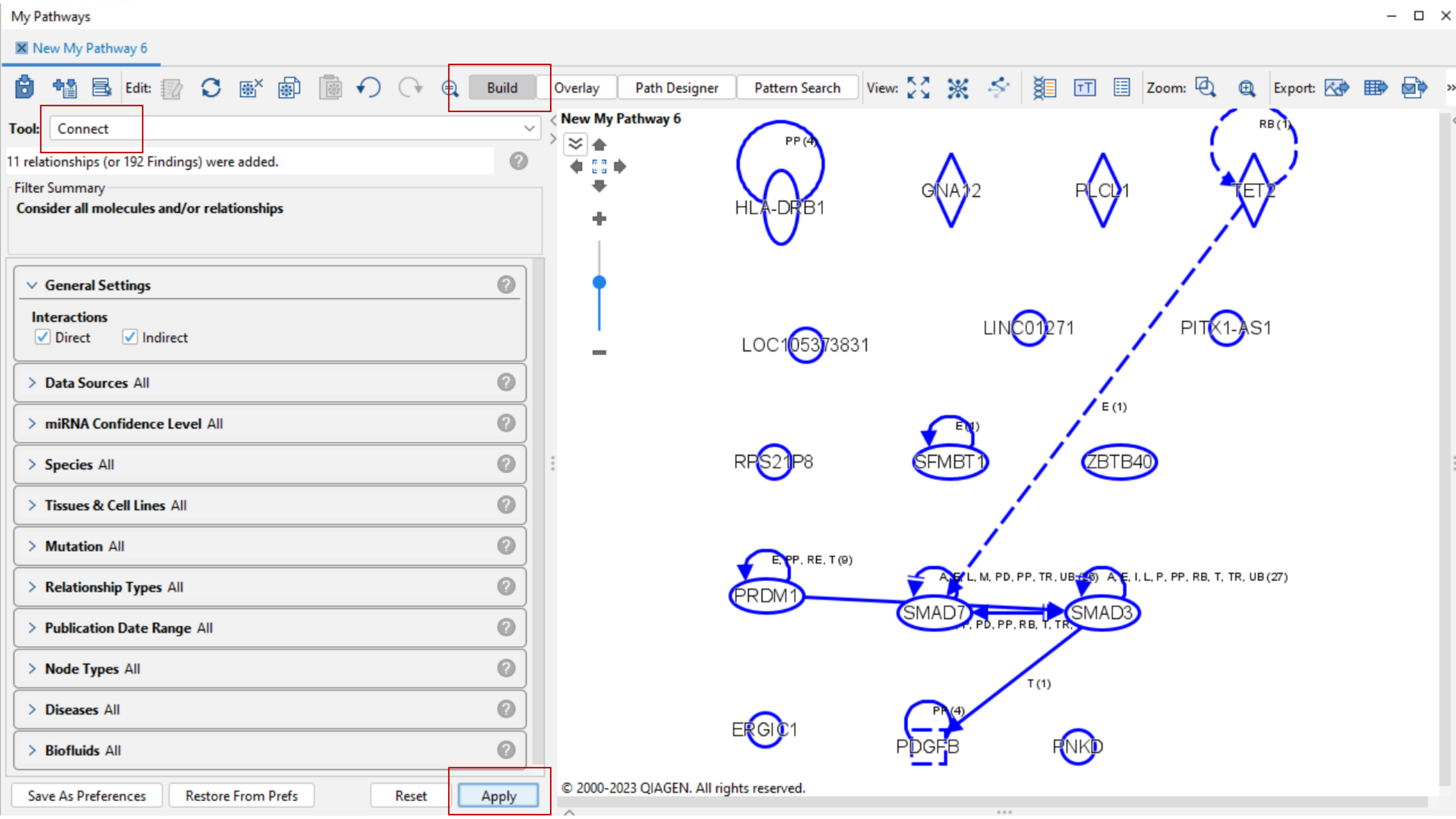
"A" - Gene/Protein/Chemical ID marked as Absent. The gene/protein/chemical will not be used as a focus molecule or appear in networks unless you also explicitly override this flag with the Override column.

Edit Dataset Settings

Analyze/Filter Dataset

Close







Genes and Chemicals Diseases and Functions Pathways and Lists Datasets and Analyses

Create New...

activation of T lymphocytes

Search

Advanced Search



Project Manager

- My Projects
 - GWAS
 - Dataset Files
 - myList
 - Ulcerative Colitis-associations2
 - colon cancer-association
 - Ulcerative Colitis-associations
 - Analyses
 - Ulcerative Colitis-associations -
 - colon cancer-association - 2023
 - Ulcerative Colitis-associations -
 - Comparison Analyses
 - Biomarker Filter Results
 - Biomarker Comparison Analyses
 - MicroRNA Target Filter Results
 - BioProfiler Results
 - IsoProfiler Results
 - My Pathways
 - My Lists
 - Human Genes Chromosomal Location
 - QIAGEN Coronavirus Networks
 - Innenuity KEGG gene lists

Search Results

Genes and Chemicals Diseases and Functions

Add To My Pathway

New My Pathway

Saved My Pathway

New My Pathway 6

The search for activation of

Diseases & Functions

Matching Diseases & Functions

Hematological System Development and Function

activation

Activation of lymphocytes

Activation of T lymphocytes

Activation of natural killer cells [activation of NK Lymphocyte, activation of NK cells,...]

Activation of B lymphocytes [activation of B-enriched lymphocytes, activation of primary B lymphocytes]

Activation of CD4+ T-lymphocytes [activation of CD4-positive T-lymphocytes, activation of CD4+ lymphocytes,...]

Activation of CD8+ T lymphocyte [activation of CD8-positive T-lymphocytes, activation of CD8+ lymphocytes,...]

Activation of helper T lymphocytes [activation of helper inducer T lymphocytes, activation of helper T lymphocytes,...]

Activation of cytotoxic T cells [activation of cytotoxic T lymphocytes, activation of cytotoxic T cells,...]

Activation of natural killer T lymphocytes

Activation of regulatory T lymphocytes

Activation of memory T lymphocytes

Activation of Th1 cells [activation of Th1 lymphocytes, activation of T helper type 1 lymphocytes,...]

Activation of naive lymphocytes

Activation of effector lymphocytes

Activation of naive T lymphocytes

Activation of alpha-beta T lymphocytes [activation of α - β T lymphocytes]

Activation of mature T lymphocytes

Activation of effector T lymphocytes

Activation of peripheral blood lymphocytes

Activation of Th2 cells [activation of Th2 lymphocytes, activation of th2 effector cells,...]

