

疾患解析から医療応用を実現する DB開発

東大医、東大病院、東海大学、日立製作所

1-1 2009/01現在での進捗

- ・ 標準SNP DB ー開発完了公開中 逐次データ追加
- ・ SNP品質管理、及び、遺伝統計解析技術の開発 ー開発中
- ・ Case-control DBの構築 ー基本機能実装完了公開中 高機能化実装中
- ・ CNV標準DBの構築 ー基本機能実装完了 高機能化実装中
- ・ ALSリシークエンシングDB ー基本機能実装完了
- ・ パーキンソン病リシークエンシングDB ー開発中
- ・ レポジトリ環境の整備 ー実施中
- ・ 詳細データ公開方針案(倫理検討、アクセス権)の作成

1-2 標準DBの構築

目的: 品質管理、健常対照者データを目的としたDBの構築

内容:

- ・ 健常者500名以上の30-100万SNPの遺伝子型頻度、アレル頻度、ハーディーワインバーク平衡検定値、Call rate等
- ・ 連鎖不平衡値 (D' , r^2)、ハプロタイプ頻度
- ・ SNPのアノテーション (機能、染色体上位置、同義/非同義等)

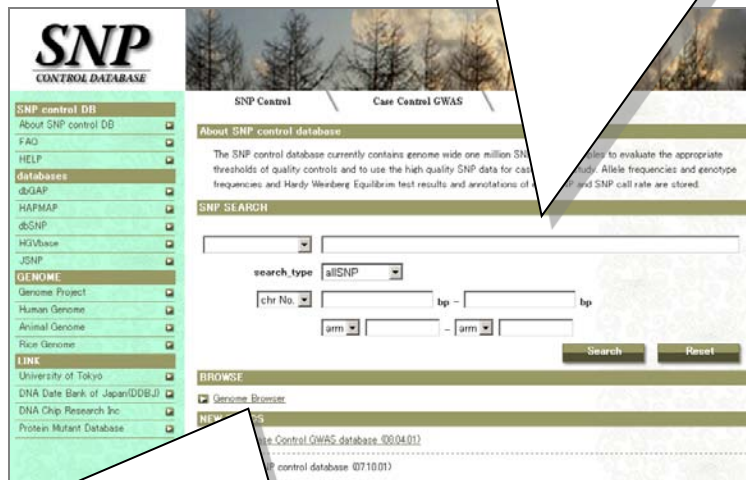
データの品質管理:

- ・ 検体のQC(欠損率、ヘテロ接合度), サンプル構成のQC(サンプル重複、混血検体の同定), SNPのQC(欠損率)

現在:

- ・ DBにはAffy500K 約500検体、Affy6.0 約200検体、Illumina317K 約200検体を蓄積。逐次データを追加。

SNPの検索（アクセッション番号、染色体上の位置、機能、疾患との関連性などで検索可能）



Genome Browserを利用して、他のデータベースコンテンツと同時に表示

SNP search

SNP ID: [NRS6663840](#)

dbSNP ID(rs): [rs6663840](#)

dbSNP ID(ss):

JSNP ID:

HGVbase ID: [chr1:g100000000](#)

Chromosome: 1

Variation Class: SNP

SNP type:

Allele:

Amino acid change:

Affymetrix:

Illumina: A/G

SNPのゲノム上の位置、SNPの種類(同義/非同義など)

Genotype 頻度、アレル頻度、ハプロタイプ頻度、HWE検定値、Call rateなど

Array kind	Ethnic group	Individual Num.	Call Rate	A	G	HWP	Allele	
				0.188	0.51	0.3	A	G
Illumina317K	Japanese	200	1.000	0.188	0.51	0.3	0.450	0.550
Affy500K	Japanese	471	0.965	0.188	0.505	0.305	0.440	0.560
HAPMAP	Japanese	44	1.000	0.272	0.431	0.295	0.490	0.510

Haplotype frequencies

Affy500K NRS12563491- NRS9424283- NRS7543006- NRS2154068- NRS6702916- NRS6702935- NRS6703035- NRS6663840- NRS9424310- NRS17403773- NRS2298225- NRS2298224- NRS17404435- NRS6683156;

