表观资源库
EWAS Open Platform介绍

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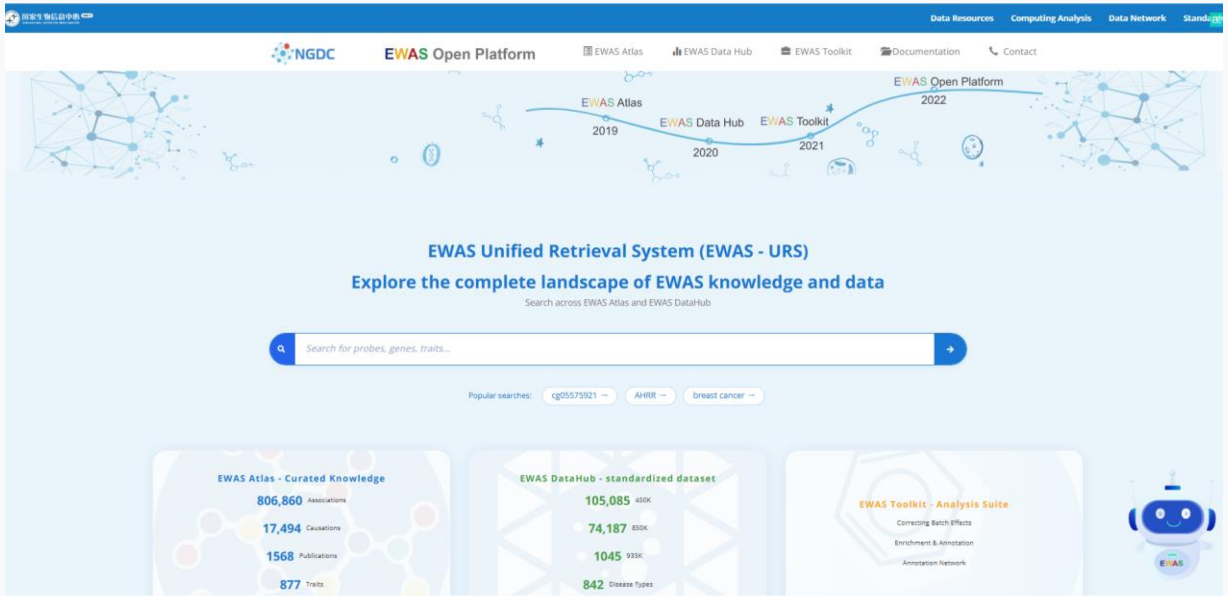
日期：2026年1月25日

https://ngdc.cncb.ac.cn/ewas/

Nucleic Acids Res, 2019, 2020, 2022,2025

表观资源库EWAS Open Platform



EWAS Unified Retrieval System (EWAS - URS)

Explore the complete landscape of EWAS knowledge and data

Search across EWAS Atlas and EWAS DataHub

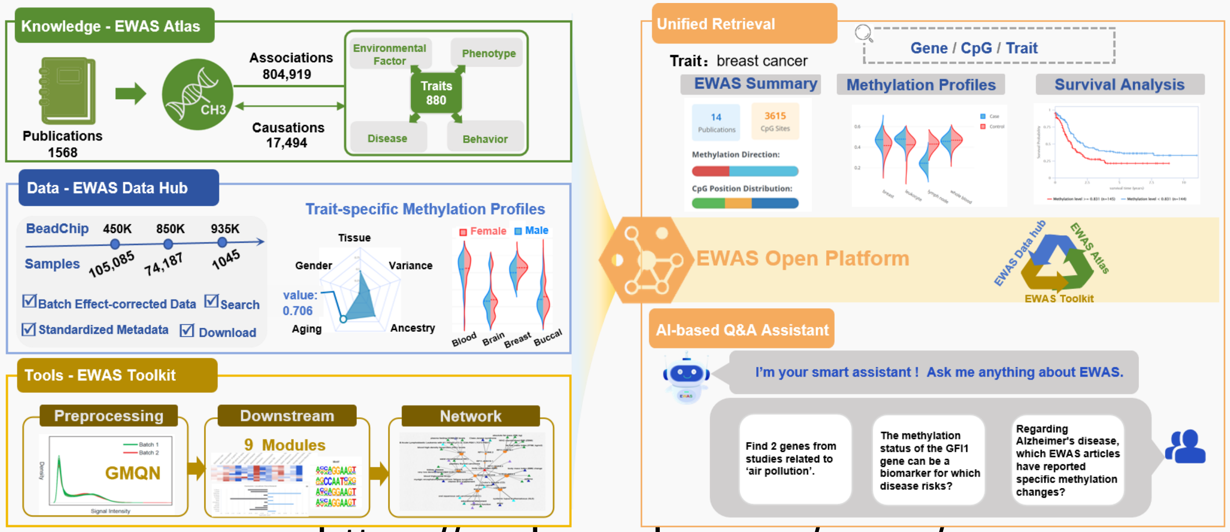
Search for probes, genes, traits...

Popular searches: cg05575921 → AHRR → breast cancer →

EWAS Atlas - Curated Knowledge	EWAS DataHub - standardized dataset	EWAS Toolkit - Analysis Suite
806,860 Associations	105,085 450K	Correcting Batch Effects
17,494 Causations	74,187 850K	Enrichment & Annotation
1568 Publications	1045 935K	Annotation Network
877 Traits	842 Disease Types	

构建集数据、工具、知识为一体的芯片甲基化资源平台，提供集成化全局检索、智能问答助手，致力为人口健康、公共安全等领域研究提供综合、高效的服务支持

- ➢ 数据
  - 通过设定受控词表，建立标准化的元数据处理流程
  - 通过去批次效应工具GMQN，建立标准化的数据处理流程
  - 组织、性别、年龄种族、BMI、疾病相关的DNA甲基化图谱
- ➢ 知识
  - 文献审编的性状-甲基化关联知识图谱
  - 文献审编的性状-多组学因果知识网络
- ➢ 工具
  - 上游：去批次效应
  - 下游：富集分析
  - 机制解析：调控网络



I'm your smart assistant ! Ask me anything about EWAS.

Find 2 genes from studies related to 'air pollution'.

The methylation status of the GFI1 gene can be a biomarker for which disease risks?

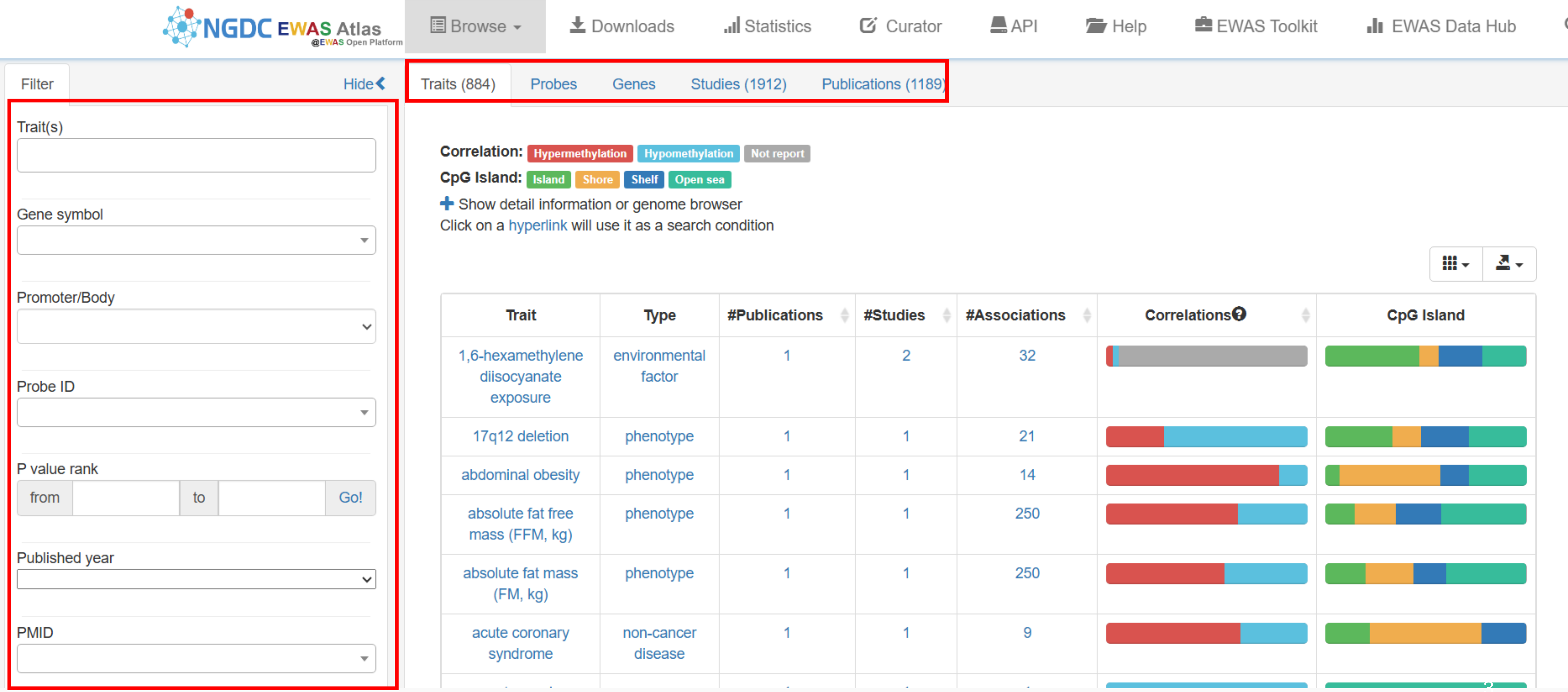
Regarding Alzheimer's disease, which EWAS articles have reported specific methylation changes?

https://ngdc.cncb.ac.cn/ewas/

Nucleic Acids Res, 2019, 2020, 2022,2025

EWAS Open Platform-关联知识信息

EWAS Atlas / Traits/ Association



Trait(s)

Gene symbol

Promoter/Body

Probe ID

P value rank from to Go!

Published year

PMID

Correlation: Hypermethylation Hypomethylation Not report

CpG Island: Island Shore Shelf Open sea

+ Show detail information or genome browser

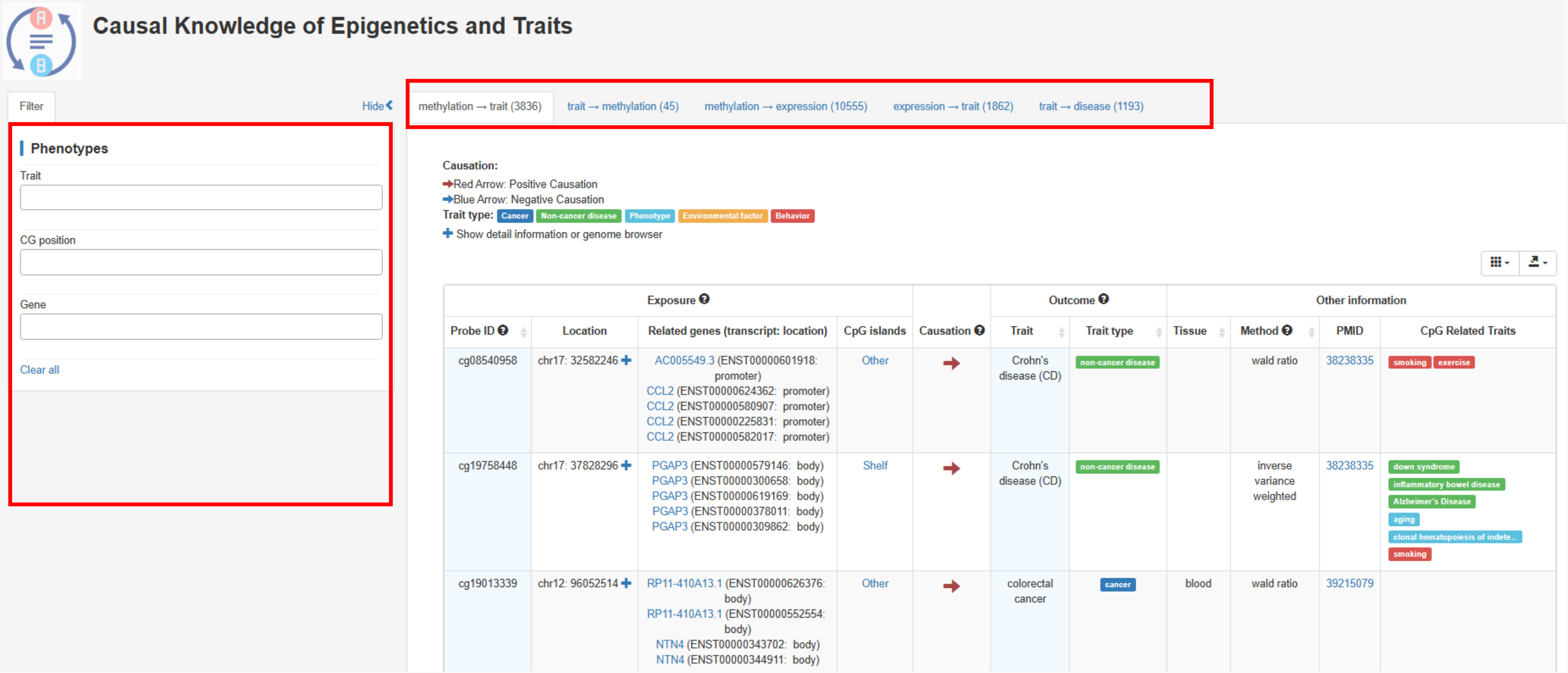
Click on a hyperlink will use it as a search condition

Trait	Type	#Publications	#Studies	#Associations	Correlations	CpG Island
1,6-hexamethylene diisocyanate exposure	environmental factor	1	2	32		
17q12 deletion	phenotype	1	1	21		
abdominal obesity	phenotype	1	1	14		
absolute fat free mass (FFM, kg)	phenotype	1	1	250		
absolute fat mass (FM, kg)	phenotype	1	1	250		
acute coronary syndrome	non-cancer disease	1	1	9		

https://ngdc.cncb.ac.cn/ewas/browse?target=traits

EWAS Open Platform-因果知识信息

EWAS Atlas / Traits/ Causality



Causal Knowledge of Epigenetics and Traits

Phenotypes

Trait

CG position

Gene

Clear all

Causation:

➡Red Arrow: Positive Causation

➡Blue Arrow: Negative Causation

Trait type: Cancer Non-cancer disease Phenotype Environmental factor Behavior

✚ Show detail information or genome browser

Exposure					Outcome		Other information			
Probe ID	Location	Related genes (transcript: location)	CpG islands	Causation	Trait	Trait type	Tissue	Method	PMID	CpG Related Traits
cg08540958	chr17: 32582246 ✚	AC005549.3 (ENST00000601918: promoter) CCL2 (ENST00000624362: promoter) CCL2 (ENST00000580907: promoter) CCL2 (ENST00000225831: promoter) CCL2 (ENST00000582017: promoter)	Other	➡	Crohn's disease (CD)	non-cancer disease		wald ratio	38238335	smoking exercise
cg19758448	chr17: 37828296 ✚	PGAP3 (ENST00000579146: body) PGAP3 (ENST00000300658: body) PGAP3 (ENST00000619169: body) PGAP3 (ENST00000378011: body) PGAP3 (ENST00000309862: body)	Shelf	➡	Crohn's disease (CD)	non-cancer disease		inverse variance weighted	38238335	down syndrome inflammatory bowel disease Alzheimer's Disease aging clonal hematopoiesis of indete... smoking
cg19013339	chr12: 96052514 ✚	RP11-410A13.1 (ENST00000626376: body) RP11-410A13.1 (ENST00000552554: body) NTN4 (ENST00000343702: body) NTN4 (ENST00000344911: body)	Other	➡	colorectal cancer	cancer	blood	wald ratio	39215079	

https://ngdc.cncb.ac.cn/ewas/browseMR

EWAS Open Platform-关联知识查询

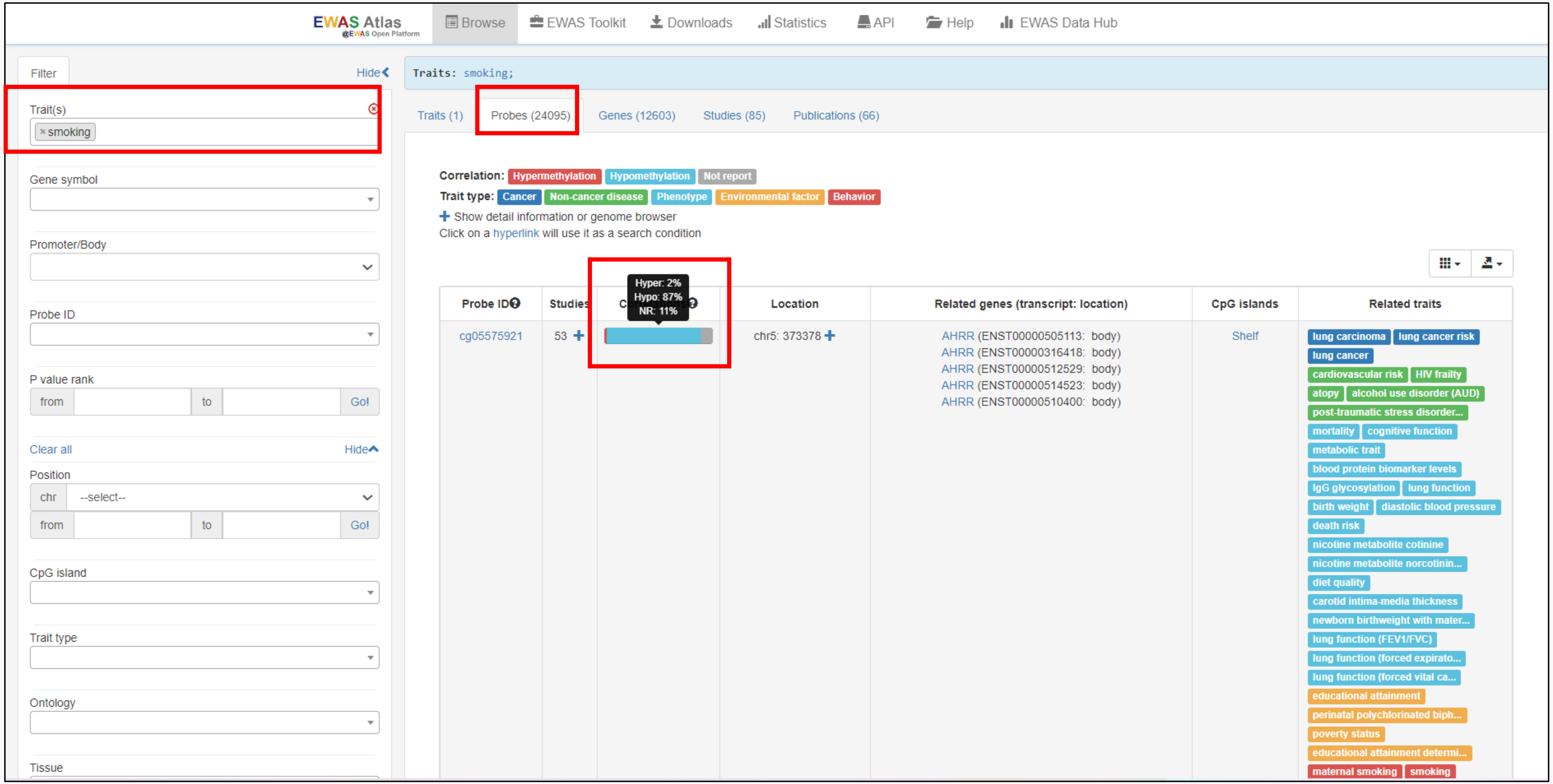
- 示例问题1：如何查看感兴趣的trait与甲基化的研究现状?

例如：吸烟与哪些位点的甲基化变化有关呢？哪些文章报道了呢？结果是否稳定呢?

EWAS Open Platform-关联知识查询

53篇研究都报道了吸烟与 AHRR相关位点的甲基化变化有关。大多都报道的是差异低甲基化。

EWAS Atlas / Probes



Trait(s): × smoking

Gene symbol

Promoter/Body

Probe ID

P value rank: from … to … Go!

Position: chr --select-- from … to … Go!