Hematological System Development and Function

Select source

Add

☒ Diseases or Functions

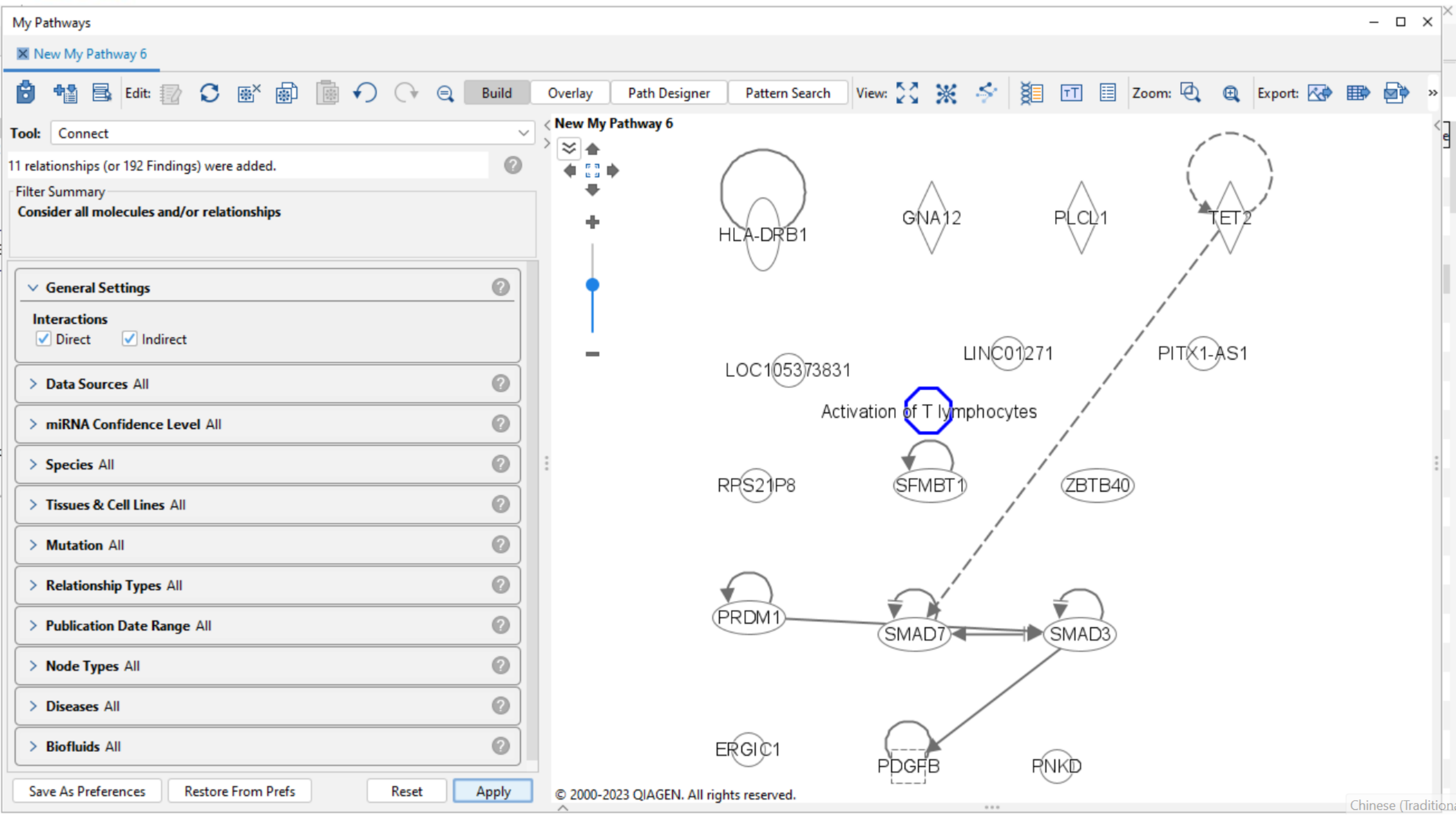
☐ Molecules

☐ Both

OK Cancel

Associated Molecule

1421
1372
1137
1134
877
262
254
163
130
88
87
59
58
49
40
35
34
32
29
28
27
26
24



Consider all molecules and/or relationships

General Settings

Interactions

☒ Direct ☒ Indirect

Set A

AddRemove

ERGIC1
GNA12
HLA-DRB1

Direction:

From Set A to Set B

>

Set B

AddRemove

Activation of T lymphocytes

☒ Use Ingenuity Knowledge Base

☐ Use Molecules from Analysis/Dataset/List...