AAAAATAGCAAAC	0.403
GGAGGCGAAAAATC	0.393
AGAGATGGAAAGTC	0.104
GGGGCGAAGAATC	0.033
AGGGCGAAGAATC	0.015
AGAGGCGAAAAATC	0.011

Gene Name: KIAA0562

EntrezGene ID : [9731](#)

Gene Symbol : [KIAA0562](#)

Refseq ID (NM-ID) : [NM_014704](#)

Refseq ID (NP-ID) : [NP_055519](#)

Gene ontology (process):

対応する遺伝子のアノテーション情報

目的: 遺伝統計以外の人も解析結果を有効利用できるDBの構築

内容:

- ・ 30-100万SNPの遺伝子型頻度、アレル頻度、ハーディーワインバーグ平衡検定値、Call rate等
- ・ P-value (2DF, 1DF), Additive risk model, recessive model, dominant model のP-value, OR, 95% CI, AICなどの遺伝統計値
- ・ ハプロタイプもしくはSNPの組み合わせに関する疾患関連性の統計値
- ・ SNPのアノテーション (機能、染色体上位置、同義/非同義など)

表示:

- ・ 上記の値を、テーブルとグラフで表示
- ・ プラットフォーム間の比較表示、既知疾患情報との比較表示が可能

現在:

- ・ 12疾患中8疾患を公開中
- ・ 機能追加部分を実装中

1-5

Case-control DBの画面例

GWAS DATABASE

SNP Control Case Control GWAS CNV Database

About This Database

About Case Control GWAS DB

HELP | FAQ

DATABASE

dbGAP

GenMDBJ

HAPMAP

dbSNP

HGVbase

LINK

DBCLS

University of Tokyo

University of Tokai

University of Tokyo Hospital

CRL, Hitachi, LTD.

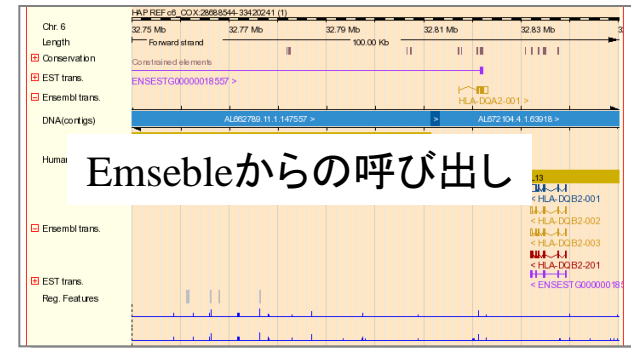
SEARCH

Case Control GWAS search

Disease Name List

Disease Related Gene List

This genome wide association database (GWAS DB) is a repository system and has been constructed to achieve permanent data management and information sharing of genome wide association data. GWAS-DB contains experimental frequency data such as allele frequencies and genotype frequencies, and statistical genetics analysis results such as allelic model, dominant model, recessive model, and additive model and provides graphic viewer to search disease related SNP candidates. Current GWAS DB contains GWAS results of several research laboratories. We greatly appreciate your GWAS data submission. This work has been supported by Ministry of Education, Culture, Sports, Science and Technology.



Disease List

ALL | A | B | C | D | E | F | G | H | I | J | K | L | M | N | O | P | Q | R | S | T | U | V | W | X | Y

- Alzheimer's disease
- Bronchial asthma
- Cerebral aneurysm
- Diabetes mellitus
- Gastric cancer
- Hypertension
- Multiple system atrophy
- Narcolepsy
- Panic disorder

Filtering

Quality: and above

HWE P-value: and above

Call Rate (control): and above

Call Rate (case): and above

SNP Type:

cSNP sSNP rSNP iSNP gSNP

Function:

Choose items

Chromosome Number

Position

SNP Type

Gene Name

AIC

Entropy

P-value

Permutation test

Multiple testing corrections

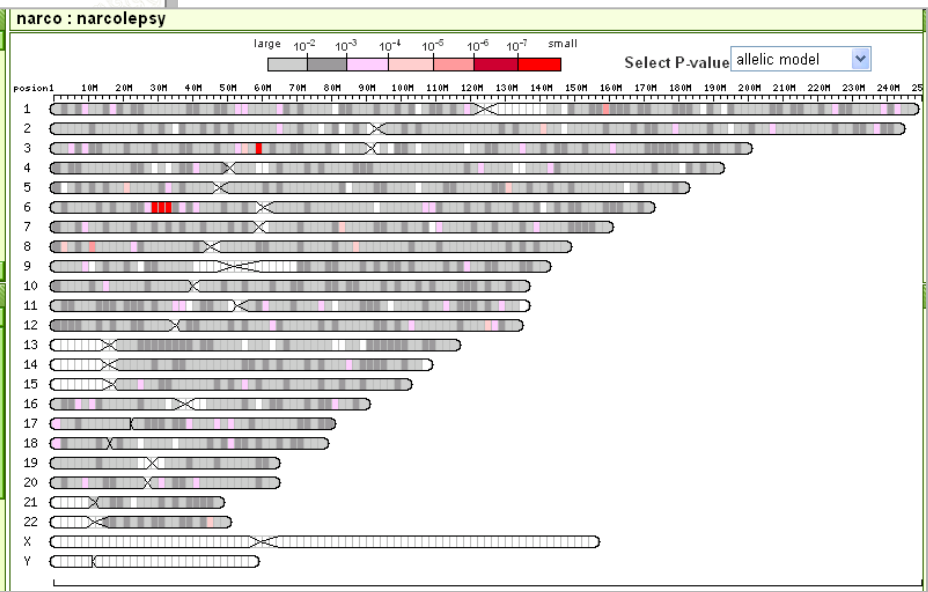
Odds Ratio

95% CI

MAF (case)

MAF (control)

HWE P-value (case)



Study list

Study ID	Disease Name	Study Name	Sample	Case	Co
brain_aneurysm	cerebral aneurysm	brain aneurysm	Aff500K	199	455
cerebral	cerebral aneurysm	cerebral aneurysm	Illumina317K	200	296

1-6

Case-control DBの機能追加例

Filtering

Quality:

MAF (control) 0.05 and above

MAF (case) 0.05 and above

HWE P-value 0.0001 and above

Call Rate (control) 0.95 and above

Call Rate (case) 0.95 and above

SNP Type:

Choose Items

SIDAK_SD

FDR_BH

FDR_BY

Odds Ratio

OR: Allelic model

OR: Genotypic model

OR: Dominant model

OR: Recessive model

OR: Additive model

Choose Graph

-log₁₀(P)

Odds Ratio

exon/intron

narco : narcolepsy

Chromosome 6 Position 30000001 - 32000000 Show 2Mbp

[Study ID]

Studyの比較

SNP ID	Chr	Position	SNP Type	Gene Name	AIC	Allelic P-value	Genotypic P-value	Dom P-value
NRS9261282	6	30143436	ISNP	PPP1R11	3.4280	0.2219	0.4749	0.2656
NRS9261301	6	30149538	ISNP	RNF39	-23.9060	9.495e-08	5.181e-07	8.82e-07
NRS2523990	6	30185208	ISNP	TRIM31	-7.2210	0.0009156	0.002192	0.03042
NRS9261471	6	30213328	ISNP	TRIM40	1.6410	0.1852	0.171	0.4167
NRS2857435	6	30214003	ISNP	TRIM40	2.3400	0.1734	0.2508	0.3165
NRS2857439	6	30214275	ISNP	TRIM40	-1.3270	0.07401	0.0365	0.2354
NRS9261485	6	30216730	ISNP	TRIM40	0.9960	0.1745	0.1255	0.4145
NRS9261488	6	30217391	ISNP	TRIM40	0.8200	0.146	0.1114	0.3656

SNP、遺伝子の検索

Plat form 1 の結果

Plat form 2 の結果

Plat form 1 の結果

Plat form 2 の結果

1-7 CNVのデータベースの表示例

Select Var Type: CNV Select Study: ALL

Position 10M 20M 30M 40M 50M 60M 70M 80M 90M 100M 110M 120M 130M 140M 150M 160M 170M 180M 190M 200M 210M 220M 230M 240M 250M