CpG island

Trait type

Ontology

Tissue

Traits: smoking;

Traits (1) | Probes (24095) | Genes (12603) | Studies (85) | Publications (66)

Correlation: Hypermethylation Hypomethylation Not report

Trait type: Cancer Non-cancer disease Phenotype Environmental factor Behavior

+ Show detail information or genome browser

Click on a hyperlink will use it as a search condition

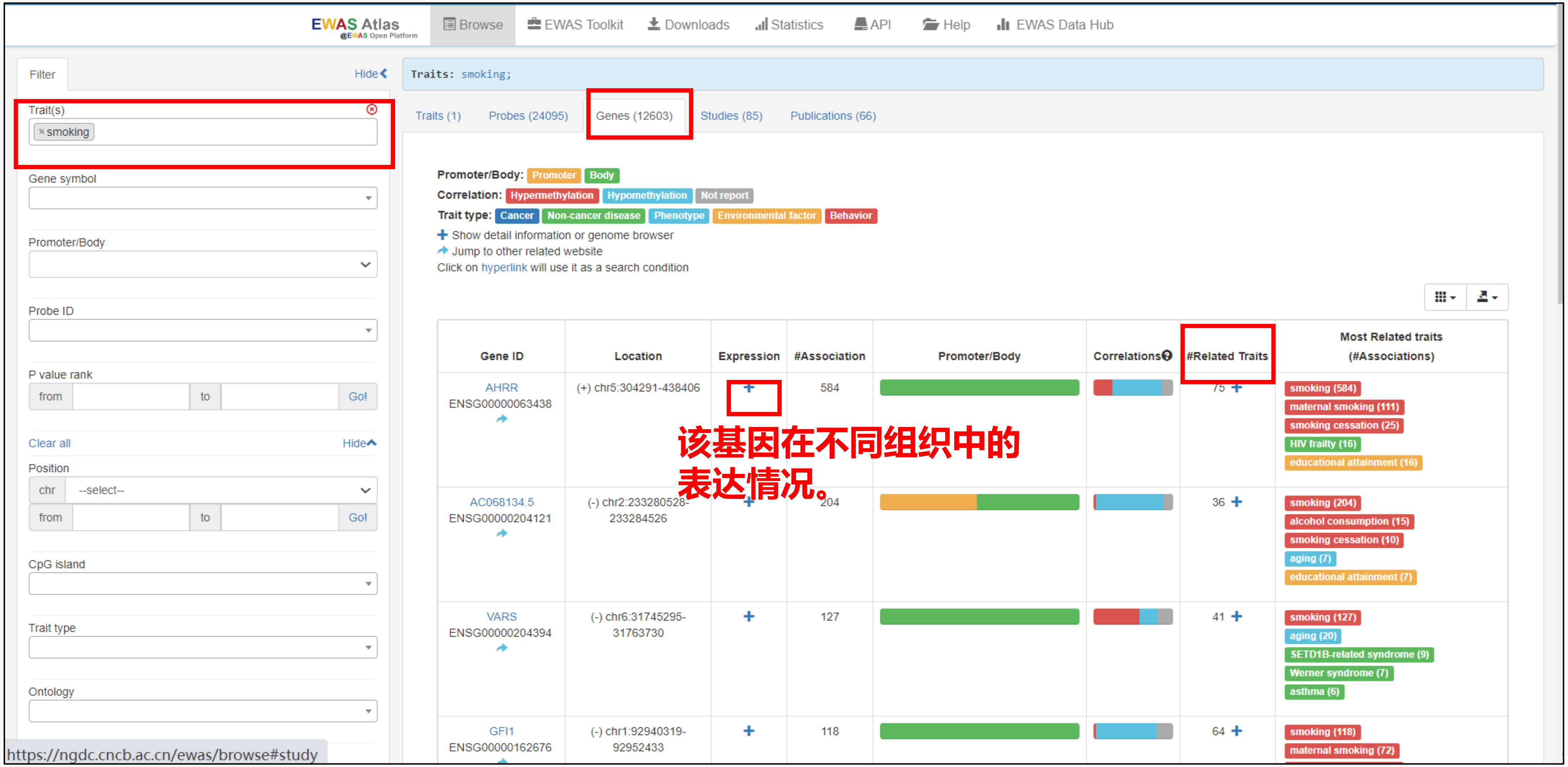
Probe ID❓	Studies	C	Location	Related genes (transcript: location)	CpG islands	Related traits
cg05575921	53 +	Hyper: 2% Hypo: 87% NR: 11%	chr5: 373378 +	AHRR (ENST00000505113: body) AHRR (ENST00000316418: body) AHRR (ENST00000512529: body) AHRR (ENST00000514523: body) AHRR (ENST00000510400: body)	Shelf	lung carcinoma lung cancer risk lung cancer cardiovascular risk HIV frailty atopy alcohol use disorder (AUD) post-traumatic stress disorder... mortality cognitive function metabolic trait blood protein biomarker levels IgG glycosylation lung function birth weight diastolic blood pressure death risk nicotine metabolite cotinine nicotine metabolite norcotinin... diet quality carotid intima-media thickness newborn birthweight with mater... lung function (FEV1/FVC) lung function (forced expirato... lung function (forced vital ca... educational attainment perinatal polychlorinated biph... poverty status educational attainment determi... maternal smoking smoking

https://ngdc.cncb.ac.cn/ewas/browse?target=traits

EWAS Open Platform-关联知识查询

AHRR基因位点除了与吸烟性状相关，还有戒烟、HIV易感性和教育程度等性状有关。

EWAS Atlas / Genes



Traits: smoking;

Promoter/Body: Promoter Body

Correlation: Hypermethylation Hypomethylation Not report

Trait type: Cancer Non-cancer disease Phenotype Environmental factor Behavior

+ Show detail information or genome browser

Jump to other related website

Click on hyperlink will use it as a search condition

Gene ID	Location	Expression	#Association	Promoter/Body	Correlations	#Related Traits	Most Related traits (#Associations)
AHRR ENSG00000063438	(+) chr5:304291-438406	+	584			75 +	smoking (584) maternal smoking (111) smoking cessation (25) HIV frailty (16) educational attainment (16)
AC068134.5 ENSG00000204121	(-) chr2:233280528-233284526	+	204			36 +	smoking (204) alcohol consumption (15) smoking cessation (10) aging (7) educational attainment (7)
VARS ENSG00000204394	(-) chr6:31745295-31763730	+	127			41 +	smoking (127) aging (20) SETD1B-related syndrome (9) Werner syndrome (7) asthma (6)
GFI1 ENSG00000162676	(-) chr1:92940319-92952433	+	118			64 +	smoking (118) maternal smoking (72)

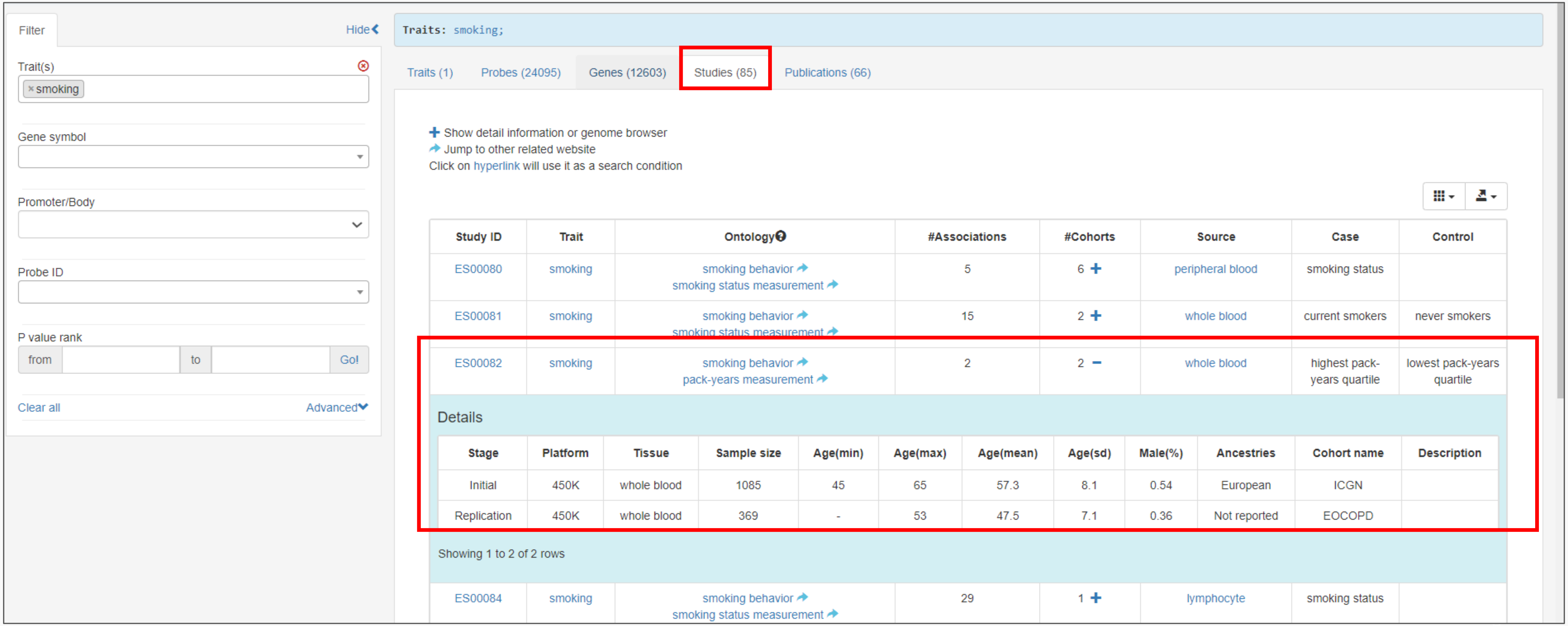
该基因在不同组织中的表达情况。

https://ngdc.cncb.ac.cn/ewas/browse?target=traits

EWAS Open Platform-关联知识查询

不同研究的结果存在差异，应该怎么寻找原因呢？查看85个吸烟相关的研究，
可以根据每个研究的队列人群的组织、年龄、性别、种族等信息来分析原因。

EWAS Atlas / Studies



Trait(s): smoking

Traits: smoking;

+ Show detail information or genome browser
Jump to other related website
Click on hyperlink will use it as a search condition

Study ID	Trait	Ontology	#Associations	#Cohorts	Source	Case	Control
ES00080	smoking	smoking behavior smoking status measurement	5	6 +	peripheral blood	smoking status	
ES00081	smoking	smoking behavior smoking status measurement	15	2 +	whole blood	current smokers	never smokers
ES00082	smoking	smoking behavior pack-years measurement	2	2 −	whole blood	highest pack-years quartile	lowest pack-years quartile

Details

Stage	Platform	Tissue	Sample size	Age(min)	Age(max)	Age(mean)	Age(sd)	Male(%)	Ancestries	Cohort name	Description
Initial	450K	whole blood	1085	45	65	57.3	8.1	0.54	European	ICGN	
Replication	450K	whole blood	369	-	53	47.5	7.1	0.36	Not reported	EOCOPD	

Showing 1 to 2 of 2 rows

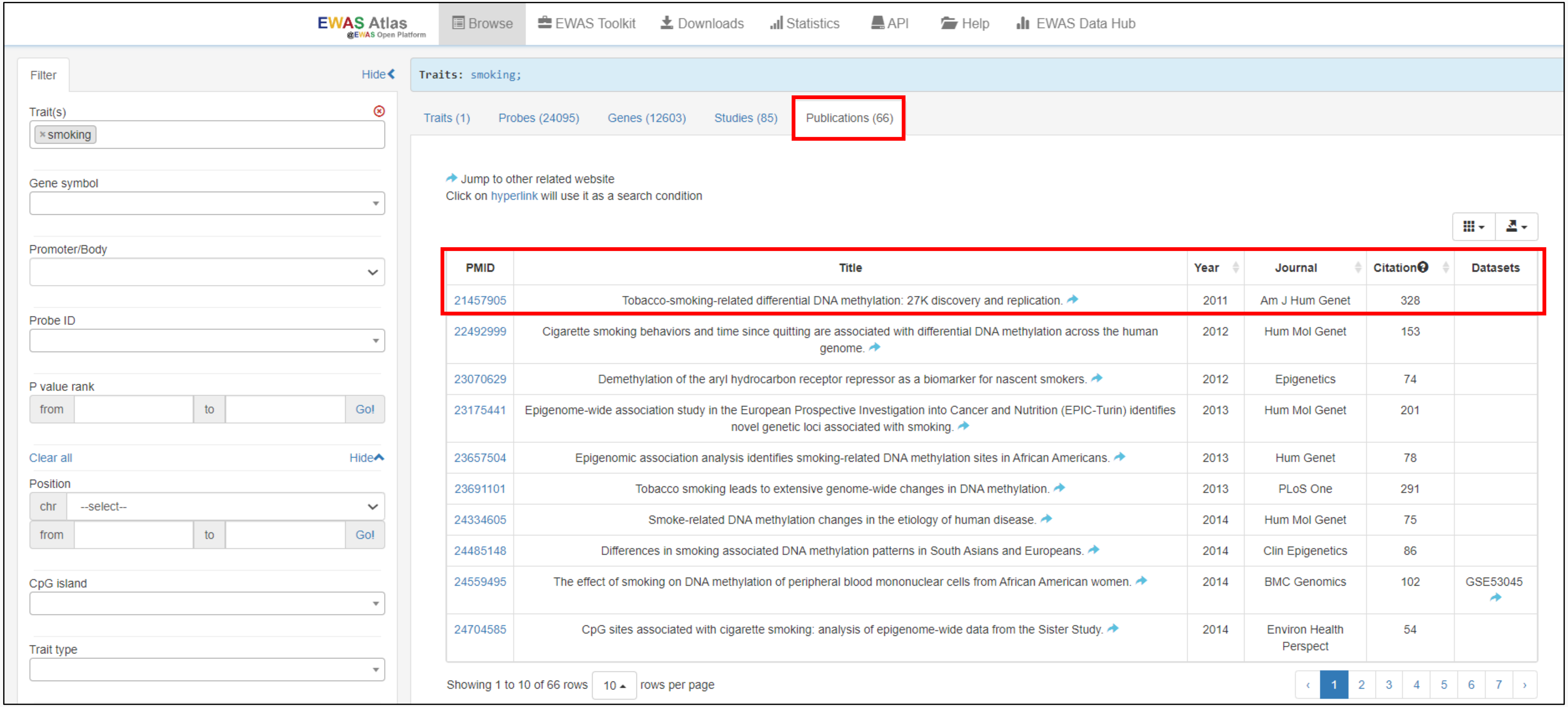
Study ID	Trait	Ontology	#Associations	#Cohorts	Source	Case	Control
ES00084	smoking	smoking behavior smoking status measurement	29	1 +	lymphocyte	smoking status	

https://ngdc.cncb.ac.cn/ewas/browse?target=traits

EWAS Open Platform-关联知识查询

EWAS Atlas / Publications

点击Publications可以查看吸烟相关甲基化位点的知识来源的期刊文章情况



Traits: smoking;

Traits (1) Probes (24095) Genes (12603) Studies (85) Publications (66)

Jump to other related website
Click on hyperlink will use it as a search condition

PMID	Title	Year	Journal	Citation	Datasets
21457905	Tobacco-smoking-related differential DNA methylation: 27K discovery and replication.	2011	Am J Hum Genet	328	
22492999	Cigarette smoking behaviors and time since quitting are associated with differential DNA methylation across the human genome.	2012	Hum Mol Genet	153	
23070629	Demethylation of the aryl hydrocarbon receptor repressor as a biomarker for nascent smokers.	2012	Epigenetics	74	
23175441	Epigenome-wide association study in the European Prospective Investigation into Cancer and Nutrition (EPIC-Turin) identifies novel genetic loci associated with smoking.	2013	Hum Mol Genet	201	
23657504	Epigenomic association analysis identifies smoking-related DNA methylation sites in African Americans.	2013	Hum Genet	78	
23691101	Tobacco smoking leads to extensive genome-wide changes in DNA methylation.	2013	PLoS One	291	
24334605	Smoke-related DNA methylation changes in the etiology of human disease.	2014	Hum Mol Genet	75	
24485148	Differences in smoking associated DNA methylation patterns in South Asians and Europeans.	2014	Clin Epigenetics	86	
24559495	The effect of smoking on DNA methylation of peripheral blood mononuclear cells from African American women.	2014	BMC Genomics	102	GSE53045
24704585	CpG sites associated with cigarette smoking: analysis of epigenome-wide data from the Sister Study.	2014	Environ Health Perspect	54	

Showing 1 to 10 of 66 rows 10 rows per page

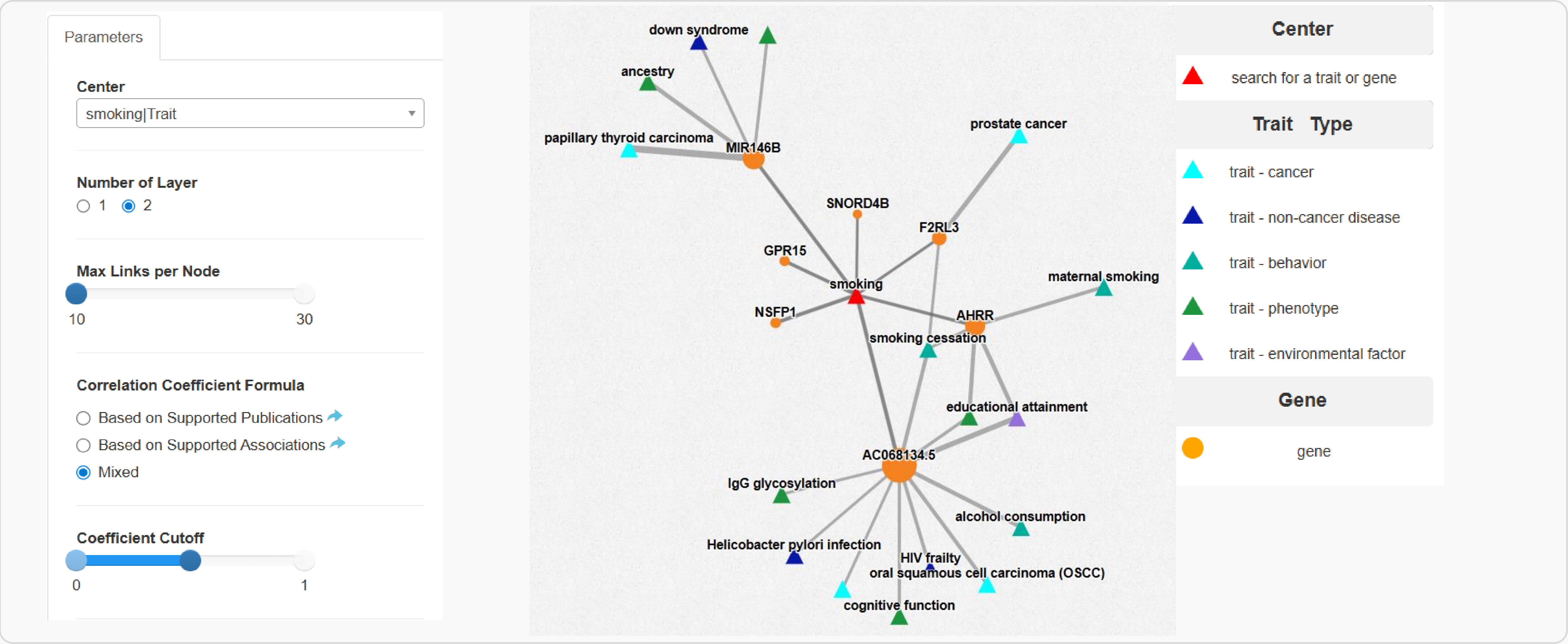
‹ 1 2 3 4 5 6 7 ›

https://ngdc.cncb.ac.cn/ewas/browse?target=traits

EWAS Open Platform-关联知识挖掘

EWAS Atlas / Network

吸烟通过***AHRR*、*F2RL3*、*MR146B***等基因的甲基化与**口腔癌、前列腺癌、甲状腺癌、唐氏综合征**等关联



Parameters

Center
smoking|Trait

Number of Layer
○ 1 ● 2

Max Links per Node
10 30

Correlation Coefficient Formula
○ Based on Supported Publications
○ Based on Supported Associations
● Mixed