Current Analysis/Dataset/List: None selected

Change Analysis/Dataset/List

> Data Sources All

☒ Direct ☒ Indirect

Add

Remove

ERGIC1
GNA12
HLA-DRB1

Direction: From Set A to Set B

Add

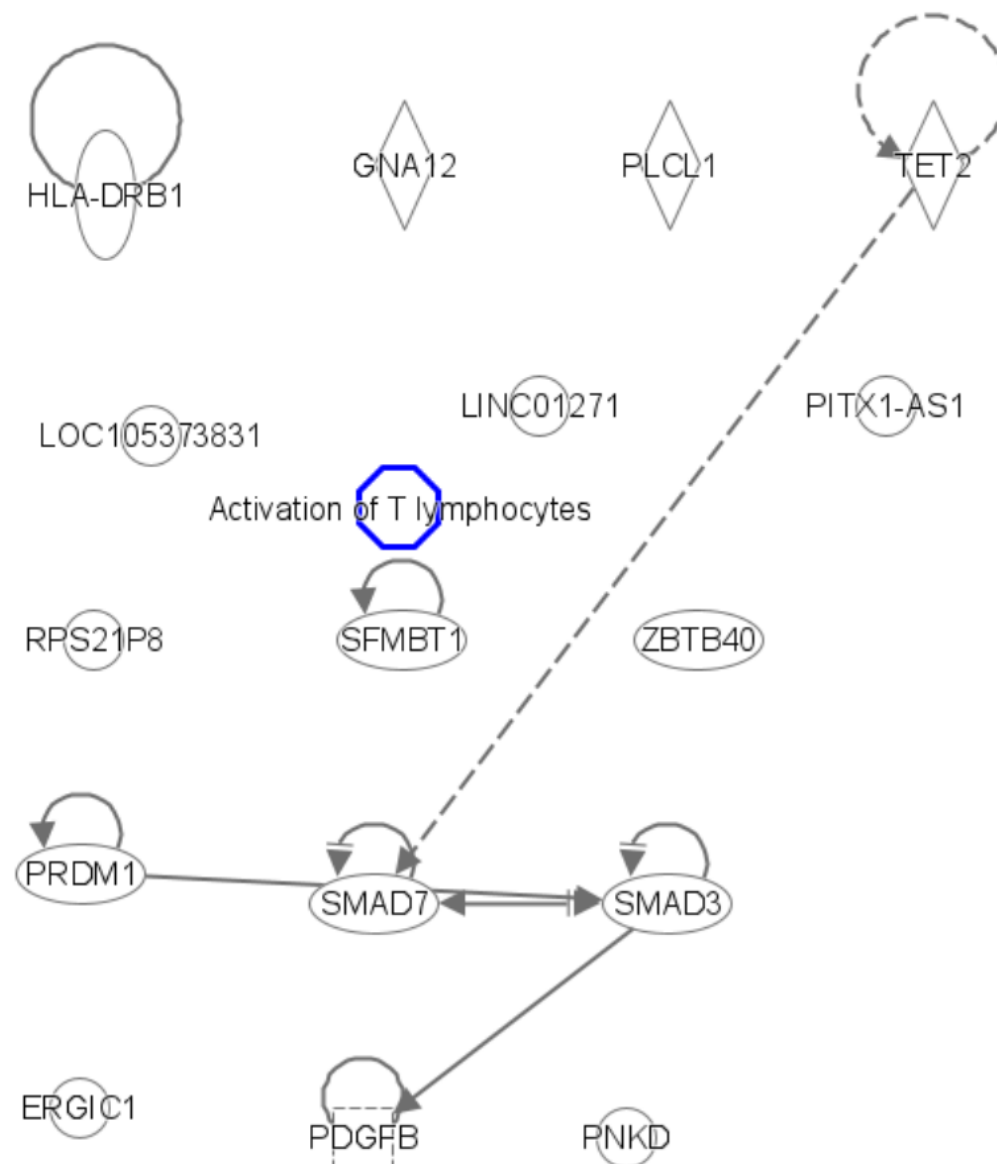
Remove

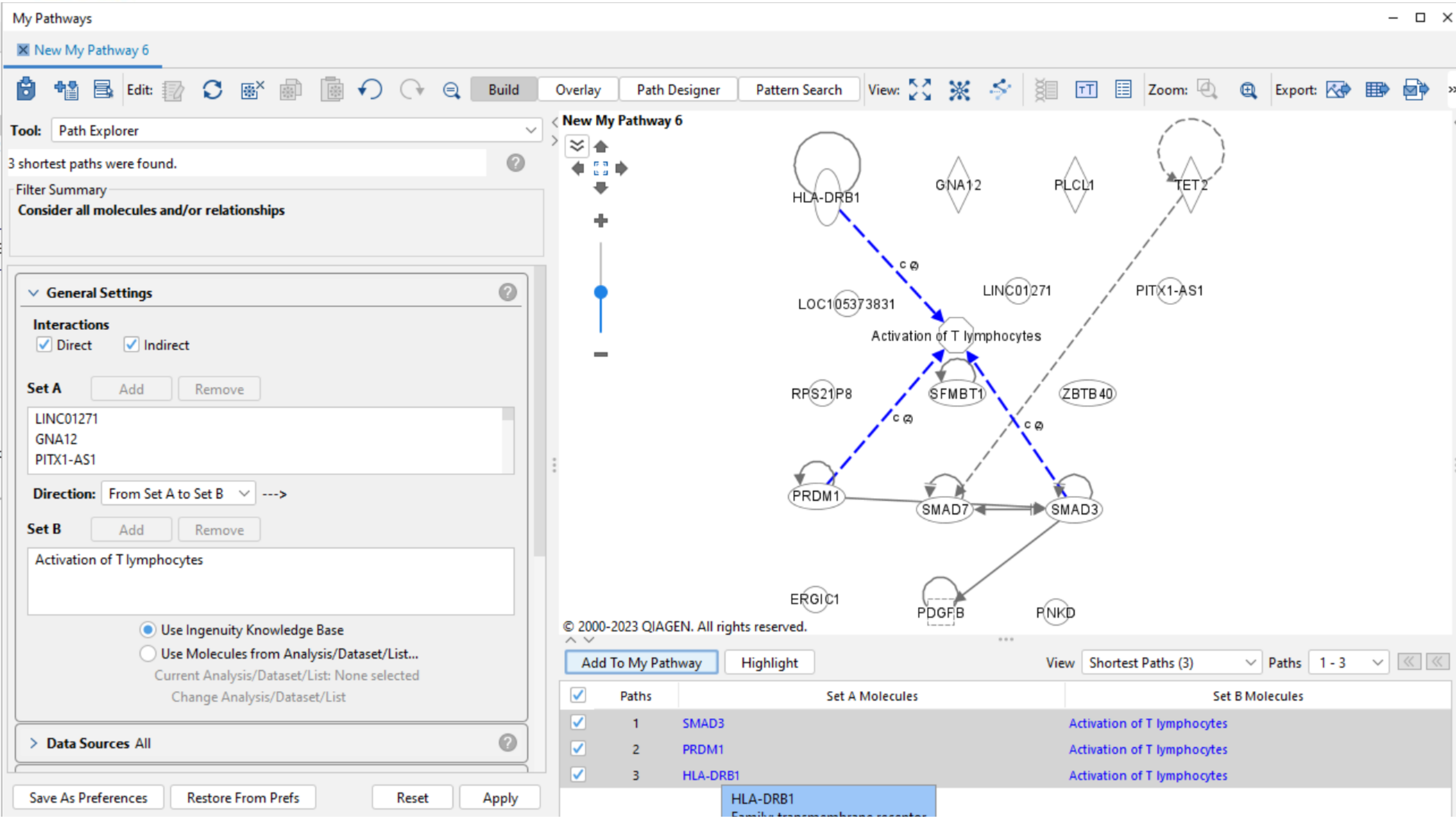
Activation of T lymphocytes

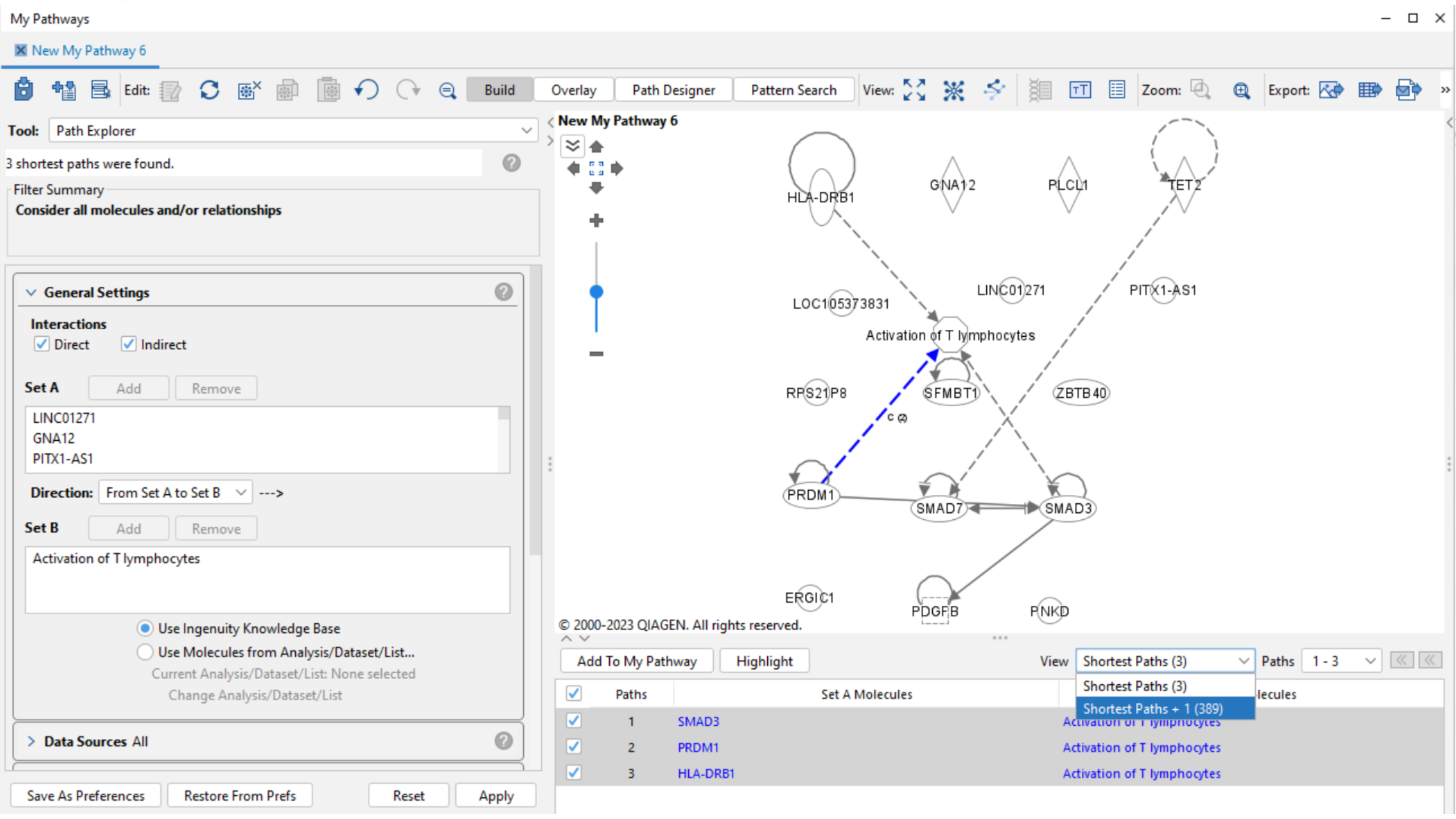
☒ Use Ingenuity Knowledge Base
☐ Use Molecules from Analysis/Dataset/List...
 Current Analysis/Dataset/List: None selected
 Change Analysis/Dataset/List

> Data Sources All

Apply







Tool: Path Explorer

389 shortest paths were found.

Filter Summary

Consider all molecules and/or relationships

General Settings

Interactions

☒ Direct ☒ Indirect

Set A

Add

Remove

LINC01271

GNA12

PITX1-AS1

Direction: From Set A to Set B

Set B

Add

Remove

Activation of T lymphocytes

☒ Use Ingenuity Knowledge Base☐ Use Molecules from Analysis/Dataset/List...

Current Analysis/Dataset/List: None selected

Change Analysis/Dataset/List

> Data Sources All

> miRNA Confidence Level All

> Species All

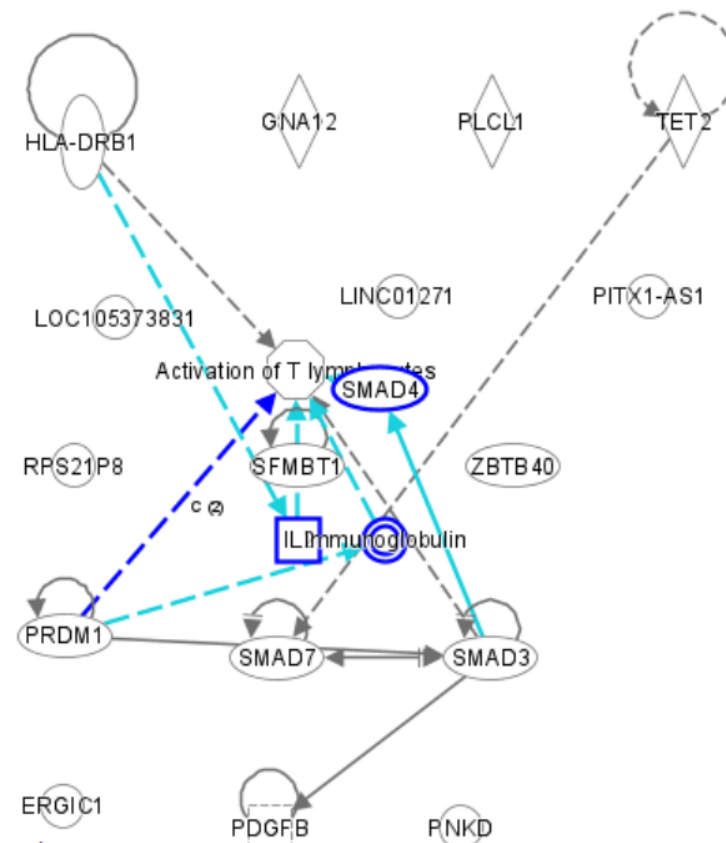
Save As Preferences

Restore From Prefs

Reset

Apply

New My Pathway 6



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Add To My Pathway

Highlight

View Shortest Paths + 1 (389)

Paths 1 - 50

	Paths	Set A Molecules	Node 1	Set B Molecules
<input checked="" type="checkbox"/>	1	SMAD3	SMAD4	Activation of T lymphocytes
<input type="checkbox"/>	2	SMAD3	IL2	Activation of T lymphocytes
<input type="checkbox"/>	3	PRDM1	IL2	Activation of T lymphocytes
<input checked="" type="checkbox"/>	4	HLA-DRB1	IL2	Activation of T lymphocytes
<input type="checkbox"/>	5	HLA-DRB1	TCR	Activation of T lymphocytes
<input checked="" type="checkbox"/>	6	PRDM1	Immunoglobulin	Activation of T lymphocytes

Tool: Keep

8 nodes and 15 relationships were kept

Filter Summary

**Keep all molecules where
connectedness > 1**

> Data Sources

> miRNA Confidence Level

> Species

> Tissues & Cell Lines

> Mutation

> Relationship Types

> Publication Date Range

> Node Types

> Diseases

> Biofluids

> Biomarkers

> Node Fill Overlay

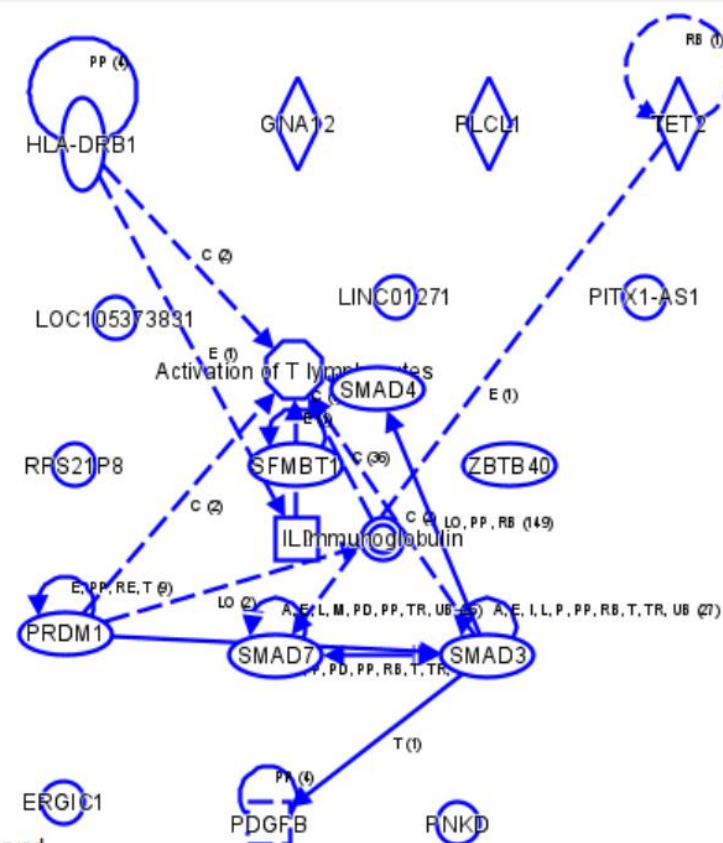
▼ Node Connectivity

Nodes connected to > 1 other nodes

Reset

Apply

New My Pathway 6



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Add To My Pathway

Highlight

View Shortest Paths + 1 (389)

Paths 1 - 50

<input type="checkbox"/>	Paths	Set A Molecules	Node 1	Set B Molecules
<input checked="" type="checkbox"/>	1	SMAD3	SMAD4	Activation of T lymphocytes
<input type="checkbox"/>	2	SMAD3	IL2	Activation of T lymphocytes
<input type="checkbox"/>	3	PRDM1	IL2	Activation of T lymphocytes
<input checked="" type="checkbox"/>	4	HLA-DRB1	IL2	Activation of T lymphocytes
<input type="checkbox"/>	5	HLA-DRB1	TCR	Activation of T lymphocytes
<input checked="" type="checkbox"/>	6	PRDM1	Immunoglobulin	Activation of T lymphocytes

Tool: Keep

8 nodes and 15 relationships were kept

Filter Summary

Keep all molecules where
connectedness > 1

> Data Sources

> miRNA Confidence Level

> Species

> Tissues & Cell Lines

> Mutation

> Relationship Types

> Publication Date Range

> Node Types

> Diseases

> Biofluids

> Biomarkers

> Node Fill Overlay

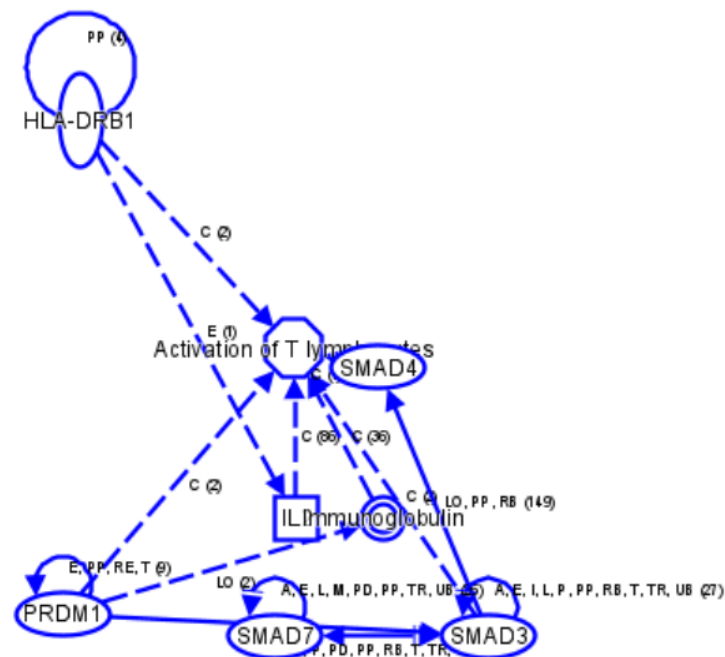
v Node Connectivity

Nodes connected to > 1 other nodes

Reset

Apply

New My Pathway 6



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Add To My Pathway

Highlight

View Shortest Paths + 1 (389)

Paths 1 - 50

<input type="checkbox"/>	Paths	Set A Molecules	Node 1	Set B Molecules
<input checked="" type="checkbox"/>	1	SMAD3	SMAD4	Activation of T lymphocytes
<input type="checkbox"/>	2	SMAD3	IL2	Activation of T lymphocytes
<input type="checkbox"/>	3	PRDM1	IL2	Activation of T lymphocytes
<input checked="" type="checkbox"/>	4	HLA-DRB1	IL2	Activation of T lymphocytes
<input type="checkbox"/>	5	HLA-DRB1	TCR	Activation of T lymphocytes
<input checked="" type="checkbox"/>	6	PRDM1	Immunoglobulin	Activation of T lymphocytes

Tool: Grow

Molecules & Canonical Pathways

Diseases & Functions

Grow from selected molecules to selected diseases & functions

Indicate diseases or functions related to Any of the selected molecules

Consider all functions

Recalculate

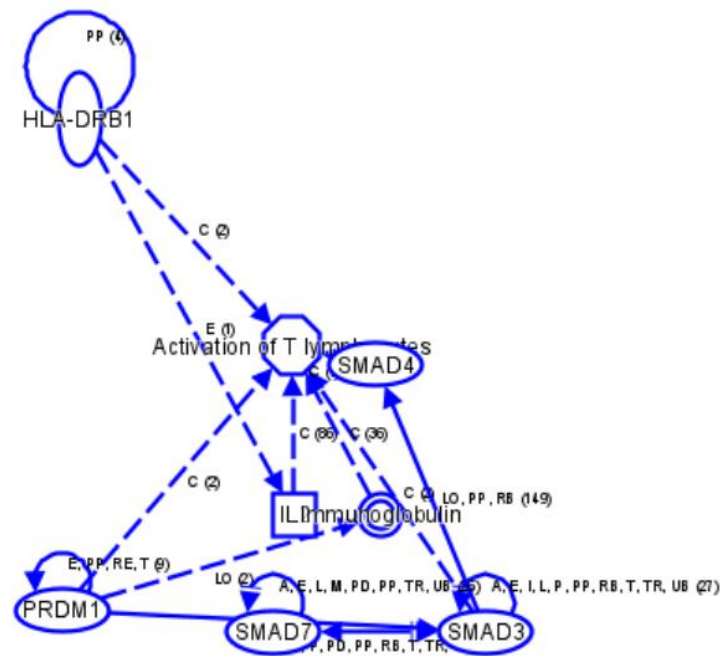
Diseases and Functions	p-value	Molecules
Induction of actin stress fiber	3.86E-10	SMAD7, SMAD3, SMAD4 ...all 3
Proliferation of activated T lymphocytes	1.50E-09	IL2, HLA-DRB1, SMAD3 ...all 5
Expansion of cancer stem cells	1.70E-09	SMAD7, SMAD3, SMAD4 ...all 3
Contractility of fibroblasts	1.70E-09	SMAD7, SMAD3, SMAD4 ...all 3
Differentiation of regulatory T lymphocytes	1.86E-09	IL2, SMAD7, HLA-DRB1 ...all 4
Cytostasis of epithelial cells	2.33E-09	SMAD7, SMAD3, SMAD4 ...all 3
Activation of CD4+ T-lymphocytes	2.67E-09	IL2, HLA-DRB1, PRDM1 ...all 4
Formation of abscess	3.55E-09	IL2, SMAD3, SMAD4 ...all 3
Proliferation of lymphocytes	4.66E-09	IL2, SMAD7, HLA-DRB1 ...all 6
Formation of skin	4.87E-09	SMAD7, HLA-DRB1, SMAD4 ...all 5
Quantity of mononuclear leukocytes	5.62E-09	IL2, SMAD7, HLA-DRB1 ...all 6
Differentiation of T lymphocytes	1.30E-08	IL2, SMAD7, HLA-DRB1 ...all 5
Activation of T lymphocytes	1.71E-08	IL2, HLA-DRB1, SMAD3 ...all 5
Transcription of DNA	3.61E-08	IL2, SMAD7, HLA-DRB1 ...all 6
Differentiation of memory T lymphocytes	4.11E-08	IL2, HLA-DRB1, PRDM1 ...all 3
Cell viability of lymphocytes	5.19E-08	IL2, SMAD3, PRDM1, ...all 4
Transactivation	5.58E-08	IL2, SMAD7, SMAD3, ...all 5
Proliferation of tumor cells	1.14E-07	IL2, SMAD7, SMAD3, ...all 5
Quantity of T lymphocytes	1.24E-07	IL2, SMAD7, HLA-DRB1 ...all 5

0/355

Reset

Apply

< New My Pathway 6



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[Add To My Pathway](#)

Highlight

View Shortest Paths + 1 (389)

Paths	1 - 50
-------	--------

<input type="checkbox"/>	Paths	Set A Molecules	Node 1	Set B Molecules
<input checked="" type="checkbox"/>	1	SMAD3	SMAD4	Activation of T lymphocytes
<input type="checkbox"/>	2	SMAD3	IL2	Activation of T lymphocytes
<input type="checkbox"/>	3	PRDM1	IL2	Activation of T lymphocytes
<input checked="" type="checkbox"/>	4	HLA-DRB1	IL2	Activation of T lymphocytes
<input type="checkbox"/>	5	HLA-DRB1	TCR	Activation of T lymphocytes
<input checked="" type="checkbox"/>	6	PRDM1	Immunoglobulin	Activation of T lymphocytes