Coefficient Cutoff
0 1

https://ngdc.cncb.ac.cn/ewas/network

Nucleic Acids Res, 2019, 2020, 2022,2025

EWAS Open Platform-关联知识查询

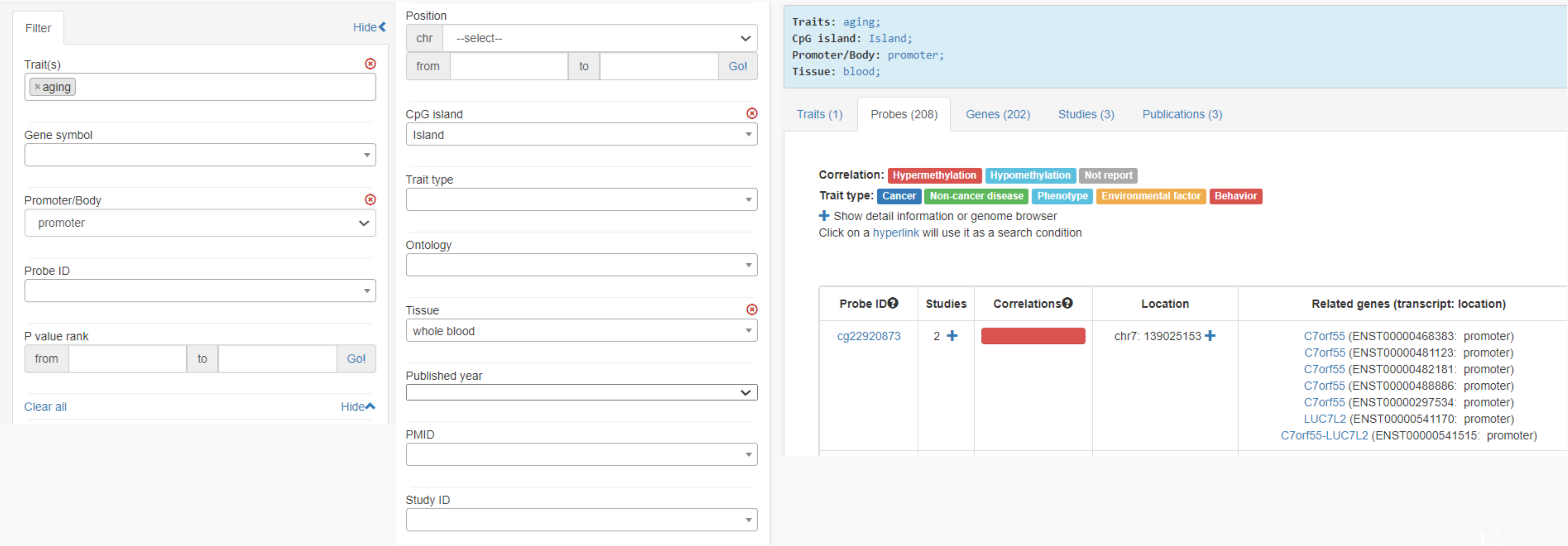
- 示例问题2：如何快速查找限定条件下trait与甲基化的关联关系？

例如：全血中， 与aging相关的位于promoter区域的CpG island甲基化

EWAS Open Platform-关联知识查询

全血中，两项研究报道了aging与C7orf55基因的promoter区域的CpG island甲基化相关。

EWAS Atlas / Probes



Trait(s): aging

Gene symbol:

Promoter/Body: promoter

Probe ID:

P value rank: from … to …

Position: chr --select-- from … to …

CpG island: Island

Trait type:

Ontology:

Tissue: whole blood

Published year:

PMID:

Study ID:

```
Traits: aging;
CpG island: Island;
Promoter/Body: promoter;
Tissue: blood;
```

Correlation: Hypermethylation Hypomethylation Not report

Trait type: Cancer Non-cancer disease Phenotype Environmental factor Behavior

+ Show detail information or genome browser

Click on a hyperlink will use it as a search condition

Probe ID	Studies	Correlations	Location	Related genes (transcript: location)
cg22920873	2 +	🟥	chr7: 139025153 +	C7orf55 (ENST00000468383: promoter) C7orf55 (ENST00000481123: promoter) C7orf55 (ENST00000482181: promoter) C7orf55 (ENST00000488886: promoter) C7orf55 (ENST00000297534: promoter) LUC7L2 (ENST00000541170: promoter) C7orf55-LUC7L2 (ENST00000541515: promoter)

EWAS Open Platform-因果知识查询

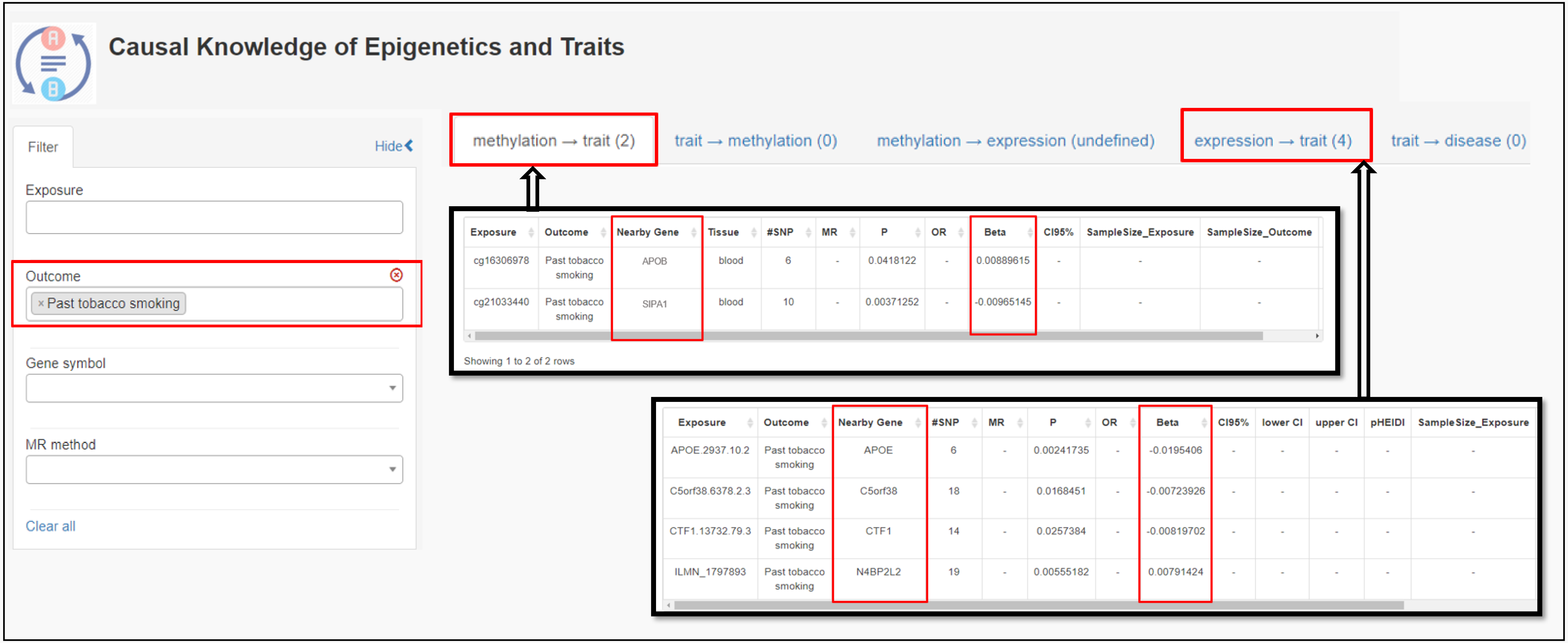
- 示例问题3：如何查看感兴趣的trait与甲基化的因果性的研究现状？

例如：哪些位点的甲基化或基因表达会引起吸烟成瘾?

EWAS Open Platform-因果知识查询

APOE基因的表达降低、 APOB（cg16306978）的甲基化升高都可能引起吸烟成瘾。

EWAS Atlas / Causality



Causal Knowledge of Epigenetics and Traits

Exposure	Outcome	Nearby Gene	Tissue	#SNP	MR	P	OR	Beta	CI95%	SampleSize_Exposure	SampleSize_Outcome
cg16306978	Past tobacco smoking	APOB	blood	6	-	0.0418122	-	0.00889615	-	-	-
cg21033440	Past tobacco smoking	SIPA1	blood	10	-	0.00371252	-	-0.00965145	-	-	-

Showing 1 to 2 of 2 rows

Exposure	Outcome	Nearby Gene	#SNP	MR	P	OR	Beta	CI95%	lower CI	upper CI	pHEIDI	SampleSize_Exposure
APOE.2937.10.2	Past tobacco smoking	APOE	6	-	0.00241735	-	-0.0195406	-	-	-	-	-
C5orf38.6378.2.3	Past tobacco smoking	C5orf38	18	-	0.0168451	-	-0.00723926	-	-	-	-	-
CTF1.13732.79.3	Past tobacco smoking	CTF1	14	-	0.0257384	-	-0.00819702	-	-	-	-	-
ILMN_1797893	Past tobacco smoking	N4BP2L2	19	-	0.00555182	-	0.00791424	-	-	-	-	-

https://ngdc.cncb.ac.cn/ewas/browseMR

EWAS Open Platform-数据查询

- 示例问题4：如何快速找到感兴趣的样本数据？

例如：寻找含有以下信息的样本

年龄：30-80岁

性别：男

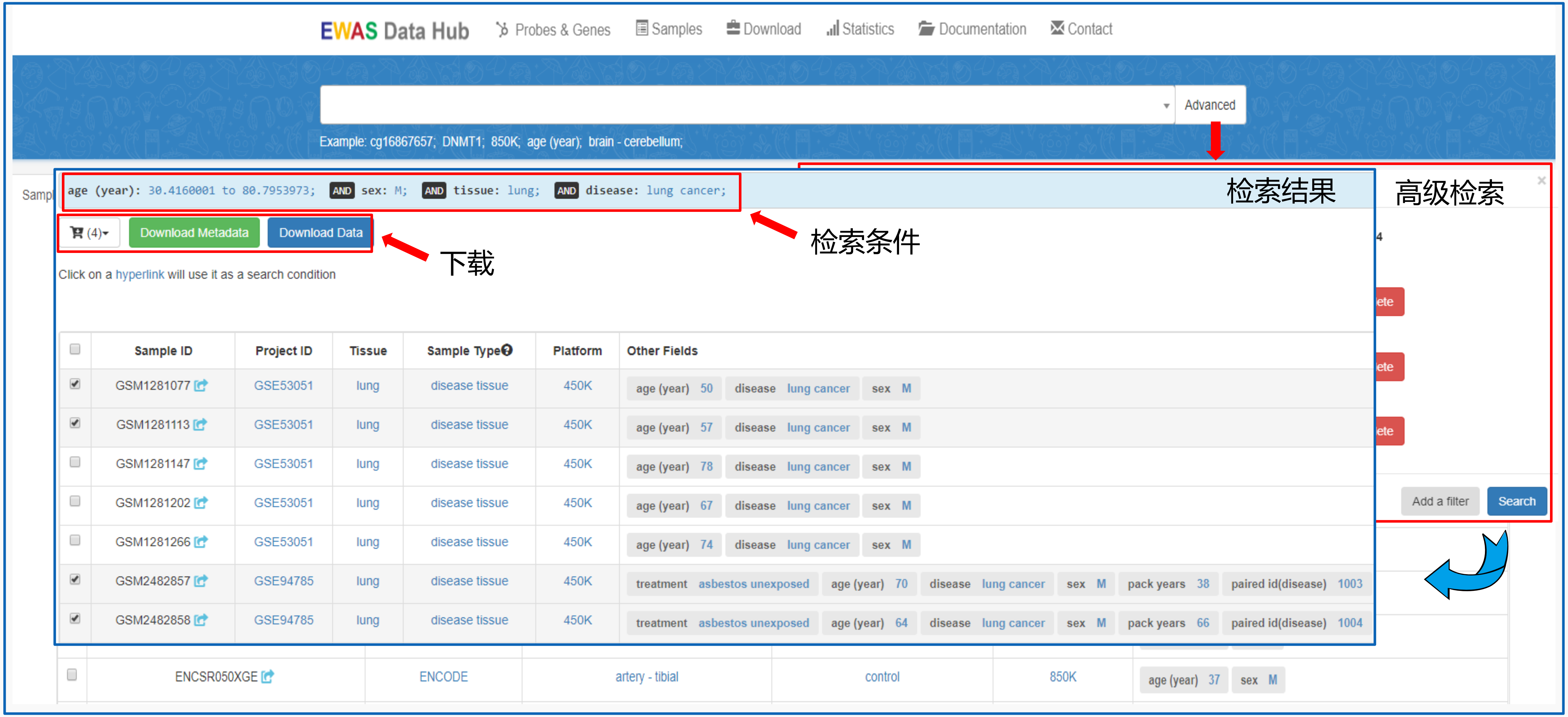
组织：肺

疾病：肺癌

EWAS Open Platform-数据查询

EWAS Data Hub / Samples

30-80岁的男性肺癌患者的甲基化样本数据查询和下载



Example: cg16867657; DNMT1; 850K; age (year); brain - cerebellum;

age (year): 30.4160001 to 80.7953973; AND sex: M; AND tissue: lung; AND disease: lung cancer;

检索结果

高级检索

检索条件

下载

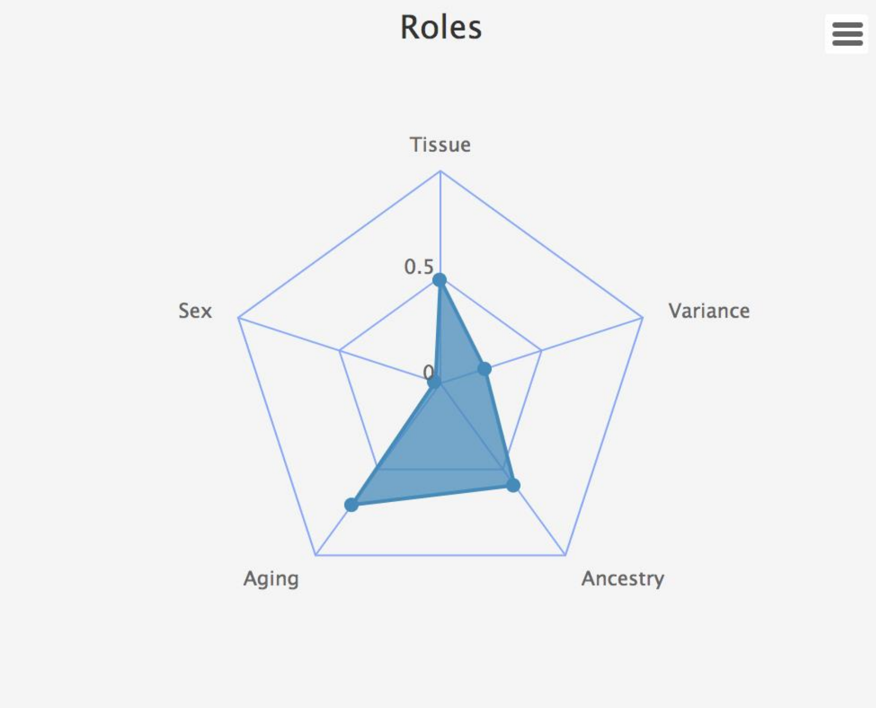
Click on a hyperlink will use it as a search condition

☐	Sample ID	Project ID	Tissue	Sample Type	Platform	Other Fields
☑	GSM1281077	GSE53051	lung	disease tissue	450K	age (year) 50 disease lung cancer sex M
☑	GSM1281113	GSE53051	lung	disease tissue	450K	age (year) 57 disease lung cancer sex M
☐	GSM1281147	GSE53051	lung	disease tissue	450K	age (year) 78 disease lung cancer sex M
☐	GSM1281202	GSE53051	lung	disease tissue	450K	age (year) 67 disease lung cancer sex M
☐	GSM1281266	GSE53051	lung	disease tissue	450K	age (year) 74 disease lung cancer sex M
☑	GSM2482857	GSE94785	lung	disease tissue	450K	treatment asbestos unexposed age (year) 70 disease lung cancer sex M pack years 38 paired id(disease) 1003
☑	GSM2482858	GSE94785	lung	disease tissue	450K	treatment asbestos unexposed age (year) 64 disease lung cancer sex M pack years 66 paired id(disease) 1004

☐	ENCSR050XGE	ENCODE	artery - tibial	control	850K	age (year) 37 sex M

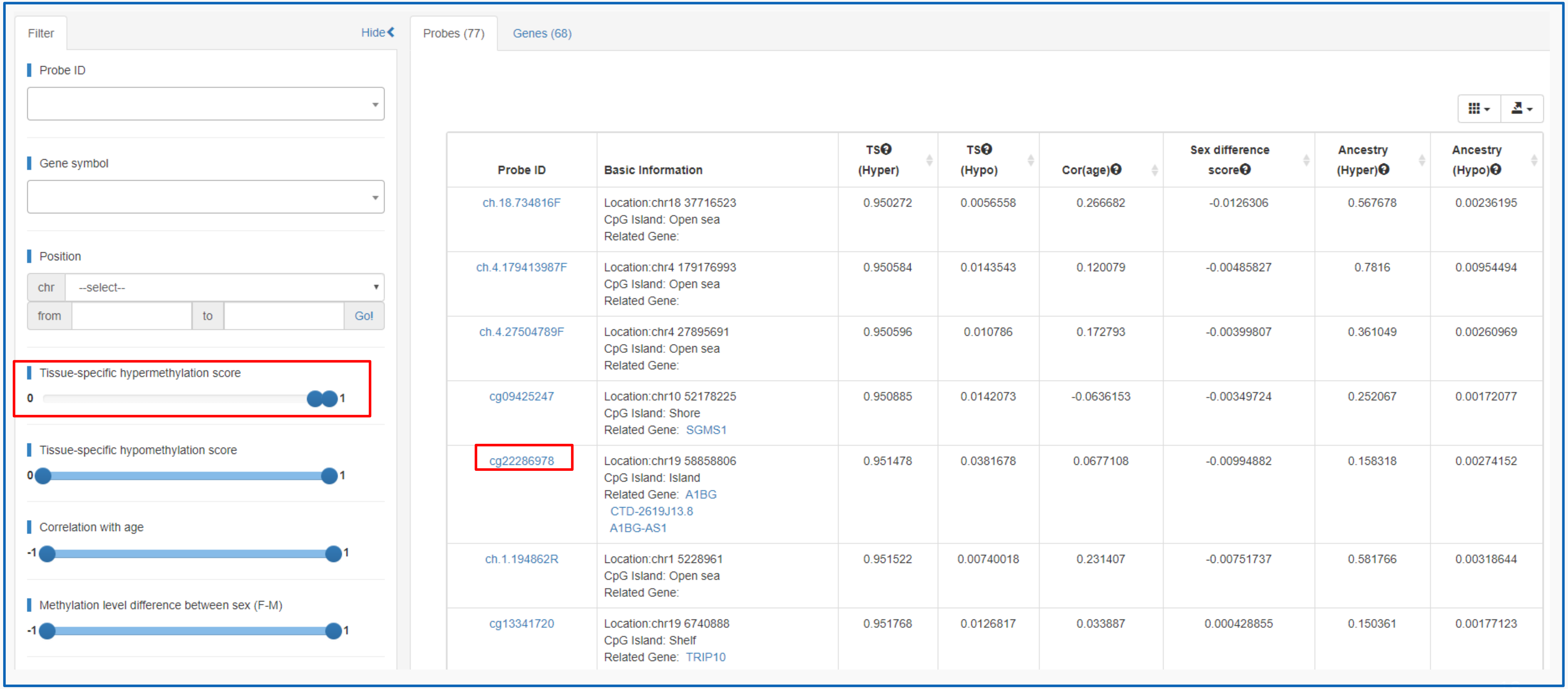
EWAS Open Platform-图谱查询

- 示例问题5：如何快速筛选出特异的甲基化位点和基因?