Tool: Grow

Molecules & Canonical Pathways Diseases & Functions

Grow from selected molecules to selected diseases & functions

Indicate diseases or functions related to Any of the selected molecules

Consider all functions

Recalculate

Diseases and Functions	p-value	Molecules
Induction of actin stress		AD3, SM...all 3
Proliferation of activated		B1, SMAD3...all 5
Expansion of cancer stem		AD3, SM...all 3
Contractility of fibroblast		AD3, SM...all 3
Differentiation of regulat		HLA-DRB1...all 4
Cytostasis of epithelial ce		AD3, SM...all 3
Activation of CD4+ T-lympho	2.07E-05	IL2, HLA-DRB1, PRDM1...all 4
Formation of abscess	3.55E-09	IL2, SMAD3, SMAD4...all 3
Proliferation of lymphocytes	4.66E-09	IL2, SMAD7, HLA-DRB1...all 6
Formation of skin	4.87E-09	SMAD7, HLA-DRB1, S...all 5
Quantity of mononuclear leu	5.62E-09	IL2, SMAD7, HLA-DRB1...all 6
Differentiation of T lymphoc	1.30E-08	IL2, SMAD7, HLA-DRB1...all 5
Activation of T lymphocytes	1.71E-08	IL2, HLA-DRB1, SMAD3...all 5
Transcription of DNA	3.61E-08	IL2, SMAD7, HLA-DRB1...all 6
Differentiation of memory T	4.11E-08	IL2, HLA-DRB1, PRDM1...all 3
Cell viability of lymphocytes	5.19E-08	IL2, SMAD3, PRDM1, ...all 4
Transactivation	5.58E-08	IL2, SMAD7, SMAD3, ...all 5
Proliferation of tumor cells	1.14E-07	IL2, SMAD7, SMAD3, ...all 5
Quantity of T lymphocytes	1.24E-07	IL2, SMAD7, HLA-DRB1...all 5

Apply

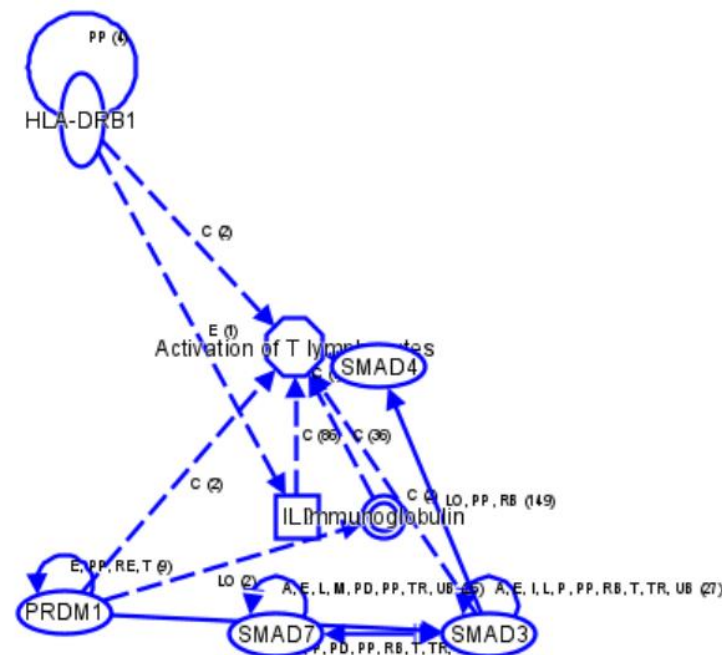
Cancel

0/355

Reset

Apply

New My Pathway 6



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Add To My Pathway

Highlight

View Shortest Paths + 1 (389)

Paths 1 - 50

<input type="checkbox"/>	Paths	Set A Molecules	Node 1	Set B Molecules
<input checked="" type="checkbox"/>	1	SMAD3	SMAD4	Activation of T lymphocytes
<input type="checkbox"/>	2	SMAD3	IL2	Activation of T lymphocytes
<input type="checkbox"/>	3	PRDM1	IL2	Activation of T lymphocytes
<input checked="" type="checkbox"/>	4	HLA-DRB1	IL2	Activation of T lymphocytes
<input type="checkbox"/>	5	HLA-DRB1	TCR	Activation of T lymphocytes
<input checked="" type="checkbox"/>	6	PRDM1	Immunoglobulin	Activation of T lymphocytes

Tool: Grow

Molecules & Canonical Pathways Diseases & Functions

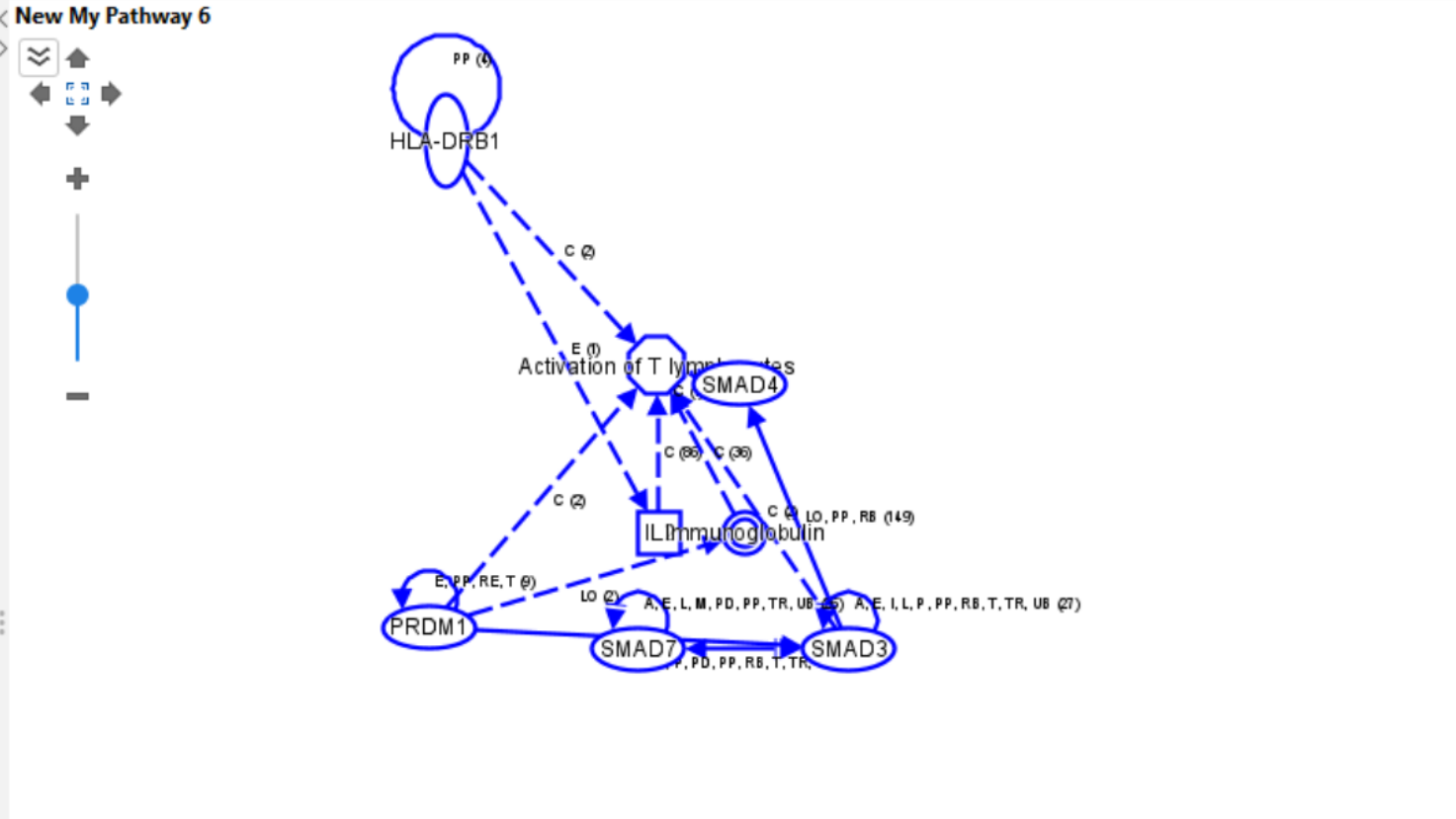
Grow from selected molecules to selected diseases & functions