EWAS Open Platform-图谱查询

EWAS Data Hub / 特异甲基化评估——组织特异高甲基化



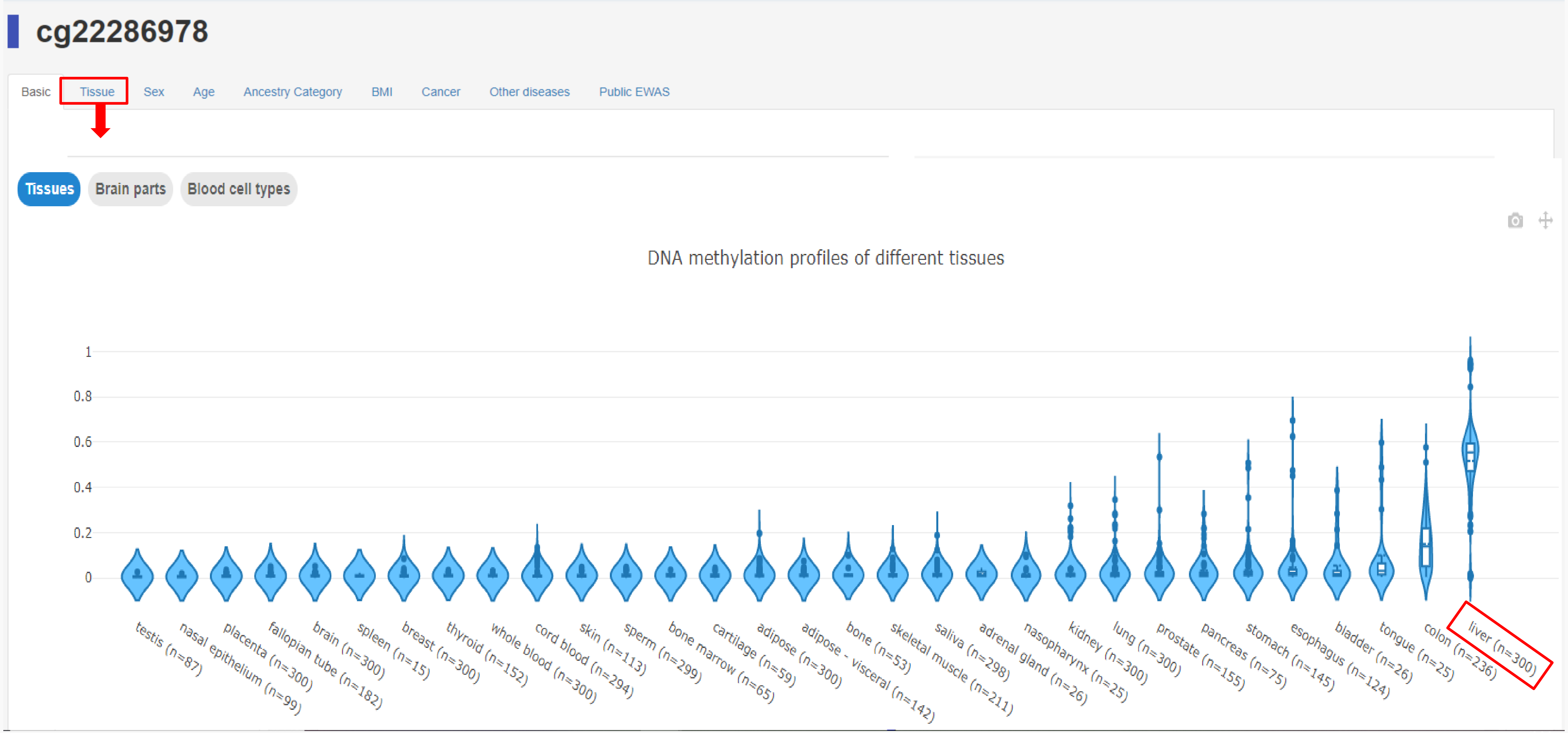
Probe ID	Basic Information	TS (Hyper)	TS (Hypo)	Cor(age)	Sex difference score	Ancestry (Hyper)	Ancestry (Hypo)
ch.18.734816F	Location:chr18 37716523 CpG Island: Open sea Related Gene:	0.950272	0.0056558	0.266682	-0.0126306	0.567678	0.00236195
ch.4.179413987F	Location:chr4 179176993 CpG Island: Open sea Related Gene:	0.950584	0.0143543	0.120079	-0.00485827	0.7816	0.00954494
ch.4.27504789F	Location:chr4 27895691 CpG Island: Open sea Related Gene:	0.950596	0.010786	0.172793	-0.00399807	0.361049	0.00260969
cg09425247	Location:chr10 52178225 CpG Island: Shore Related Gene: SGMS1	0.950885	0.0142073	-0.0636153	-0.00349724	0.252067	0.00172077
cg22286978	Location:chr19 58858806 CpG Island: Island Related Gene: A1BG CTD-2619J13.8 A1BG-AS1	0.951478	0.0381678	0.0677108	-0.00994882	0.158318	0.00274152
ch.1.194862R	Location:chr1 5228961 CpG Island: Open sea Related Gene:	0.951522	0.00740018	0.231407	-0.00751737	0.581766	0.00318644
cg13341720	Location:chr19 6740888 CpG Island: Shelf Related Gene: TRIP10	0.951768	0.0126817	0.033887	0.000428855	0.150361	0.00177123

https://ngdc.cncb.ac.cn/ewas/datahub/exploration

EWAS Open Platform-图谱查询

A1BG相关位点在肝脏liver中特异性的高甲基化

EWAS Data Hub / 特异甲基化评估——组织特异高甲基化

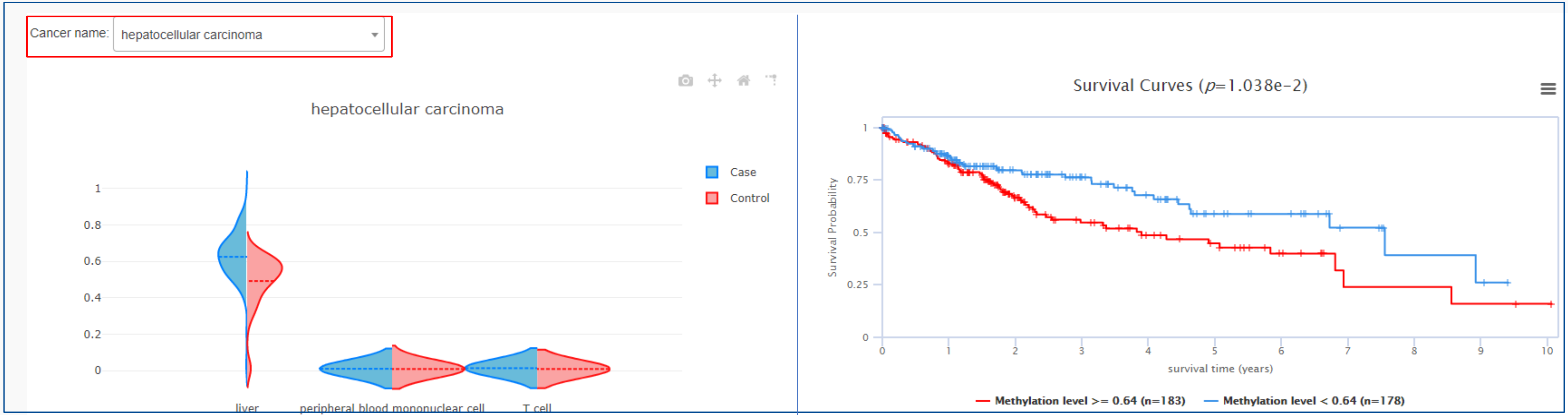


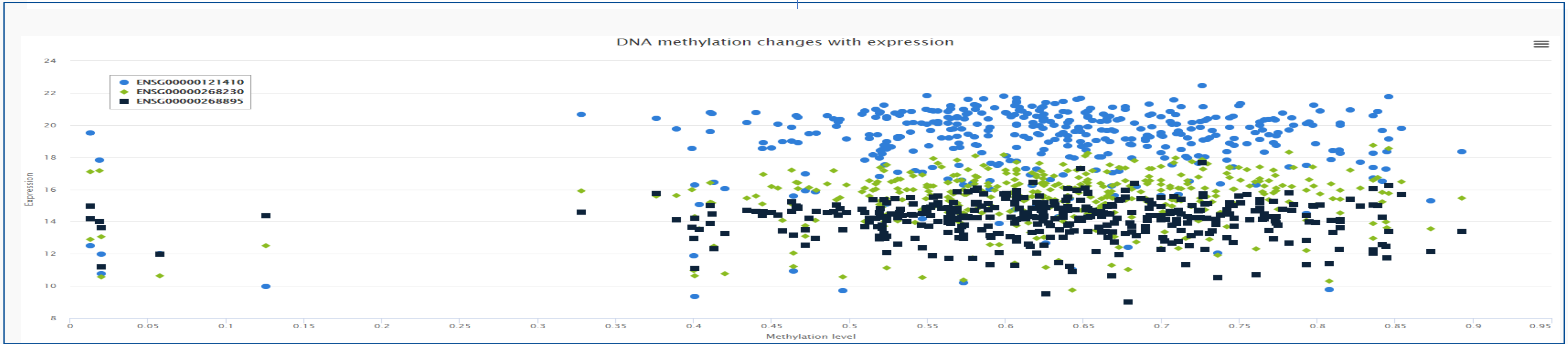
https://ngdc.cncb.ac.cn/ewas/datahub/probe/cg22286978

EWAS Open Platform-图谱查询

肝癌患者相比正常样本的A1BG相关位点甲基化升高，并且甲基化升高的样本的生存结果更差。

EWAS Data Hub / 特异甲基化评估——组织特异高甲基化


Cancer name: hepatocellular carcinoma



EWAS Open Platform-图谱查询

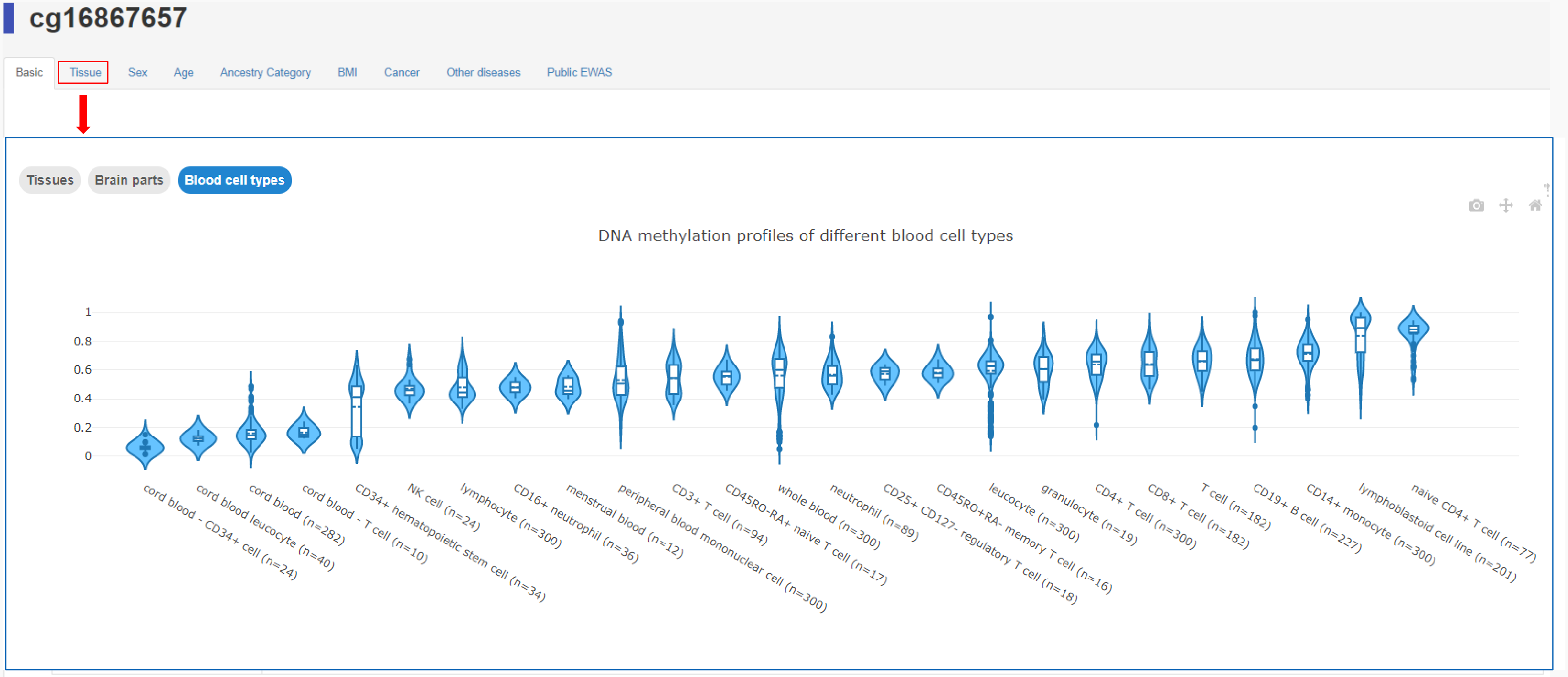
- 示例问题6：如何查看关注的甲基化位点和基因的多模态甲基化图谱?

探针/基因

- 基本模块
- 组织模块
- 年龄模块
- 性别模块
- 种族模块
- BMI模块
- 肿瘤模块
- 疾病模块
- 文献模块

EWAS Open Platform-图谱查询

EWAS Data Hub / 多模态DNA甲基化图谱

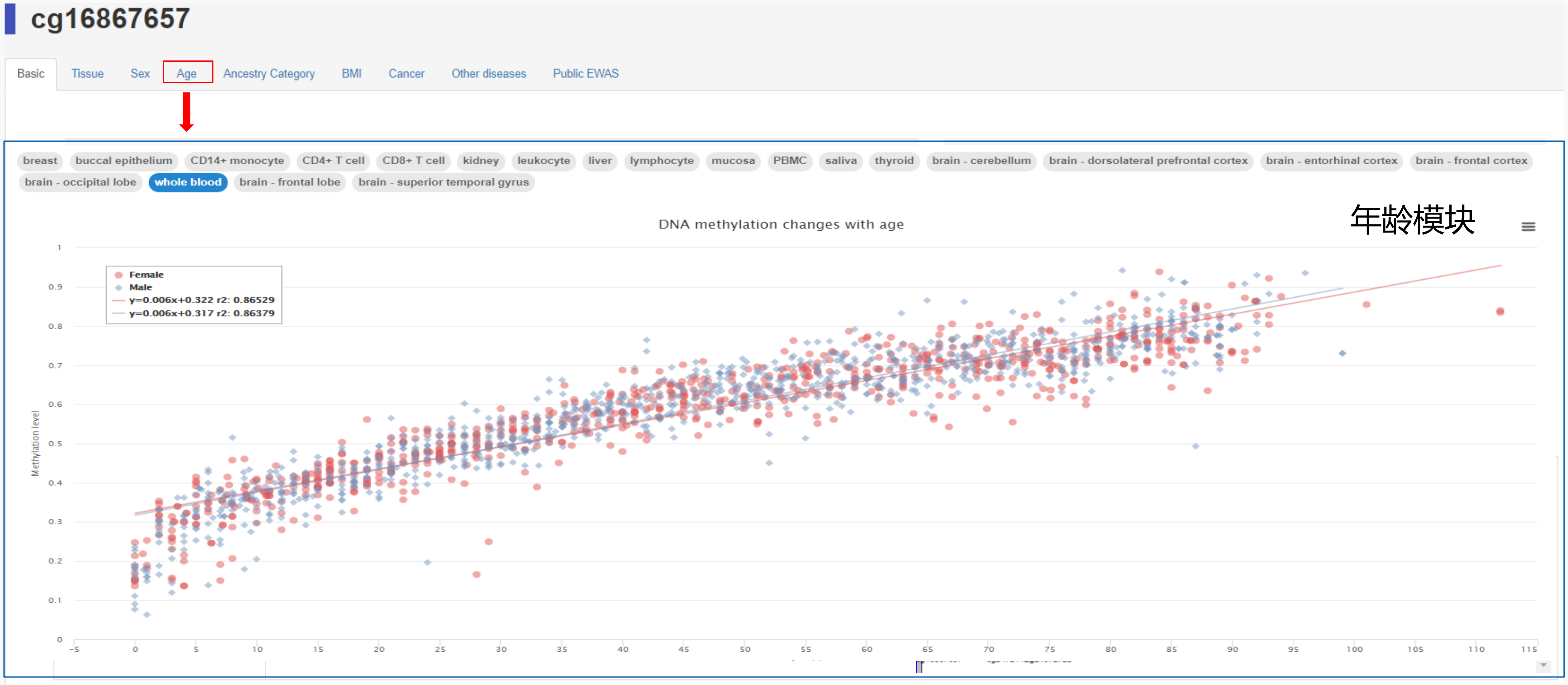


cg16867657

https://ngdc.cncb.ac.cn/ewas/datahub/probe/cg16867657

EWAS Open Platform-图谱查询

EWAS Data Hub / 多模态DNA甲基化图谱



年龄模块

https://ngdc.cncb.ac.cn/ewas/datahub/probe/cg16867657

EWAS Open Platform-图谱查询

EWAS Data Hub / 多模态DNA甲基化图谱

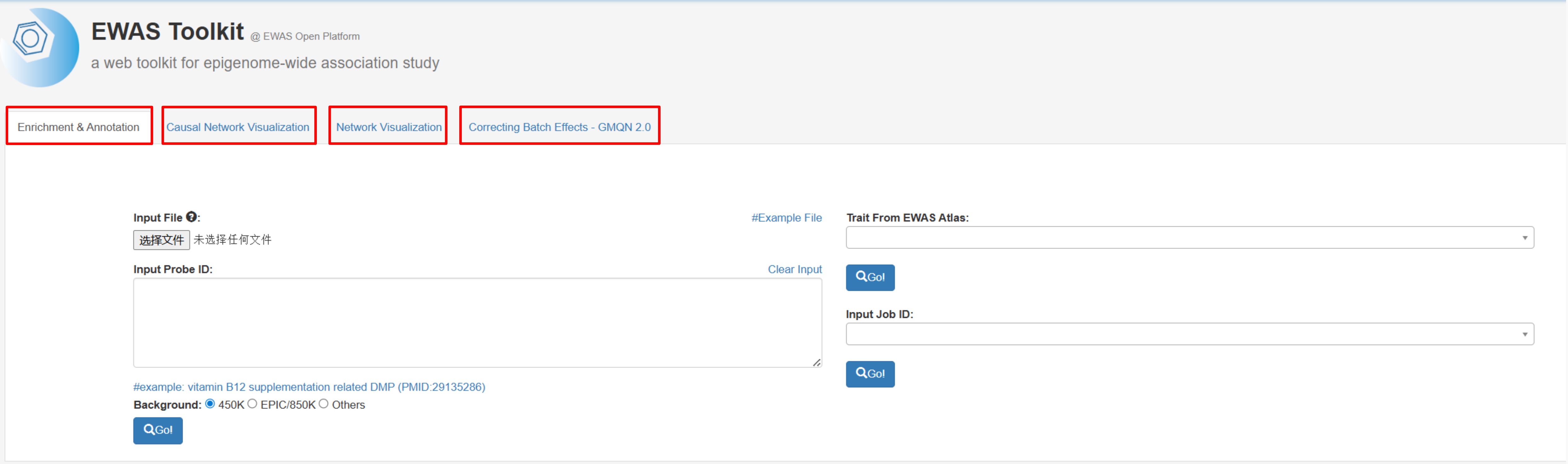
cg16867657

Cancer name: acute myeloid leukemia

肿瘤模块

https://ngdc.cncb.ac.cn/ewas/datahub/probe/cg16867657

EWAS Open Platform-工具使用



EWAS Toolkit @ EWAS Open Platform

a web toolkit for epigenome-wide association study

Enrichment & Annotation | Causal Network Visualization | Network Visualization | Correcting Batch Effects - GMQN 2.0

Input File ❓: #Example File

选择文件 未选择任何文件

Input Probe ID: Clear Input

#example: vitamin B12 supplementation related DMP (PMID:29135286)

Background: ● 450K ○ EPIC/850K ○ Others

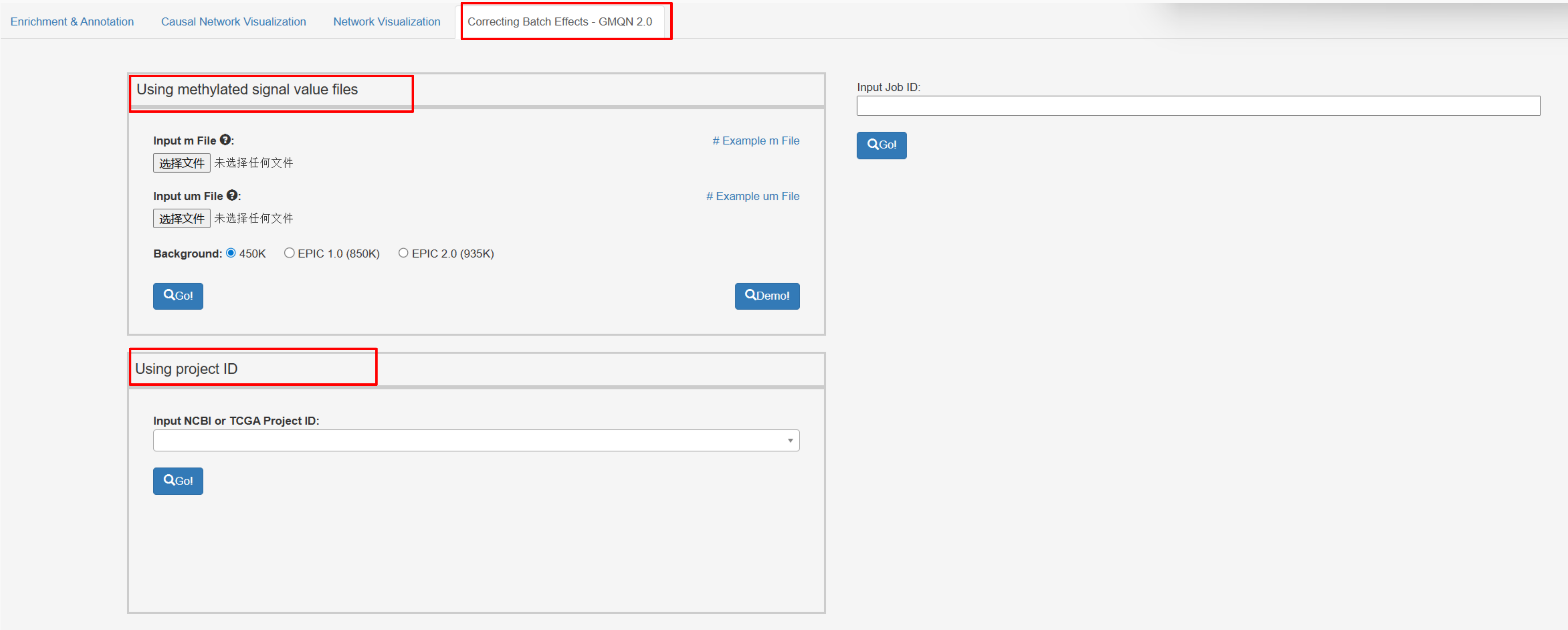
Trait From EWAS Atlas:

Input Job ID:

https://ngdc.cncb.ac.cn/ewas/toolkit

EWAS Open Platform-工具使用

支持两种输入文件（甲基化信号数据、项目ID）的在线去批次效应工具



Using methylated signal value files

Input m File ?: 选择文件 未选择任何文件

Input um File ?: 选择文件 未选择任何文件

Background: ● 450K ○ EPIC 1.0 (850K) ○ EPIC 2.0 (935K)

Using project ID

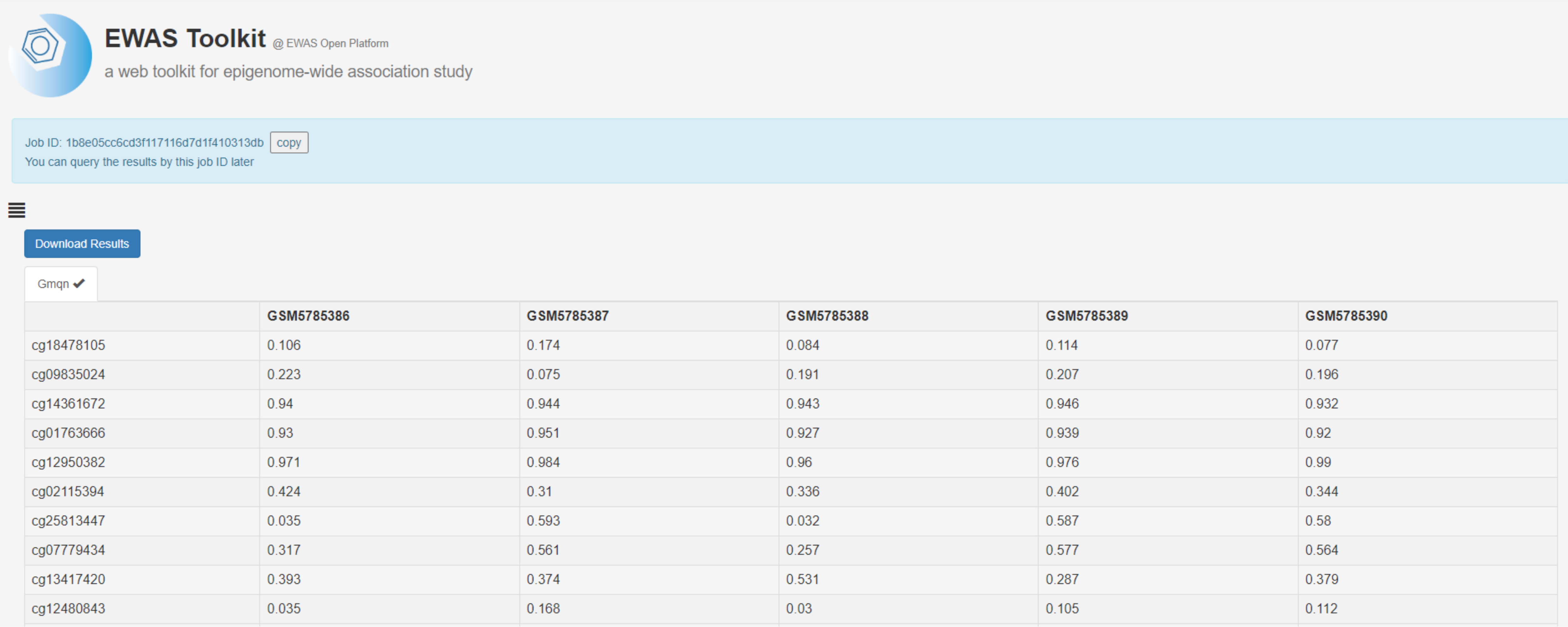
Input NCBI or TCGA Project ID:

Input Job ID:

https://ngdc.cncb.ac.cn/ewas/toolkit

EWAS Open Platform-工具使用

在线去批次效应工具结果表的查看和便捷化下载



EWAS Toolkit @ EWAS Open Platform

a web toolkit for epigenome-wide association study

Job ID: 1b8e05cc6cd3f117116d7d1f410313db

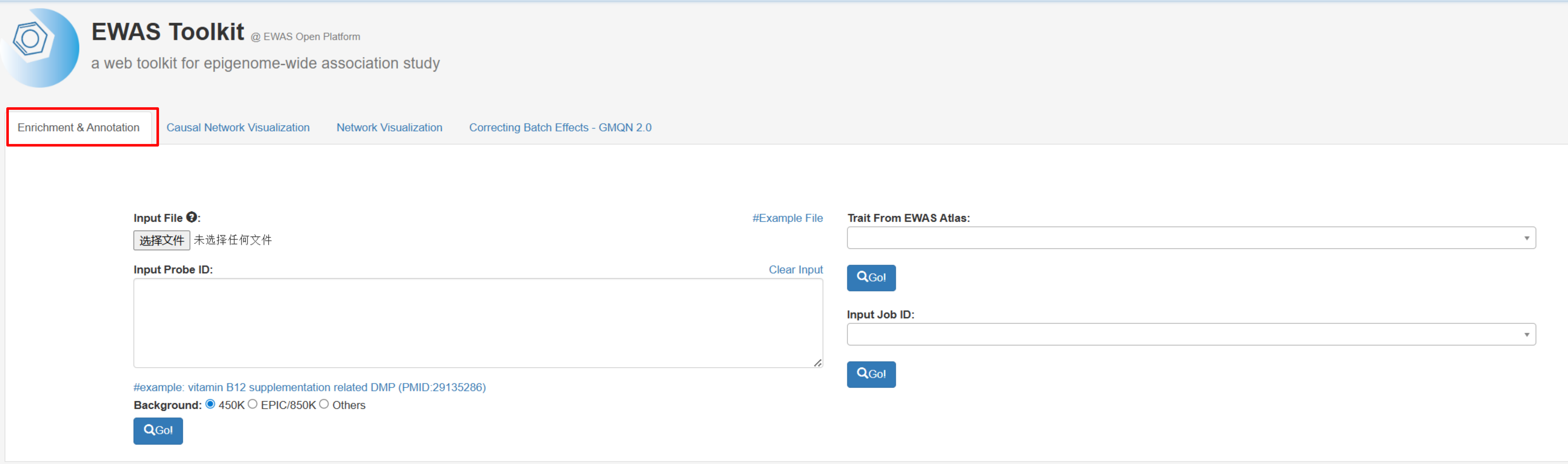
You can query the results by this job ID later

	GSM5785386	GSM5785387	GSM5785388	GSM5785389	GSM5785390
cg18478105	0.106	0.174	0.084	0.114	0.077
cg09835024	0.223	0.075	0.191	0.207	0.196
cg14361672	0.94	0.944	0.943	0.946	0.932
cg01763666	0.93	0.951	0.927	0.939	0.92
cg12950382	0.971	0.984	0.96	0.976	0.99
cg02115394	0.424	0.31	0.336	0.402	0.344
cg25813447	0.035	0.593	0.032	0.587	0.58
cg07779434	0.317	0.561	0.257	0.577	0.564
cg13417420	0.393	0.374	0.531	0.287	0.379
cg12480843	0.035	0.168	0.03	0.105	0.112