Indicate diseases or functions related to Any of the selected molecules

Consider only functions with names like proliferation*

Recalculate

Diseases and Functions	p-value	Molecules
Proliferation of activated T lyr	1.50E-09	IL2, HLA-DRB1, SMAD3, ...all 5
Proliferation of lymphocytes	4.66E-09	IL2, SMAD7, HLA-DRB1, ...all 6
Proliferation of tumor cells	1.14E-07	IL2, SMAD7, SMAD3, ...all 5
Proliferation of beta islet cell:	3.57E-07	IL2, SMAD7, SMAD3 ...all 3
Proliferation of B lymphocyte:	4.62E-07	IL2, SMAD7, SMAD3, ...all 4
Proliferation of cancer cells	3.63E-06	IL2, SMAD7, SMAD3, ...all 4
Proliferation of hepatocytes	7.03E-06	SMAD7, SMAD3, SMA...all 3
Proliferation of Th1 cells	1.94E-05	IL2, HLA-DRB1 ...all 2
Proliferation of T lymphocyte:	2.47E-05	IL2, HLA-DRB1, SMAD4 ...all 3
Proliferation of hematopoieti	2.98E-05	IL2, PRDM1, SMAD4 ...all 3
Proliferation of eye cell lines	4.19E-05	SMAD3, SMAD4 ...all 2
Proliferation of splenocytes	1.25E-04	IL2, SMAD3 ...all 2
Proliferation of lymphokine a	1.47E-04	IL2 ...all 1
Proliferation of hepatic stella	1.57E-02	SMAD3 ...all 1

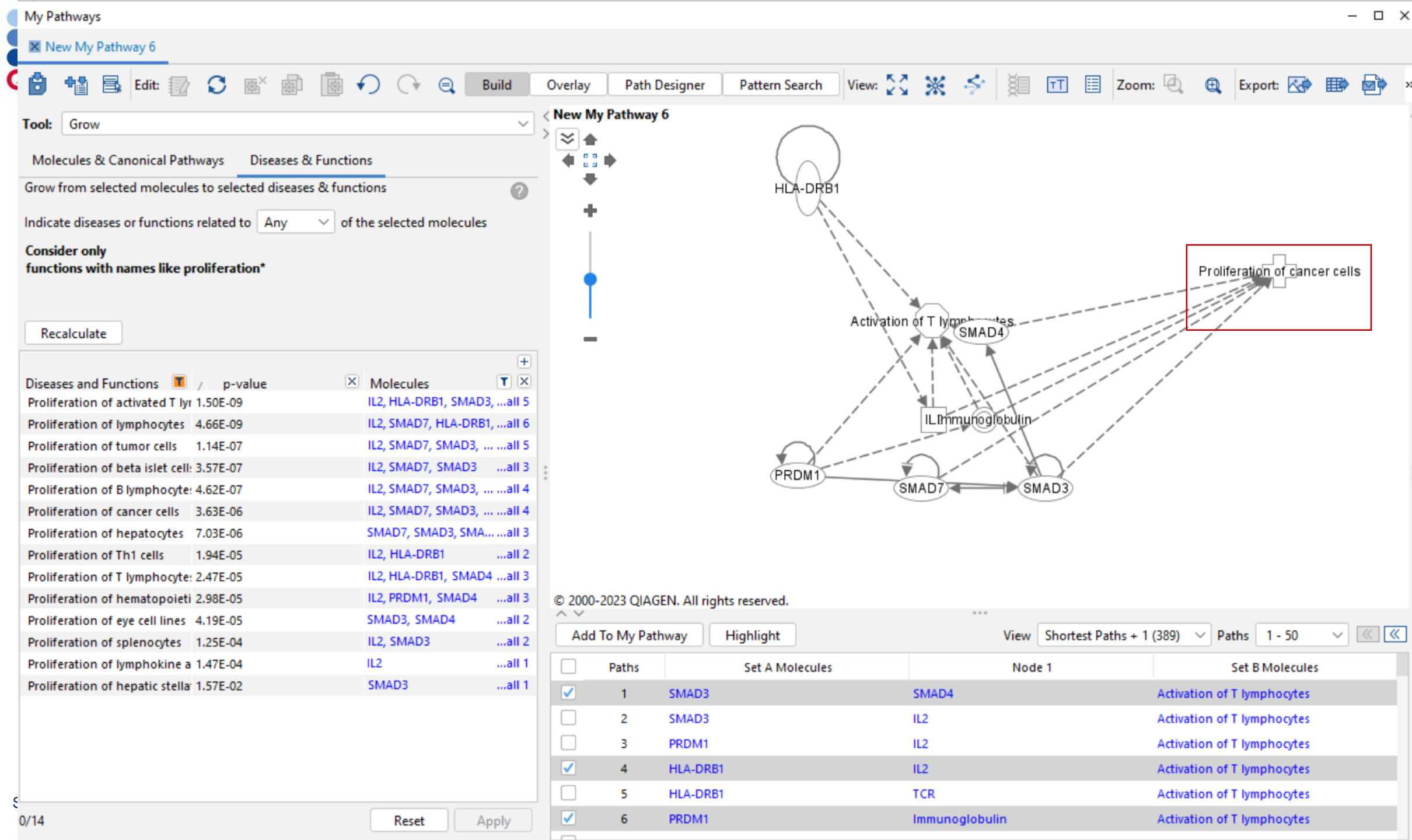


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Add To My Pathway Highlight

View Shortest Paths + 1 (389) Paths 1 - 50

	Paths	Set A Molecules	Node 1	Set B Molecules
<input checked="" type="checkbox"/>	1	SMAD3	SMAD4	Activation of T lymphocytes
<input type="checkbox"/>	2	SMAD3	IL2	Activation of T lymphocytes
<input type="checkbox"/>	3	PRDM1	IL2	Activation of T lymphocytes
<input checked="" type="checkbox"/>	4	HLA-DRB1	IL2	Activation of T lymphocytes
<input type="checkbox"/>	5	HLA-DRB1	TCR	Activation of T lymphocytes
<input checked="" type="checkbox"/>	6	PRDM1	Immunoglobulin	Activation of T lymphocytes



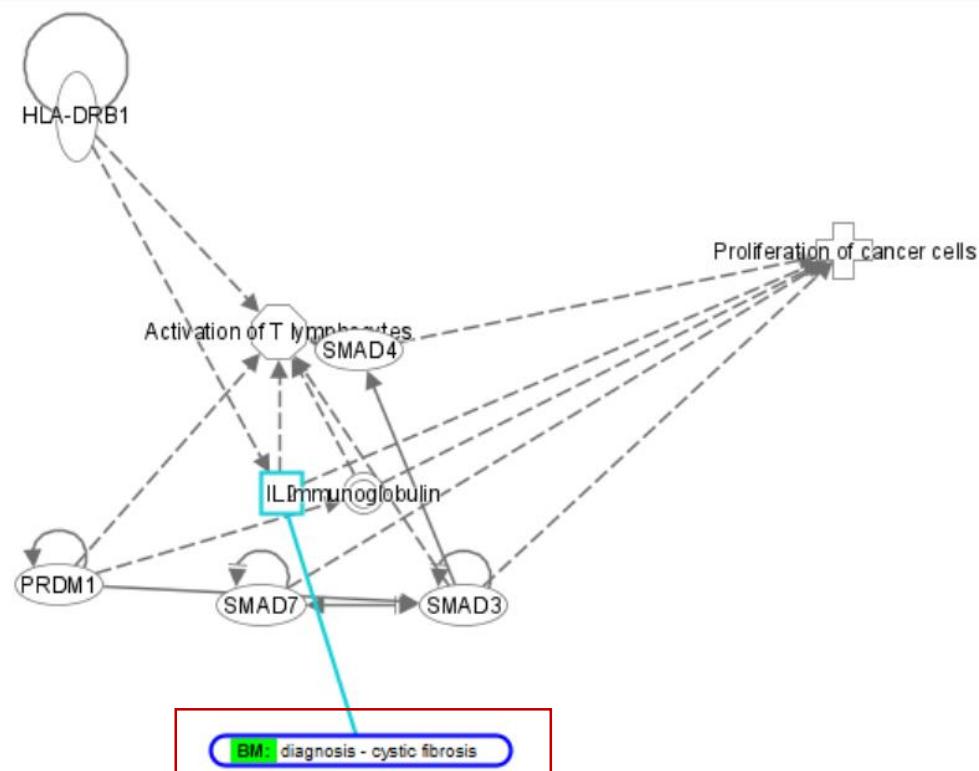
Overlay: Biomarkers

Select Biomarker labels from table to be displayed on pathway.

<input type="checkbox"/>	Application	Disease	# Molecules	Molecule(s)
<input type="checkbox"/>	diagnosis	non-insulin-dependent...	1	IL2
<input checked="" type="checkbox"/>	diagnosis	cystic fibrosis	1	IL2
<input type="checkbox"/>	diagnosis	sarcoidosis	1	IL2
<input type="checkbox"/>	diagnosis	sarcopenia	1	IL2
<input type="checkbox"/>	diagnosis	Parkinson's disease	1	IL2
<input type="checkbox"/>	diagnosis	candidiasis	1	IL2
<input type="checkbox"/>	diagnosis	asthma	1	IL2
<input type="checkbox"/>	disease progression	Parkinson's disease	1	IL2
<input type="checkbox"/>	efficacy	HIV infection	1	IL2
<input type="checkbox"/>	efficacy	small B-cell lymphocytic ...	1	IL2
<input type="checkbox"/>	efficacy	leukemia	1	IL2
<input type="checkbox"/>	efficacy	lymphoma	1	IL2
<input type="checkbox"/>	efficacy	multiple myeloma	1	IL2
<input type="checkbox"/>	efficacy	obesity	1	IL2
<input type="checkbox"/>	efficacy	non-Hodgkin lymphoma	1	IL2
<input type="checkbox"/>	efficacy	rheumatoid arthritis	1	IL2
<input type="checkbox"/>	efficacy	myeloproliferative synd...	1	IL2
<input type="checkbox"/>	efficacy	acute myeloid leukemia	1	IL2
<input type="checkbox"/>	efficacy	myelodysplastic syndro...	1	IL2
<input type="checkbox"/>	efficacy	marginal zone cell lym...	1	IL2
<input type="checkbox"/>	efficacy	follicular lymphoma	1	IL2
<input type="checkbox"/>	efficacy	breast cancer	1	IL2
<input type="checkbox"/>	prognosis	colorectal cancer	1	SMAD4
<input type="checkbox"/>	response to therapy	rheumatoid arthritis	1	IL2
<input type="checkbox"/>	unspecified applic...	rheumatoid arthritis	1	SMAD4
<input type="checkbox"/>	unspecified applic...	gastric mucosa-associat...	1	Immunoglobulin
<input type="checkbox"/>	unspecified applic...	ovarian cancer	1	Immunoglobulin
<input type="checkbox"/>	unspecified applic...	colorectal carcinoma	1	SMAD4