https://ngdc.cncb.ac.cn/ewas/toolkit

EWAS Open Platform-工具使用

甲基化位点集的在线化富集和注释功能

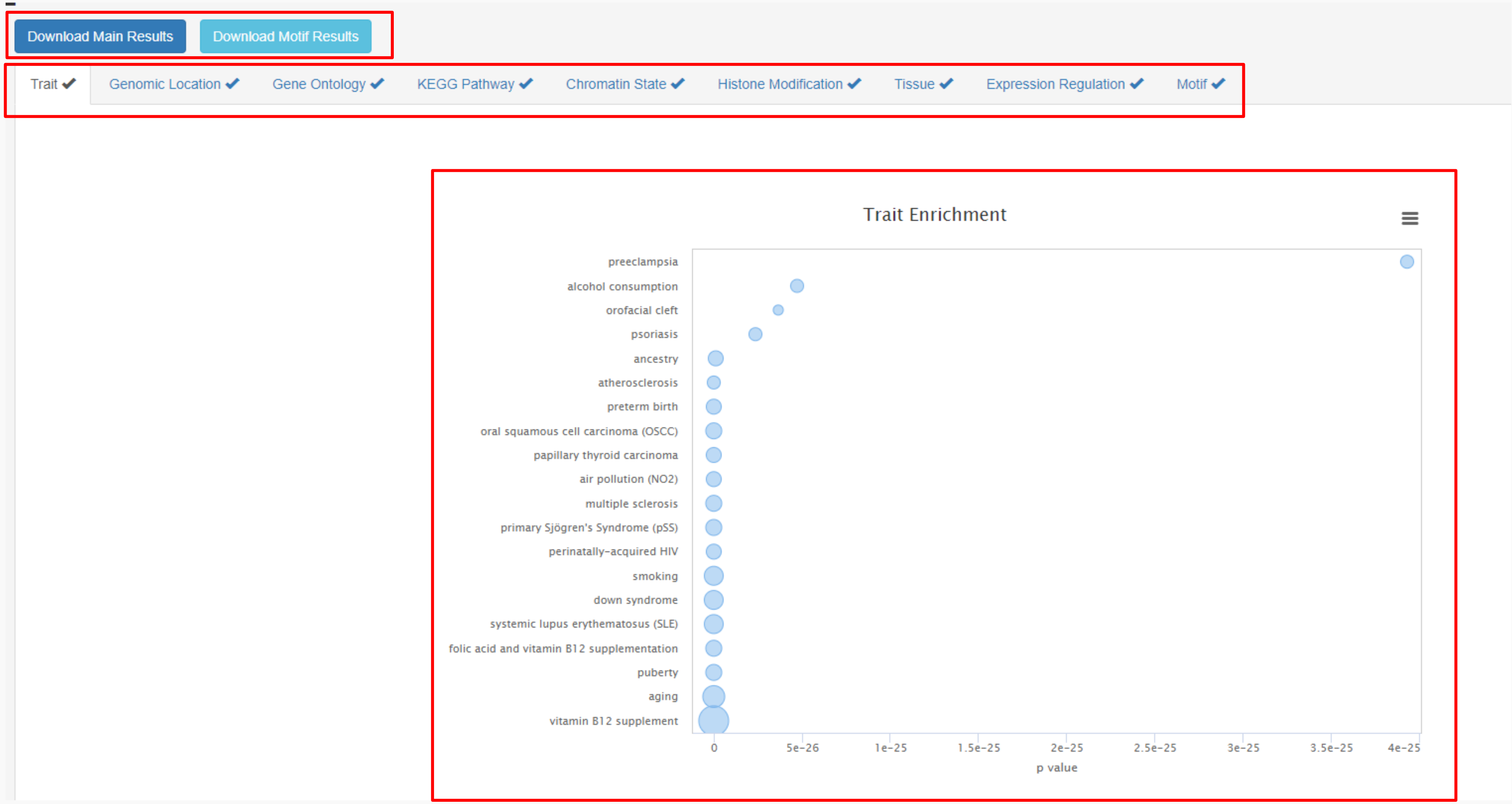


Background: ● 450K ○ EPIC/850K ○ Others

https://ngdc.cncb.ac.cn/ewas/toolkit

EWAS Open Platform-工具使用

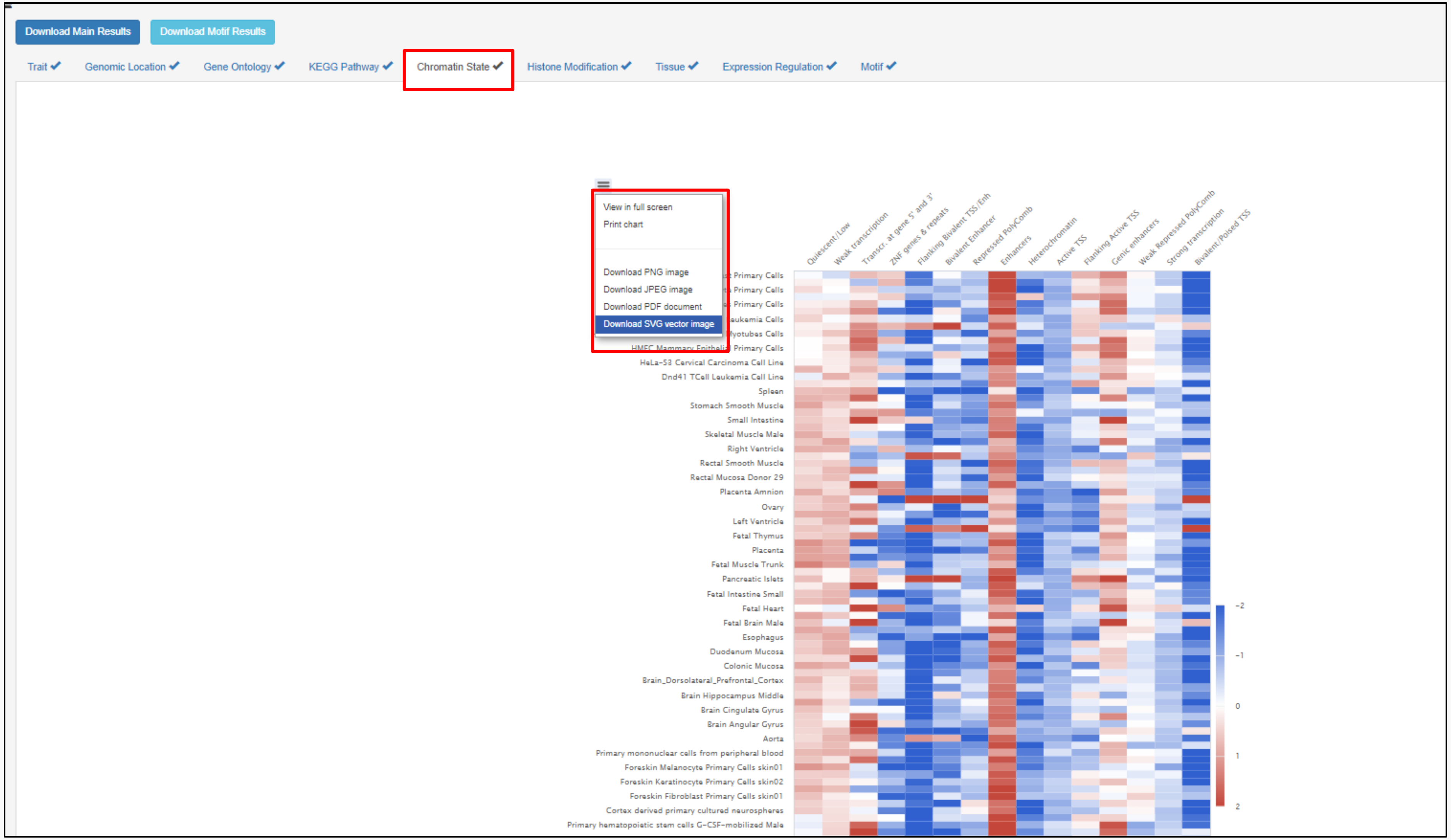
EWAS Toolkit / Enrichment & Annotation



https://ngdc.cncb.ac.cn/ewas/toolkit

EWAS Open Platform-工具使用

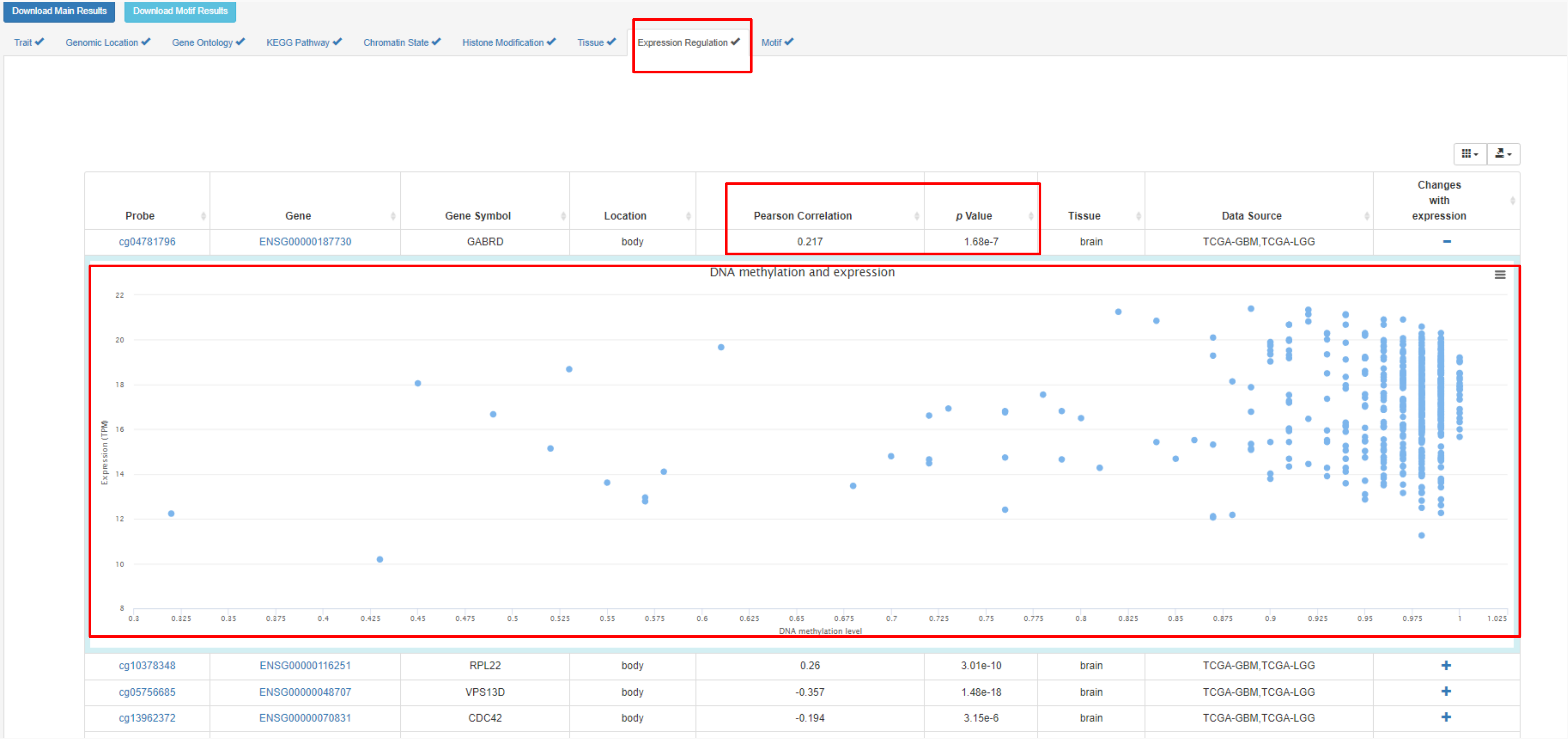
EWAS Toolkit / Enrichment & Annotation



https://ngdc.cncb.ac.cn/ewas/toolkit

EWAS Open Platform-工具使用

EWAS Toolkit / Enrichment & Annotation

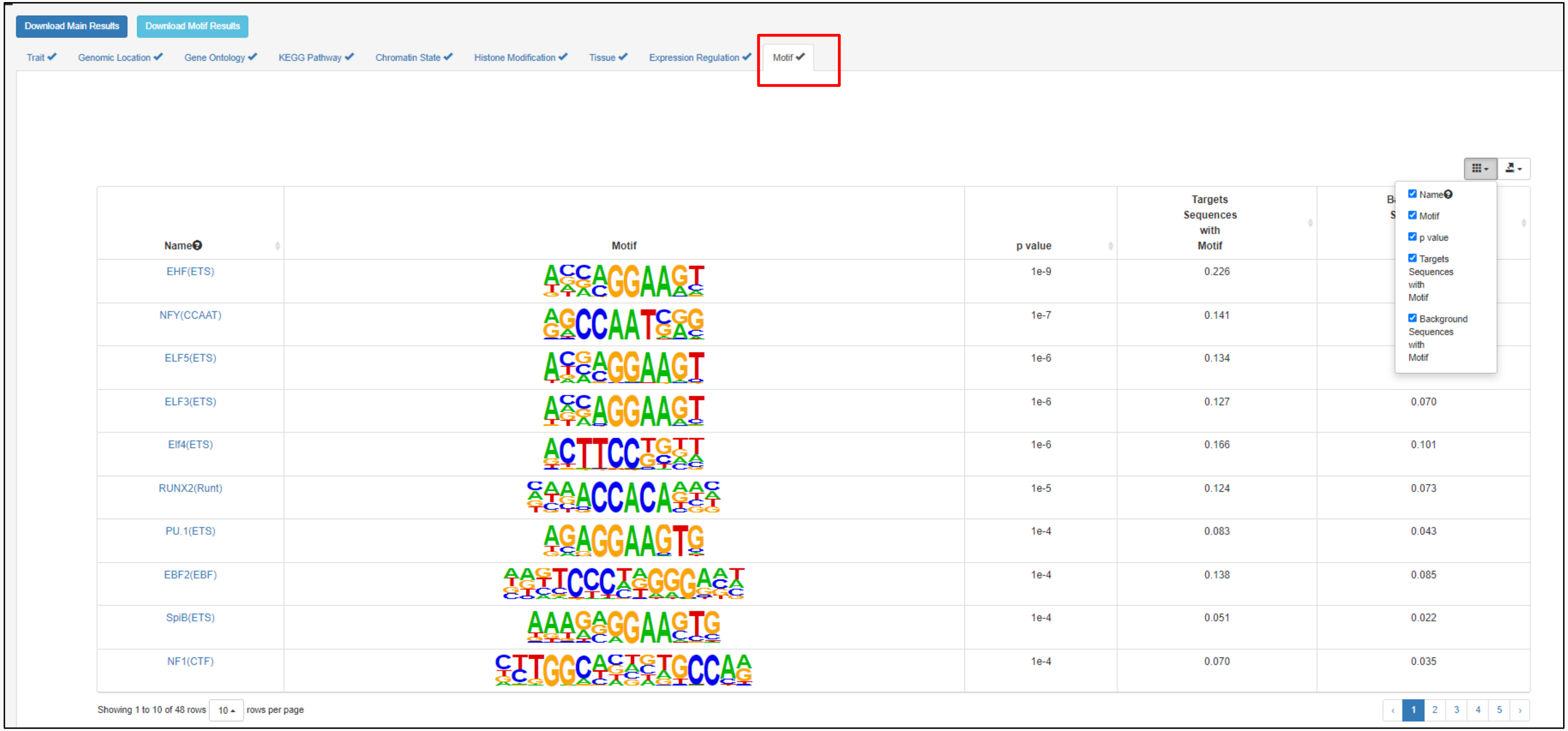


Probe	Gene	Gene Symbol	Location	Pearson Correlation	*p* Value	Tissue	Data Source	Changes with expression
cg04781796	ENSG00000187730	GABRD	body	0.217	1.68e-7	brain	TCGA-GBM,TCGA-LGG	−
cg10378348	ENSG00000116251	RPL22	body	0.26	3.01e-10	brain	TCGA-GBM,TCGA-LGG	+
cg05756685	ENSG00000048707	VPS13D	body	-0.357	1.48e-18	brain	TCGA-GBM,TCGA-LGG	+
cg13962372	ENSG00000070831	CDC42	body	-0.194	3.15e-6	brain	TCGA-GBM,TCGA-LGG	+

https://ngdc.cncb.ac.cn/ewas/toolkit

EWAS Open Platform-工具使用

EWAS Toolkit / Enrichment & Annotation



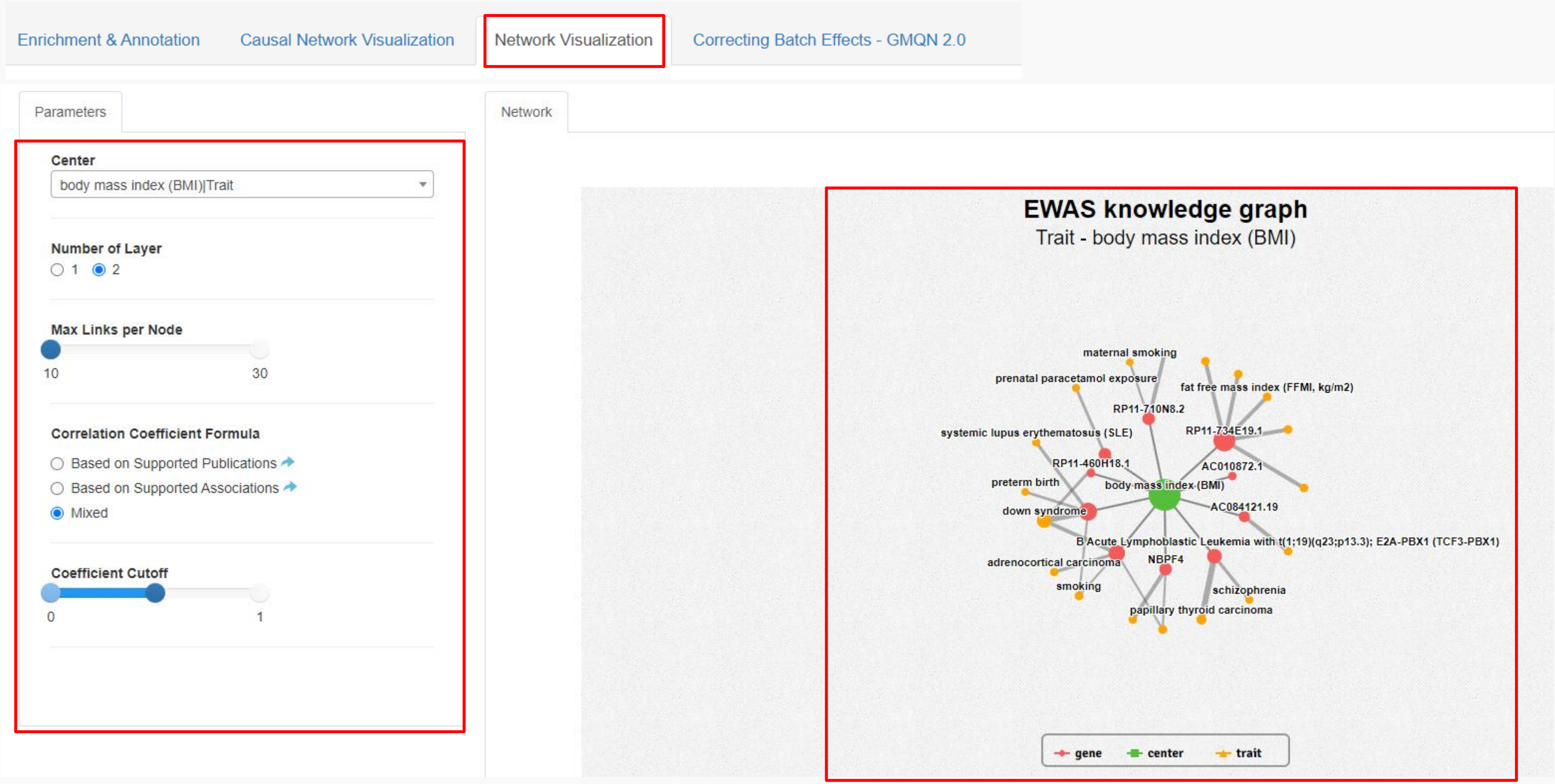
☑ Name
☑ Motif
☑ p value
☑ Targets Sequences with Motif
☑ Background Sequences with Motif

Name	Motif	p value	Targets Sequences with Motif	B S
EHF(ETS)		1e-9	0.226	
NFY(CCAAT)		1e-7	0.141	
ELF5(ETS)		1e-6	0.134	
ELF3(ETS)		1e-6	0.127	0.070
Elf4(ETS)		1e-6	0.166	0.101
RUNX2(Runt)		1e-5	0.124	0.073
PU.1(ETS)		1e-4	0.083	0.043
EBF2(EBF)		1e-4	0.138	0.085
SpiB(ETS)		1e-4	0.051	0.022
NF1(CTF)		1e-4	0.070	0.035

Showing 1 to 10 of 48 rows 10 rows per page

https://ngdc.cncb.ac.cn/ewas/toolkit

EWAS Open Platform-工具使用



Center
body mass index (BMI)|Trait

Number of Layer
○ 1 ● 2

Max Links per Node
10 30

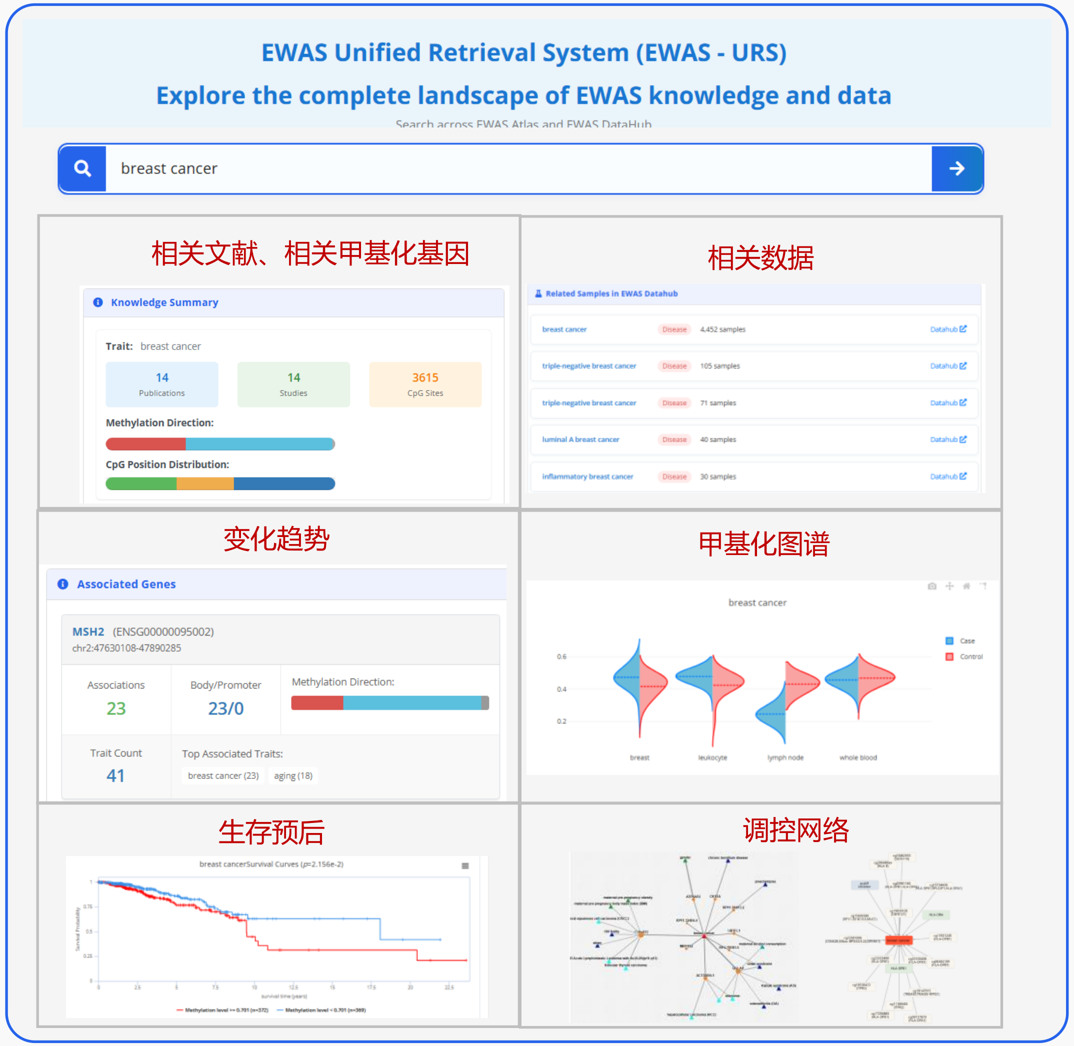
Correlation Coefficient Formula
○ Based on Supported Publications
○ Based on Supported Associations
● Mixed

Coefficient Cutoff
0 1

https://ngdc.cncb.ac.cn/ewas/toolkit

EWAS Open Platform-融合检索

构建深度融合数据、知识与工具的全局检索系统，提供综合性、精炼化、可溯源的结果



EWAS Unified Retrieval System (EWAS - URS)

Explore the complete landscape of EWAS knowledge and data

Search across EWAS Atlas and EWAS DataHub

breast cancer

相关文献、相关甲基化基因

Knowledge Summary

Trait: breast cancer

14	14	3615
Publications	Studies	CpG Sites

Methylation Direction:

CpG Position Distribution:

相关数据

Related Samples in EWAS Datahub

breast cancer	Disease	4,452 samples	Datahub
triple-negative breast cancer	Disease	105 samples	Datahub
triple-negative breast cancer	Disease	71 samples	Datahub
luminal A breast cancer	Disease	40 samples	Datahub
inflammatory breast cancer	Disease	30 samples	Datahub

变化趋势

Associated Genes

MSH2 (ENSG00000095002)
chr2:47630108-47890285

Associations: 23

Body/Promoter: 23/0

Methylation Direction:

Trait Count: 41

Top Associated Traits: breast cancer (23) aging (18)

甲基化图谱

生存预后

调控网络

基因、甲基化、疾病 、环境因子、表型

- 提供数据和知识的综合性多模块展示
  - 总结
  - EWAS研究
  - 甲基化和表达
  - 疾病图谱
  - 文献

EWAS Open Platform-智能问答

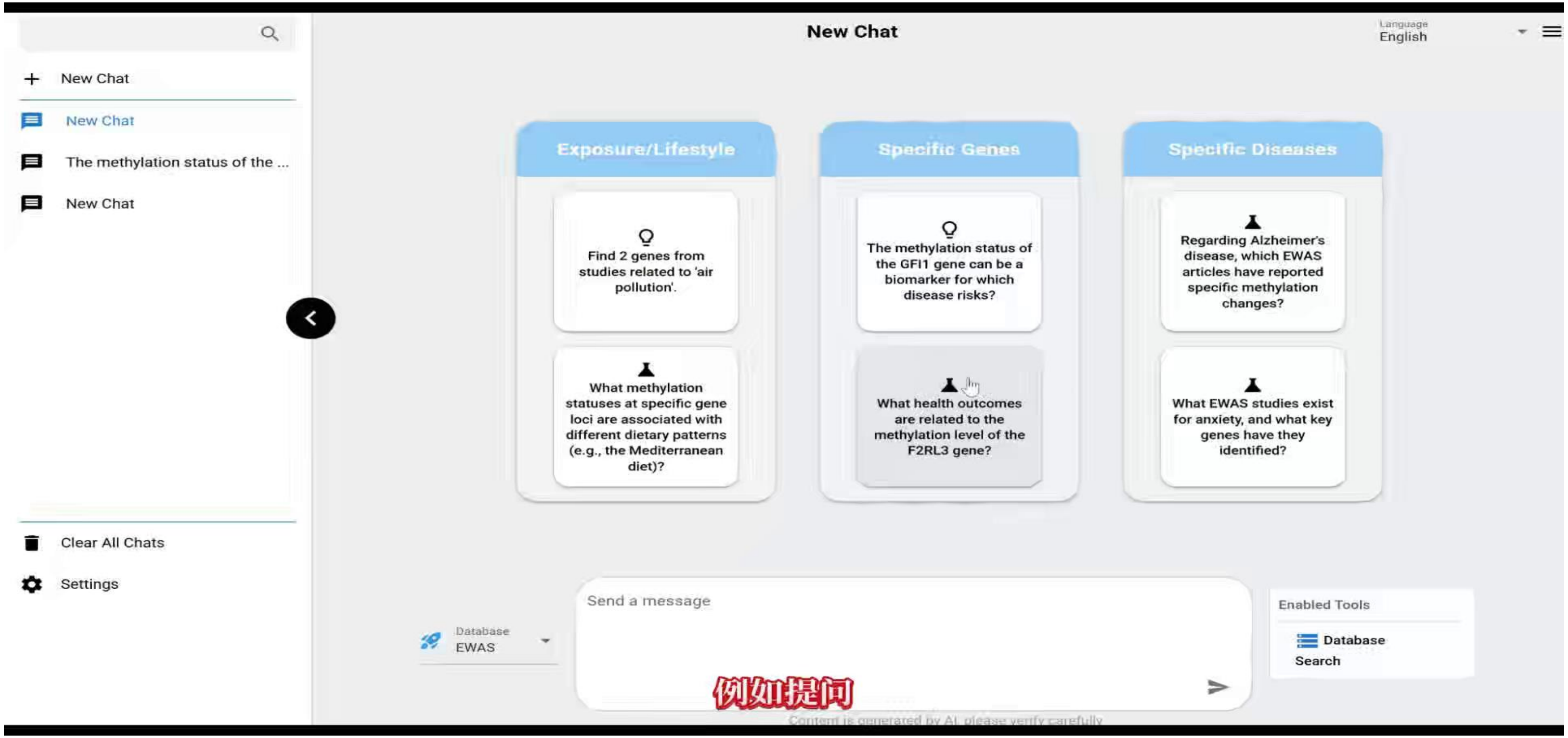
为使研究人员能更灵活、无障碍、直观地获取复杂性状（含疾病）的表观信息，上线了智能问答助手，支持自然语言对话查询，可基于平台整合的EWAS资源自动返回结构化答案

Click here for Intelligent Q&A!

Your smart assistant awaits

根据您的查询，我在表观关联研究(EWAS)数据库中找到了与空气污染(包括PM2.5暴露)相关的 11,133 个基因和13,851个甲基化位点。

这些基因和位点在 40 项研究中被识别出来，涉及 26 种不同的空气污染相关性状。



Find 2 genes from studies related to 'air pollution'.