Mode Label Interactive OFF

New My Pathway 6



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Add To My Pathway

Highlight

View Shortest Paths + 1 (389)

Paths 1 - 50

<input type="checkbox"/>	Paths	Set A Molecules	Node 1	Set B Molecules
<input checked="" type="checkbox"/>	1	SMAD3	SMAD4	Activation of T lymphocytes
<input type="checkbox"/>	2	SMAD3	IL2	Activation of T lymphocytes
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<input checked="" type="checkbox"/>	4	HLA-DRB1	IL2	Activation of T lymphocytes
<input type="checkbox"/>	5	HLA-DRB1	TCR	Activation of T lymphocytes
<input checked="" type="checkbox"/>	6	PRDM1	Immunoglobulin	Activation of T lymphocytes

☐ Ignore Analysis Cutoff

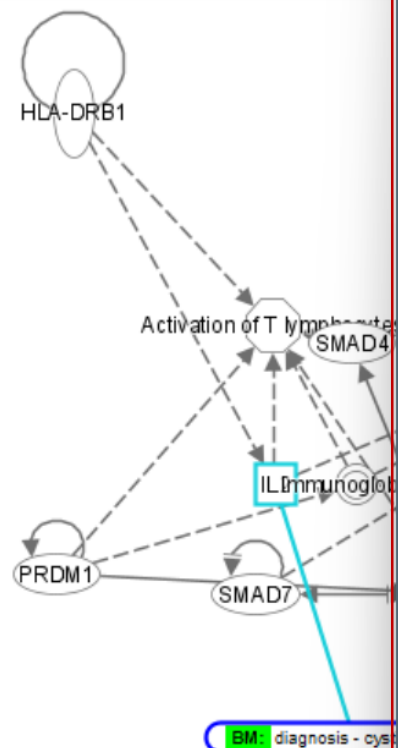
Index	Name <i>(select to overlay)</i>
Please add a dataset, analysis or list.	

Please add a dataset, analysis or list.

Add...

Show node charts for multi overlay: ☒ Always ☐ For rows selected above ☐

Graph overlay options

☒ Display isoform badges on nodes

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[Add To My Pathway](#)

Highlight

<input type="checkbox"/>	Paths	Set A Molecules	Node 1	Set B Molecules
<input checked="" type="checkbox"/>	1	SMAD3	SMAD4	Activation of T lymphocytes
<input type="checkbox"/>	2	SMAD3	IL2	Activation of T lymphocytes
<input type="checkbox"/>	3	PRDM1	IL2	Activation of T lymphocytes
<input checked="" type="checkbox"/>	4	HLA-DRB1	IL2	Activation of T lymphocytes
<input type="checkbox"/>	5	HLA-DRB1	TCR	Activation of T lymphocytes
<input checked="" type="checkbox"/>	6	PRDM1	Immunoglobulin	Activation of T lymphocytes

Select a dataset, analysis or list

Please manually select analyses, datasets, and lists to overlay, or use search below

A-Z Sort

ulcerative colitis

Search

Refresh

▼ **My Projects**

➤ **GWAS**

➤ Human Genes Chromosomal Location


QIAGEN Coronavirus Networks

- Ingenuity KEGG gene lists

➤ **Tissue Expression**

➤ Example Analyses

- Training

>  Shared Projects

Use search (above) for Analysis Match datasets and analyses

OK

Cancel

ulcerative colitis

Search



Examples: liver, "mouse OR rat", "HeLa NOT 3T3", "p?3", "ovar**"

Search Results

Showing first 1779 results out of 1779 in 29233ms for query [ulcerative colitis]

Folder Types

- [dataset \(973\)](#)
- [analysis \(806\)](#)

Projects

- [HumanDisease \(1348\)](#)

Open

Add to Overlay

Customize Table

Crea... 2023/... - 2023/... (1/45)



Name	Matching Term	Type	Creation Date
1- ulcerative colitis (UC) [colonic mucosa] 26910	exp_meta_data.control.dis...	analysis	2023/09/30 23:22
1- ulcerative colitis (UC) [colonic mucosa] 33570	exp_meta_data.case.disea...	analysis	2023/09/30 23:22
1- ulcerative colitis (UC) [colon] 10226	exp_meta_data.control.dis...	analysis	2023/09/30 23:22
1- ulcerative colitis (UC) [colonic mucosa] 16417	exp_meta_data.compariso...	analysis	2023/09/30 23:21
1- ulcerative colitis (UC) [ileal mucosa] 10425	exp_meta_data.case.disea...	analysis	2023/09/30 23:21
1- ulcerative colitis (UC) [intestinal organoid] 1527	exp_meta_data.compariso...	analysis	2023/09/30 23:21
1- ulcerative colitis (UC) [colonic mucosa] 17871	exp_meta_data.compariso...	analysis	2023/09/30 23:20
1- ulcerative colitis (UC) [mesenteric adipose tissue] 25716	exp_meta_data.compariso...	analysis	2023/09/30 23:20
1- ulcerative colitis (UC) [colonic mucosa] 19885	exp_meta_data.compariso...	analysis	2023/09/30 23:20
1- ulcerative colitis (UC) [pouch mucosa] 26938	exp_meta_data.compariso...	analysis	2023/09/30 23:20
1- ulcerative colitis (UC) [colonic mucosa] 25110	exp_meta_data.compariso...	analysis	2023/09/30 23:19
1- ulcerative colitis (UC) [rectum] 8043	exp_meta_data.compariso...	analysis	2023/09/30 23:19
1- ulcerative colitis (UC) [colonic mucosa] 26770	exp_meta_data.compariso...	analysis	2023/09/30 23:19
1- ulcerative colitis (UC) [sigmoid colon] 19333	exp_meta_data.control.dis...	analysis	2023/09/30 23:19
1- ulcerative colitis (UC) [colonic mucosa] 29017	exp_meta_data.compariso...	analysis	2023/09/30 23:18
1- ulcerative colitis (UC) [peripheral blood] 14819	exp_meta_data.compariso...	analysis	2023/09/30 23:18
1- ulcerative colitis (UC) [peripheral blood] 154	exp_meta_data.control.dis...	analysis	2023/09/30 23:17
1- ulcerative colitis (UC) [rectal mucosa] 2363	exp_meta_data.compariso...	analysis	2023/09/30 23:17

Libraries > OmicSoft > DiseaseLand > HumanDisease > Analyses

[1- ulcerative colitis \(UC\) \[colon\] 10226](#)

Case/Control Differences

Key	Case	Control
response	sensitive	resistant

Comparison Context

comparisoncategory **Responder vs. Non-Responder**comparisoncontrast **TNFInadequateResponder:Response => NA -> sensitive vs resistant**diseasestate **ulcerative colitis (UC)**dosage **100 mg**organism **human**platformname **NGS.Illumina.HiSeq2000**subjecttreatment **etrolizumab**tissue **colon**treatmentstatus **none**

All Experiment Metadata

case.diseasestate **ulcerative colitis (UC)**case.dosage **100 mg**case.response **sensitive**case.sampleids **GSM1872906;GSM1872912;GSM1872913;GSM1872914**

ulcerative colitis

Search



Examples: liver, "mouse OR rat", "HeLa NOT 3T3", "p?3", "ovar*"

Search Results

Showing first 1779 results out of 1779 in 29233ms for query [ulcerative colitis]

Folder Types

- [dataset \(973\)](#)
- [analysis \(806\)](#)

Projects

- [HumanDisease \(1348\)](#)

Open

Add to Overlay

Customize Table

Crea... 2023/... - 2023/... (1/45)



Name	Matching Term	Type	Creation Date
1- ulcerative colitis (UC) [colonic mucosa] 26910	exp_meta_data.control.dis...	analysis	2023/09/30 23:22
1- ulcerative colitis (UC) [colonic mucosa] 33570	exp_meta_data.case.disea...	analysis	2023/09/30 23:22
1- ulcerative colitis (UC) [colon] 10226	exp_meta_data.control.dis...	analysis	2023/09/30 23:22
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1- ulcerative colitis (UC) [ileal mucosa] 10425	exp_meta_data.case.disea...	analysis	2023/09/30 23:21
1- ulcerative colitis (UC) [intestinal organoid] 1527	exp_meta_data.compariso...	analysis	2023/09/30 23:21
1- ulcerative colitis (UC) [colonic mucosa] 17871	exp_meta_data.compariso...	analysis	2023/09/30 23:20
1- ulcerative colitis (UC) [mesenteric adipose tissue] 25716	exp_meta_data.compariso...	analysis	2023/09/30 23:20
1- ulcerative colitis (UC) [colonic mucosa] 19885	exp_meta_data.compariso...	analysis	2023/09/30 23:20
1- ulcerative colitis (UC) [pouch mucosa] 26938	exp_meta_data.compariso...	analysis	2023/09/30 23:20
1- ulcerative colitis (UC) [colonic mucosa] 25110	exp_meta_data.compariso...	analysis	2023/09/30 23:19