What methylation statuses at specific gene loci are associated with different dietary patterns (e.g., the Mediterranean diet)?

The methylation status of the GFI1 gene can be a biomarker for which disease risks?

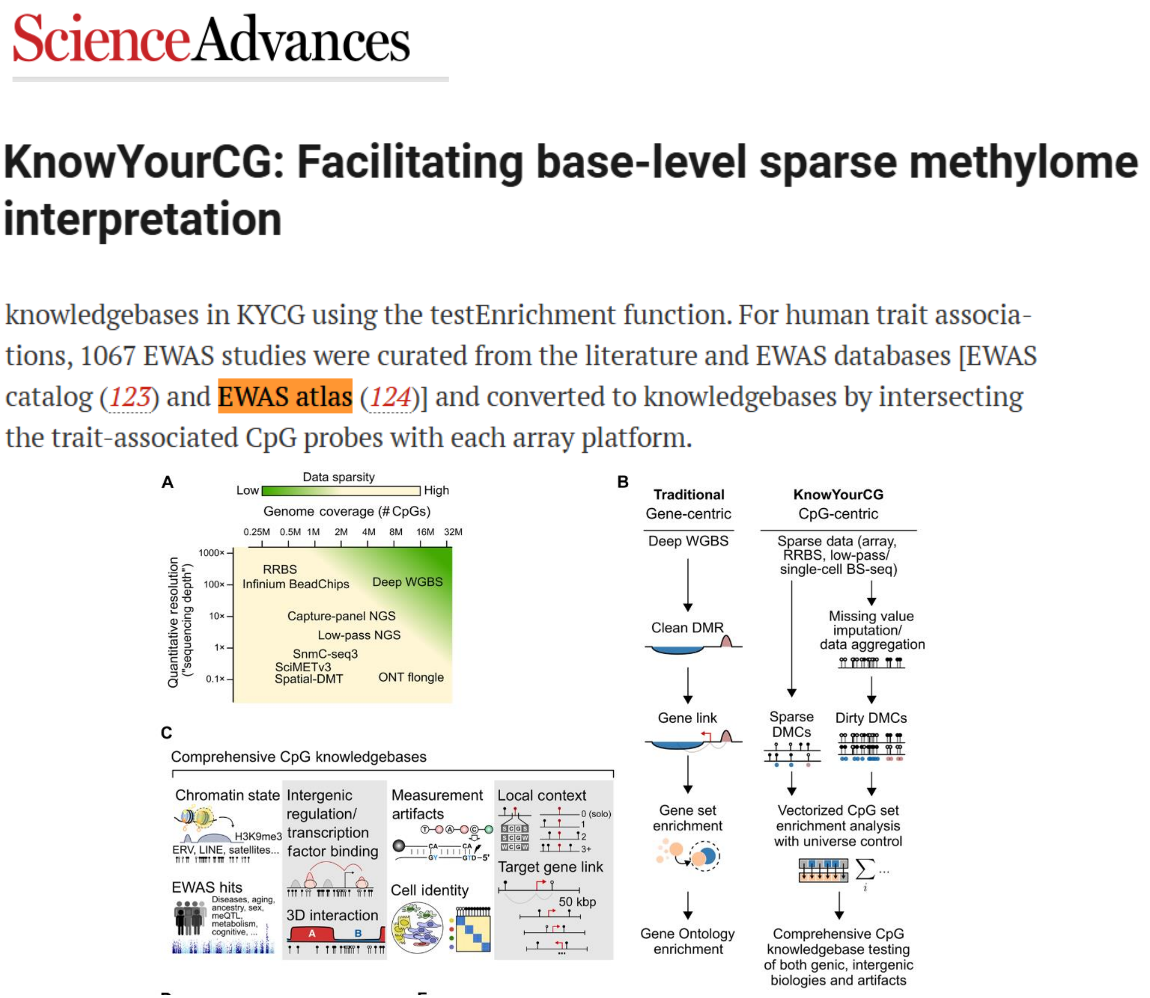
What health outcomes are related to the methylation level of the F2RL3 gene?

Regarding Alzheimer's disease, which EWAS articles have reported specific methylation changes?

What EWAS studies exist for anxiety, and what key genes have they identified?

EWAS Open Platform用户使用案例 1

EWAS Atlas 为KnowYourCG工具提供了系统性的上游知识支撑



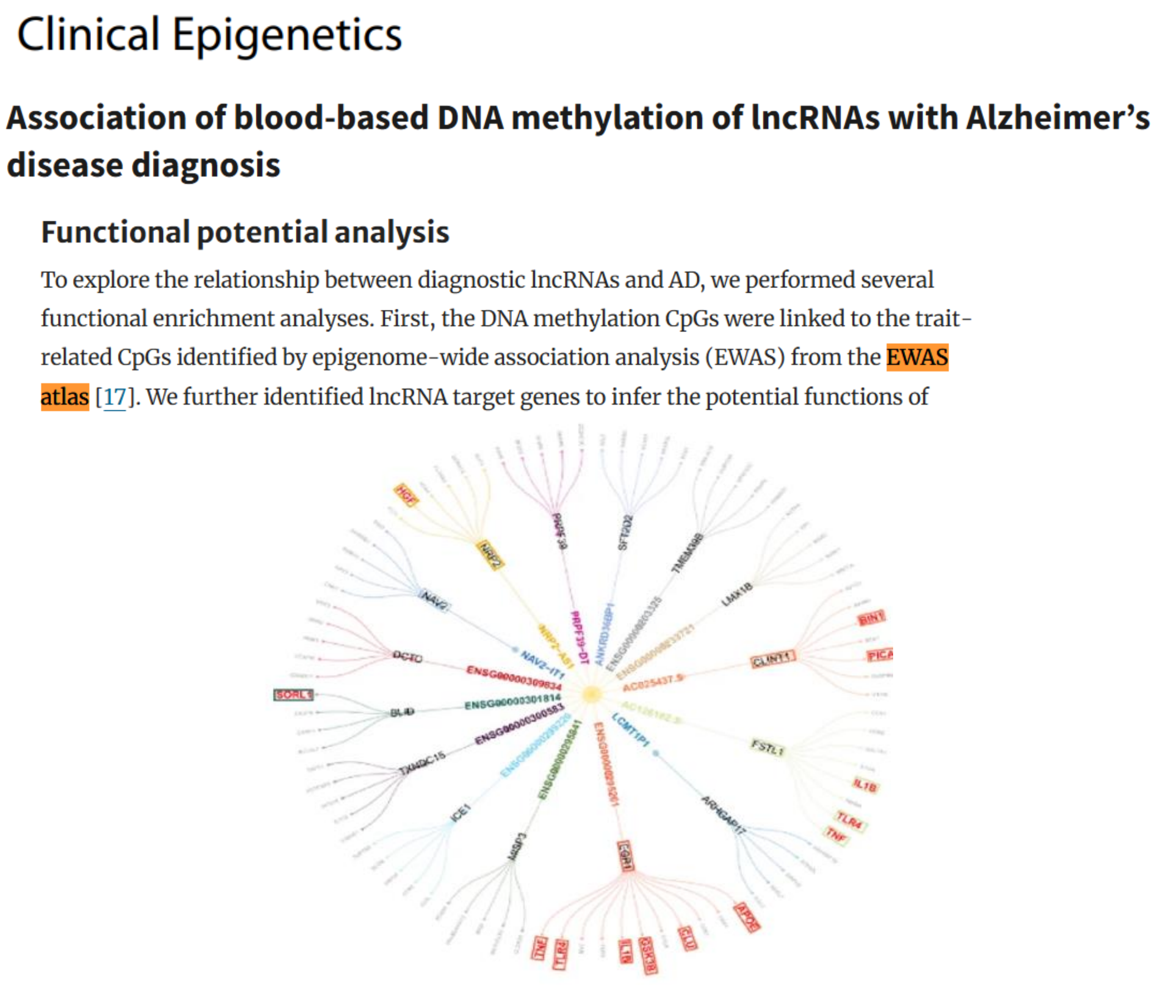
ScienceAdvances

KnowYourCG: Facilitating base-level sparse methylome interpretation

knowledgebases in KYCG using the testEnrichment function. For human trait associations, 1067 EWAS studies were curated from the literature and EWAS databases [EWAS catalog (*123*) and EWAS atlas (*124*)] and converted to knowledgebases by intersecting the trait-associated CpG probes with each array platform.

直接进行碱基层面筛查的DNA甲基化数据解读框架，通过快速匹配海量知识库，有效破解各类稀疏甲基化数据，为生物学机制研究和检测技术优化提供关键见解 Science Advances (2025)

EWAS Atlas为阿兹海默症的早诊生物标志物研究提供支持



Clinical Epigenetics

Association of blood-based DNA methylation of lncRNAs with Alzheimer's disease diagnosis

Functional potential analysis

To explore the relationship between diagnostic lncRNAs and AD, we performed several functional enrichment analyses. First, the DNA methylation CpGs were linked to the trait-related CpGs identified by epigenome-wide association analysis (EWAS) from the EWAS atlas [17]. We further identified lncRNA target genes to infer the potential functions of

血液中发现一组与阿尔茨海默病密切相关的lncRNA，基于此构建出高精度的诊断模型，为AD的无创早期诊断提供了新型血液潜在生物标志物，并揭示了疾病发生的新分子机制。linical Epigenetics (2025)

EWAS Open Platform用户使用案例 2

为国内外的多个面向人口健康与疾病预测的AI模型提供高质量数据支撑

基于高质量人群DNA甲基化数据集训练的 单样本甲基化填补模型 OSMI

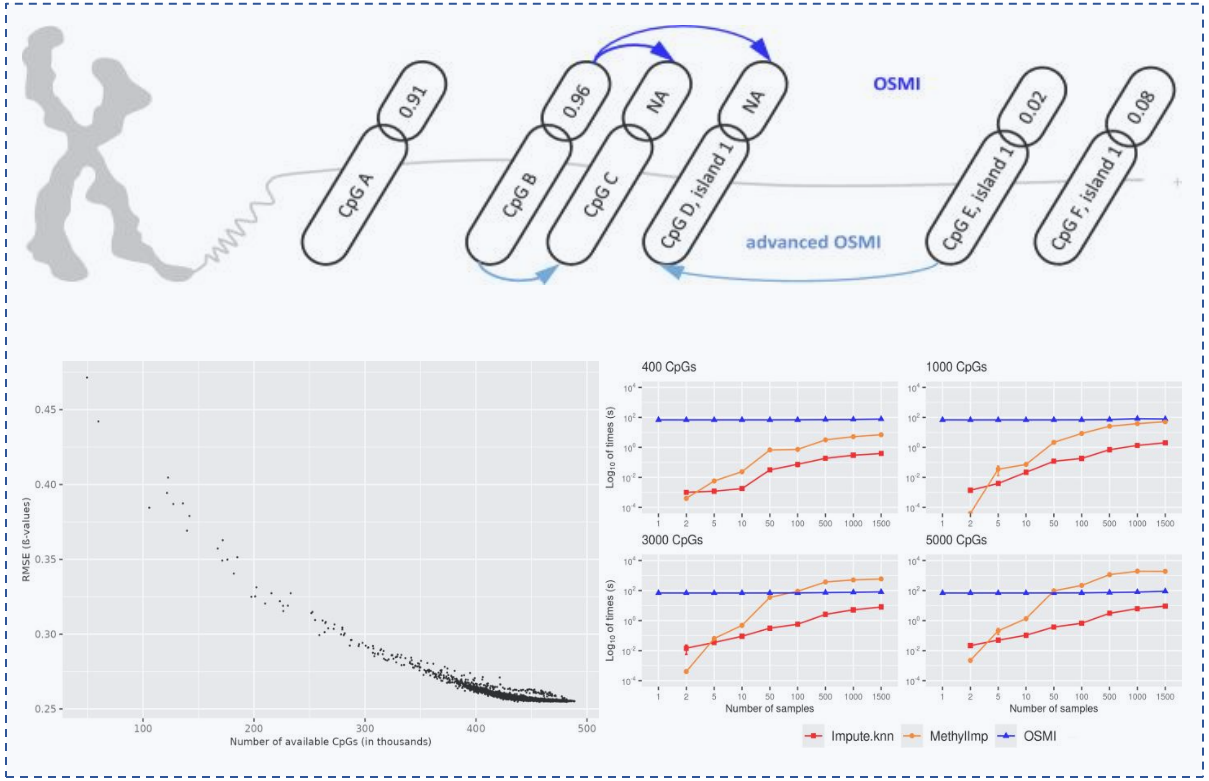
□ 提高了插补精度，极大降低了因缺失数据导致的重复检测和实验成本

□ 对内存和计算量的要求较低，可促进了个性化医疗方案的普及

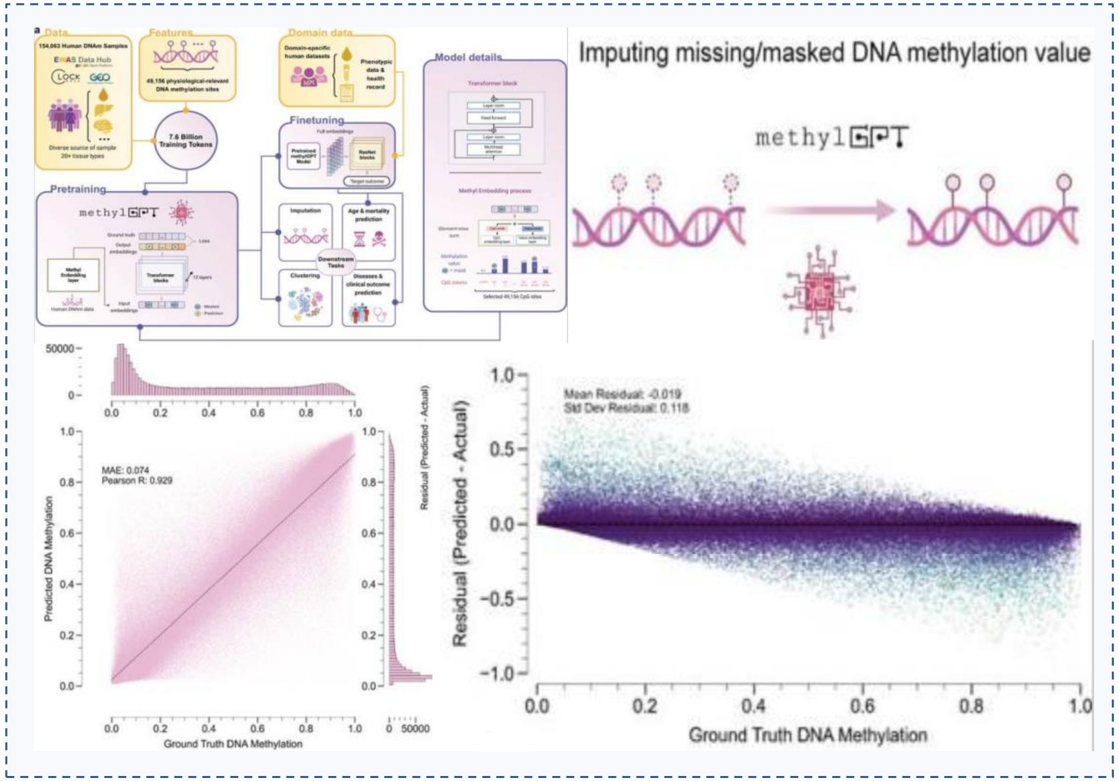
基于高质量人群DNA甲基化数据集训练的 DNA 甲基化组的基础模型MethylGPT

□ 数据缺失70%的下游任务中保持稳定性能，大大降低检测和试验成本

□ 能够系统评估干预对疾病风险的影响，展示了临床应用的潜力



在单个受试者案例中产生了RMSE = 0.2713的平均插补精度
（95%可信区间为0.2696至0.2730）
BMC Bioinformatics 31, 26:143 (2025)



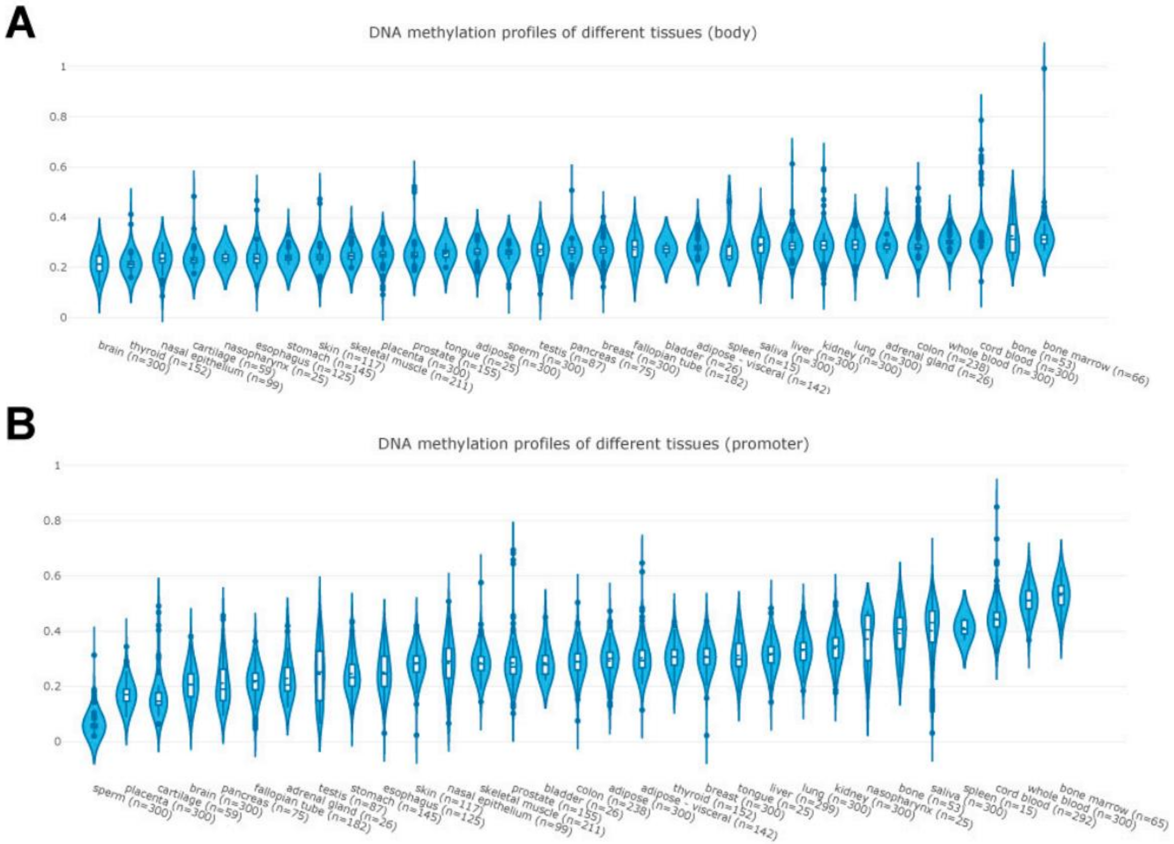
模型表现出稳健的甲基化值预测（Pearson R=0.929）
bioRxiv 4:2024.10.30.621013 (2024)

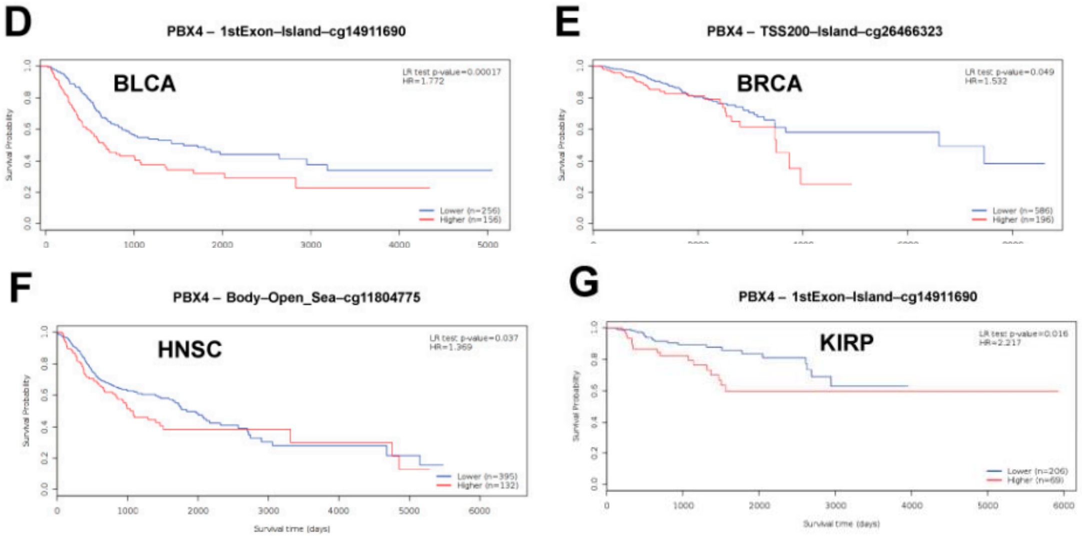
EWAS Open Platform用户使用案例 3

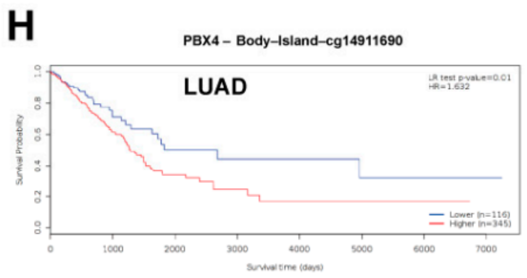
Title: Identifying the Potential Roles of PBX4 in Human Cancers Based on Integrative Analysis.