Analyses to overlay

1- ulcerative colitis (UC) [colon] 10226

Up ^

Down v

Remove

Clear All

Overlay Now

Libraries > OmicSoft > DiseaseLand > HumanDisease > Analyses

[1- ulcerative colitis \(UC\) \[colon\] 10226](#)

Case/Control Differences

Key	Case	Control
response	sensitive	resistant

Comparison Context

comparisoncategory **Responder vs. Non-Responder**comparisoncontrast **TNFInadequateResponder:Response => NA -> sensitive vs resistant**diseasestate **ulcerative colitis (UC)**dosage **100 mg**organism **human**platformname **NGS.Illumina.HiSeq2000**subjecttreatment **etrolizumab**tissue **colon**treatmentstatus **none**

All Experiment Metadata

case.diseasestate **ulcerative colitis (UC)**case.dosage **100 mg**case.response **sensitive**case.sampleids **GSM1872906;GSM1872912;GSM1872913;GSM1872914**

位

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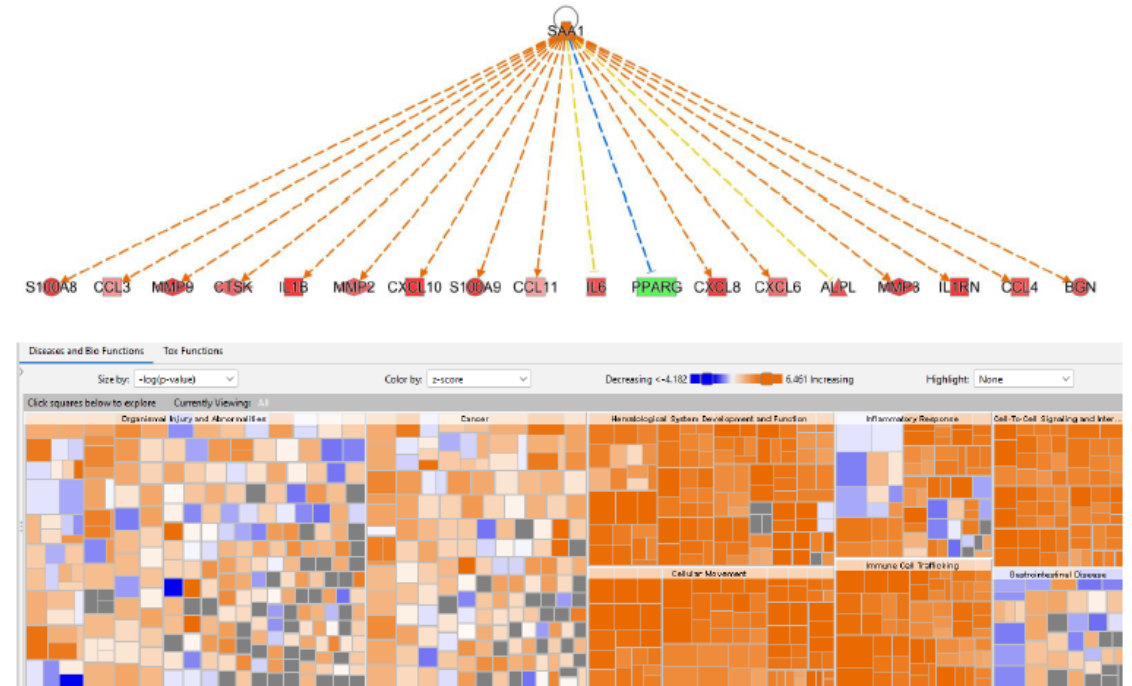
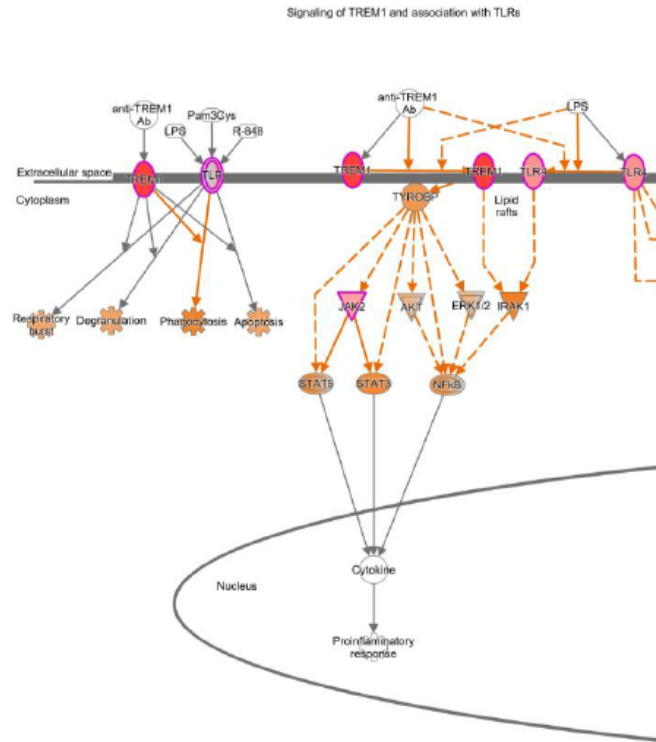
位

位

位

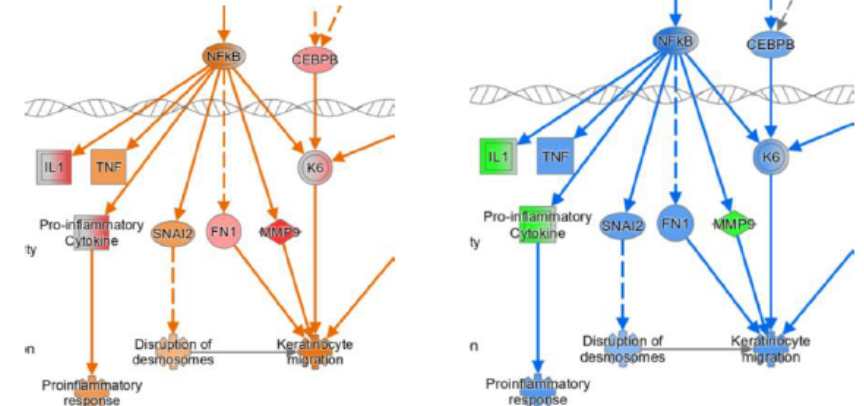
位

	A	B	C
1	geneid	UCvsNormalLog2FoldChange	UCvsNormal.pval
2	DDX11L1	-0.1067	0.2878
3	WASH7P	-0.1883	0.0097
4	FAM138F	-0.0761	0.4699
5	OR4F5	0.1474	0.5311
6	LOC729737	0.4789	0.0017
7	LOC100133331	0.4789	0.0017
8	LOC100132062	0.4789	0.0017
9	OR4F29	0.2495	0.2389
10	JA429831	0.1215	0.3338
11	JB137814	-0.674	1.6381E-06
12	M37726	-1.0651	0.00000576
13	LINC00115	-0.1696	0.021
14	LOC643837	0.1025	0.3021
15	FAM41C	0.2098	0.2554
16	SAIMD11	-0.0552	0.4088
17	NOC2L	0.3408	1.2575E-06
18	KLHL17	0.1497	0.0082
19	PLEKHN1	0.1463	0.0088
20	C10orf170	-0.1649	0.0085
1546	TTTY13	-0.3543	0.0003
1547	RBM1E	-0.3167	0.016
1548	PRY2	-0.1792	0.0084
1549	TTTY6	-0.2051	0.0005
1550	RBM1J	-0.3167	0.016
1551	TTTY5	-0.105	0.0743
1552	RBM2FP	-0.5248	0.0939
1553	RBM1F	-0.5248	0.0939
1554	PRY	-0.1792	0.0084
1555	BPY2	-0.1446	0.0562
1556	DAZ1	0.0734	0.5811
1557	DAZ4	0.0734	0.5811
1558	DAZ2	0.0734	0.5811
1559	DAZ3	0.0734	0.5811
1560	CDY1	-0.0319	0.6541
1561	CSPG4P1Y	-0.0678	0.3043
1562	GOLGA2P2Y	-0.0872	0.1077
1563	U6_83	0.21	0.1



- Data upload and analysis setup
- Canonical pathways
- Upstream regulators
- Diseases and functions
- Comparison analysis

Ulcerative Colitis vs Normal Treatment vs Baseline





Better Care with Better Knowledge

若有需要進一步的資訊或在使用軟體上遇到問題歡迎聯繫以下窗口:

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熊嘉妮 專案主任 ChristineHsiung@gga.asia 02-2795 1777 #3028

Bioinfo@GGA.ASIA