PBX4 是参与多种病理生理过程的 HOX 蛋白的转录辅助因子，前期研究已表明PBX4 的失调与多种疾病密切相关，但多种癌症中PBX4 的作用研究仍然不足，因此，作者在33种癌症中进行了PBX4相关的泛癌分析以深入探究其潜在作用。

其中，作者通过直接使用EWAS Open Platform中的不同组织的DNA甲基化分析图谱和不同癌症中的生存分析图，发现PBX4的甲基化主要发生在骨骼、血液、肺、肾和肝组织中，膀胱癌（BLCA）、乳腺癌（BRCA）、头颈癌(HNSC)、肾癌（KIRP） 和 肺腺癌(LUAD) 的 PBX4 高甲基化水平与不良预后显著相关。





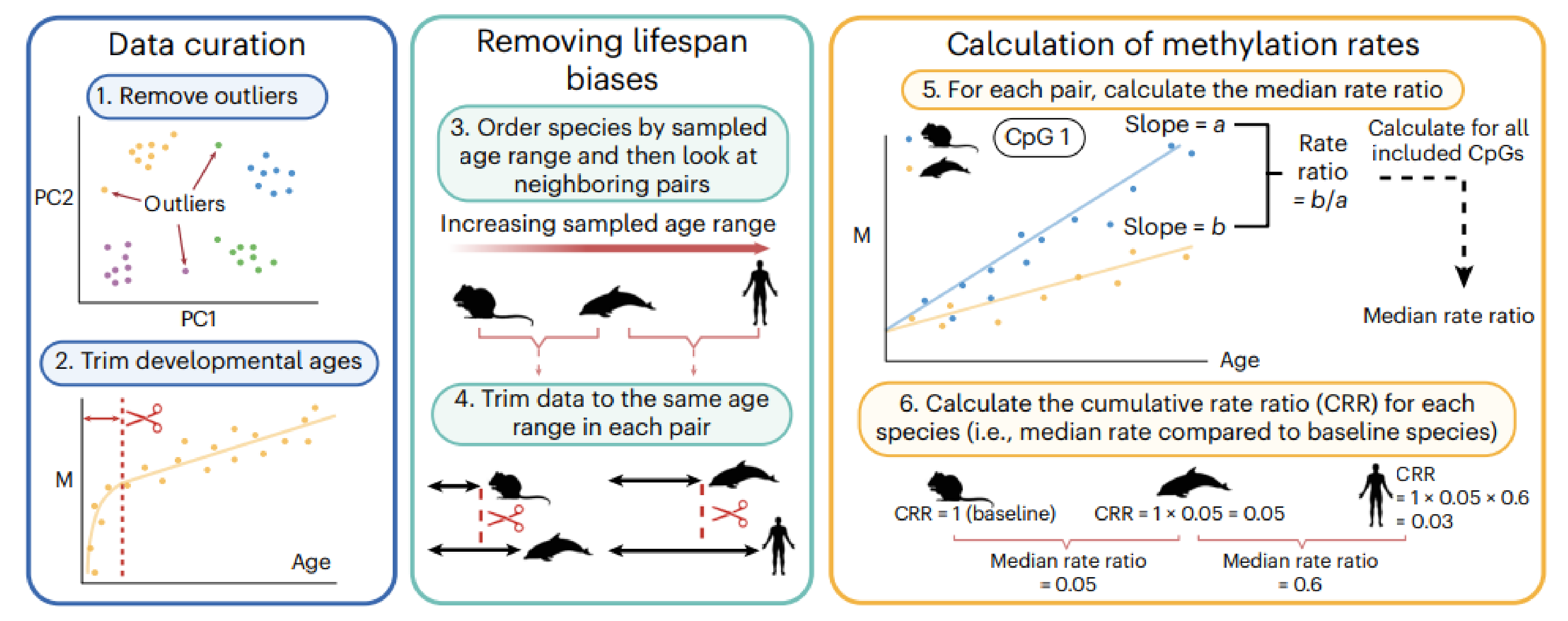


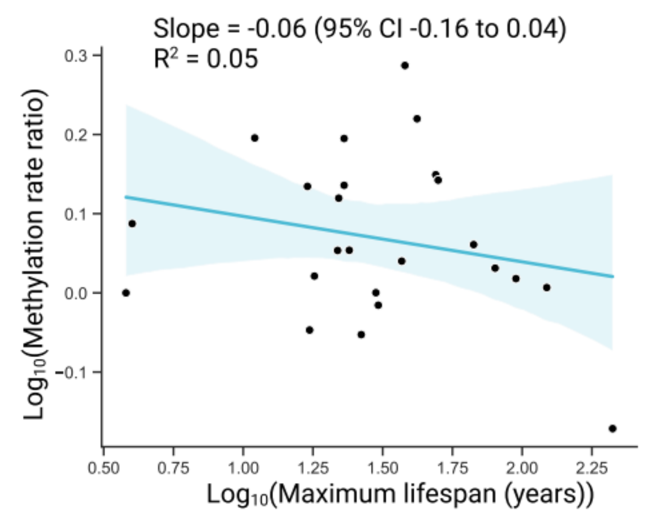
Song et al. *Biomolecules*. 2022

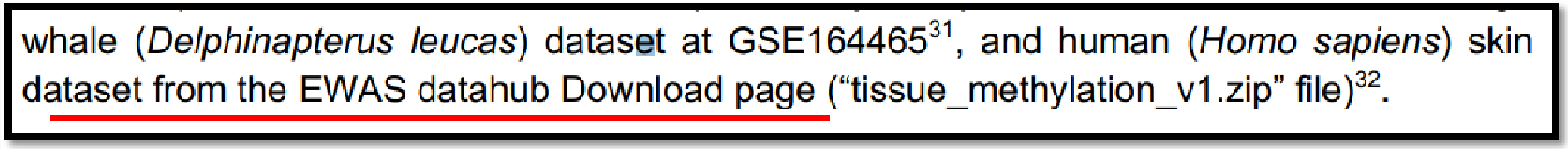
EWAS Open Platform用户使用案例 4

Title: DNA methylation rates scale with maximum lifespan across mammals.

DNA甲基化变化速率先前已被发现广泛与哺乳动物的最大寿命相关，但没有观察到精确的关系。作者开发了一个方法用来比较哺乳动物中保守年龄相关位点的甲基化率。**通过直接使用EWAS Open Platform中整合的去批次效应后的皮肤组织的DNA甲基化数据**， 作者发现，DNA甲基化率与最大寿命呈负相关。甲基化变化速率可能与哺乳动物最大寿命的演化约束有关。






whale (*Delphinapterus leucas*) dataset at GSE164465[31], and human (*Homo sapiens*) skin dataset from the EWAS datahub Download page (“tissue_methylation_v1.zip” file)[32].

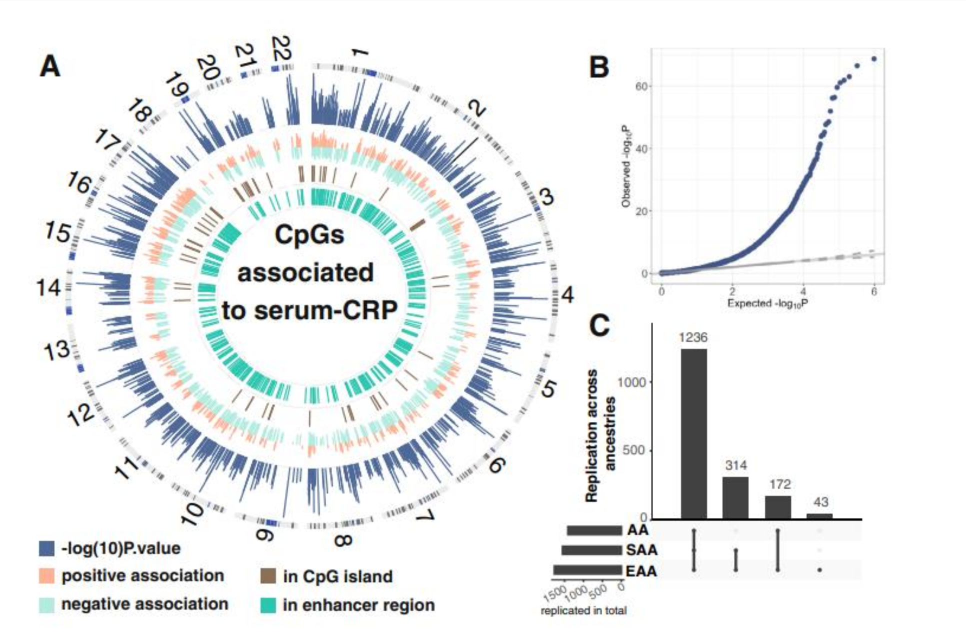
Samuel et al. ***Nature Aging***. 2024

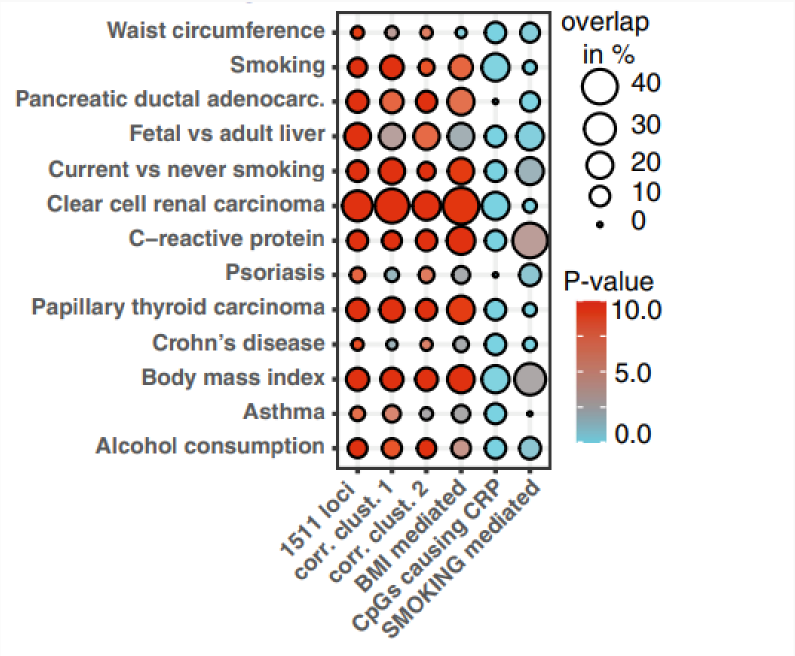
EWAS Open Platform用户使用案例 5

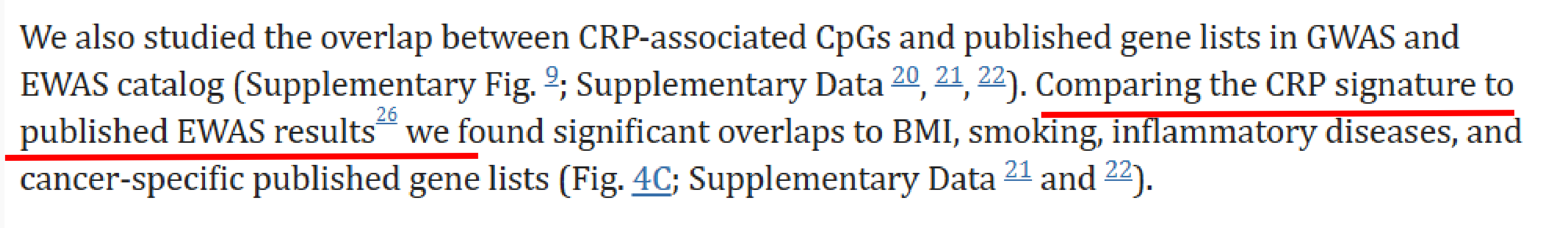
Title: DNA methylation signature of chronic low-grade inflammation and its role in cardio-respiratory diseases.

以 C 反应蛋白为标志的慢性炎症与甲基化变化有关，但因果关系尚不清楚。作者使用 22,774 人的队列对 C 反应蛋白水平进行了表观基因组范围的关联荟萃分析，发现这些甲基化变化可能是炎症的结果，并可能导致疾病。

其中，作者直接使用EWAS Open Platform中整合的表观知识图谱，发现C反应蛋白相关的甲基化基因与BMI、吸烟、炎症性疾病和癌症特异性已发表的甲基化基因列表有显著重叠，进一步侧面反映了C反应蛋白可能参与炎症和癌症过程。






We also studied the overlap between CRP-associated CpGs and published gene lists in GWAS and EWAS catalog (Supplementary Fig. 9; Supplementary Data 20, 21, 22). Comparing the CRP signature to published EWAS results[26] we found significant overlaps to BMI, smoking, inflammatory diseases, and cancer-specific published gene lists (Fig. 4C; Supplementary Data 21 and 22).

Matthias et al. *Nature Communications*. 2022

EWAS Open Platform用户使用案例 6

Title: Epigenome-wide meta-analysis identifies DNA methylation biomarkers associated with diabetic kidney disease.

1 型糖尿病影响全球超过 900 万人，作者评估了1304 个 1 型糖尿病患者和已知肾脏状况的血液来源全基因组 DNA 甲基化的差异。在 1 型糖尿病中鉴定了 32 个差异甲基化位点，**基于EWAS Open Platform的工具分析，发现**其与糖尿病肾病的病理特征相关。进一步研究表明，32 个 位点中有 21 个的甲基化可预测肾衰竭的发展，有助于识别出 1 型糖尿病中患糖尿病肾病风险更高的个体。

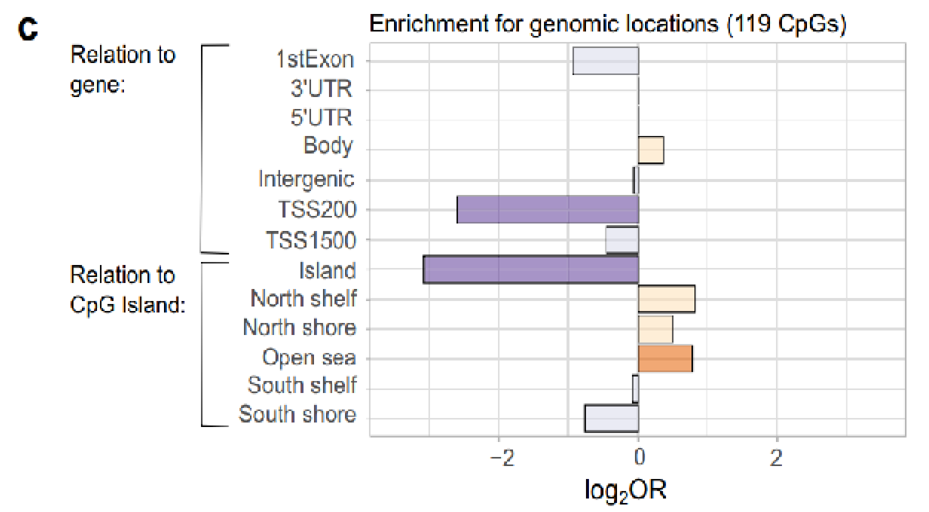


Fig. 2 | The location of the 31 DKD-associated CpGs in the minimal model ($p \leq 9.9 \times 10^{-8}$) in relation to the CpG island (a) or the gene (b) and enriched/depleted genomic locations for CpGs with $p < 1.0 \times 10^{-5}$ in the meta-analysis minimal model. Locations in panel **a** and **b** were retrieved from Illuminas Infinium MethylationEPIC v1.0 B4 Manifest file. The genomic enrichment analyses in panel **c**, including DKD-associated CpGs with a $p < 10^{-5}$, was performed within the web-based EWAS atlas platform. Significant results ($p < 0.05$) are denoted by darker shades. Enriched regions (orange colour) were Open Sea ($p = 5.5 \times 10^{-3}$), and depleted regions (purple colours) were TSS200 ($p = 1.4 \times 10^{-3}$) and Island ($p = 2.8 \times 10^{-7}$). The EWAS toolkit uses the Weighted Fisher's Exact test (two-sided) to compute *p*-values. *DKD* Diabetic kidney disease, *TSS* transcription starting site, *UTR* Untranslated region.

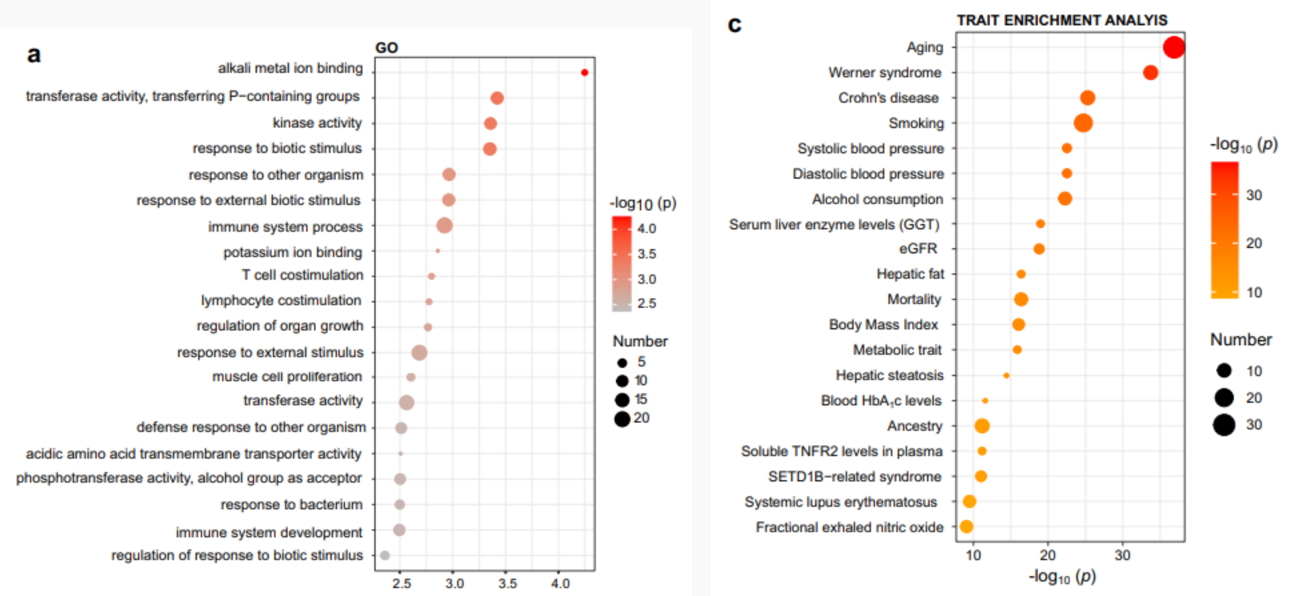


Fig. 4 | Gene ontology (GO) and Kyoto Encyclopaedia of Genes and Genomes (KEGG) enrichment analyses for genes annotated to CpGs with a $p < 1.0 \times 10^{-5}$ and enriched trait associations in the EWAS Atlas for the individual CpGs. The size of the points reflects the number of differentially methylated genes associated with each term (panel **a** and **b**) or the number of CpGs identified for this phenotype in the EWAS Atlas (panel **c**). The colour gradient reflects the association strength.

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