1
2
3
4
5
6
7
8
9
10
11
12
13
14

Chromosome 16 Region 20500001 - 23000001 Show 2.5Mbp GO

var.1259.1.1
var.1259.1.2
var.1259.1.3

var.1260.1.1
var.1260.1.2
var.1260.1.3

NR237096
NR237097
NR237098
NR237099
NR237100
NR237101
NR237102
NR237103
NR237104
NR237105
NR237106
NR237107
NR237108
NR237109
NR237110
NR237111
NR237112
NR237113
NR237114
NR237115
NR237116
NR237117
NR237118
NR237119
NR237120
NR237121
NR237122
NR237123
NR237124
NR237125
NR237126
NR237127
NR237128
NR237129
NR237130
NR237131
NR237132
NR237133
NR237134
NR237135
NR237136
NR237137
NR237138
NR237139
NR237140
NR237141
NR237142
NR237143
NR237144
NR237145
NR237146
NR237147
NR237148
NR237149
NR237150
NR237151
NR237152
NR237153
NR237154
NR237155
NR237156
NR237157
NR237158
NR237159
NR237160
NR237161
NR237162
NR237163
NR237164
NR237165
NR237166
NR237167
NR237168
NR237169
NR237170
NR237171
NR237172
NR237173
NR237174
NR237175
NR237176
NR237177
NR237178
NR237179
NR237180
NR237181
NR237182
NR237183
NR237184
NR237185
NR237186
NR237187
NR237188
NR237189
NR237190
NR237191
NR237192
NR237193
NR237194
NR237195
NR237196
NR237197
NR237198
NR237199
NR237200
NR237201
NR237202
NR237203
NR237204
NR237205
NR237206
NR237207
NR237208
NR237209
NR237210
NR237211
NR237212
NR237213
NR237214
NR237215
NR237216
NR237217
NR237218
NR237219
NR237220
NR237221
NR237222
NR237223
NR237224
NR237225
NR237226
NR237227
NR237228
NR237229
NR237230
NR237231
NR237232
NR237233
NR237234
NR237235
NR237236
NR237237
NR237238
NR237239
NR237240
NR237241
NR237242
NR237243
NR237244
NR237245
NR237246
NR237247
NR237248
NR237249
NR237250
NR237251
NR237252
NR237253
NR237254
NR237255
NR237256
NR237257
NR237258
NR237259
NR237260
NR237261
NR237262
NR237263
NR237264
NR237265
NR237266
NR237267
NR237268
NR237269
NR237270
NR237271
NR237272
NR237273
NR237274
NR237275
NR237276
NR237277
NR237278
NR237279
NR237280
NR237281
NR237282
NR237283
NR237284
NR237285
NR237286
NR237287
NR237288
NR237289
NR237290
NR237291
NR237292
NR237293
NR237294
NR237295
NR237296
NR237297
NR237298
NR237299
NR237300
NR237301
NR237302
NR237303
NR237304
NR237305
NR237306
NR237307
NR237308
NR237309
NR237310
NR237311
NR237312
NR237313
NR237314
NR237315
NR237316
NR237317
NR237318
NR237319
NR237320
NR237321
NR237322
NR237323
NR237324
NR237325
NR237326
NR237327
NR237328
NR237329
NR237330
NR237331
NR237332
NR237333
NR237334
NR237335
NR237336
NR237337
NR237338
NR237339
NR237340
NR237341
NR237342
NR237343
NR237344
NR237345
NR237346
NR237347
NR237348
NR237349
NR237350
NR237351
NR237352
NR237353
NR237354
NR237355
NR237356
NR237357
NR237358
NR237359
NR237360
NR237361
NR237362
NR237363
NR237364
NR237365
NR237366
NR237367
NR237368
NR237369
NR237370
NR237371
NR237372
NR237373
NR237374
NR237375
NR237376
NR237377
NR237378
NR237379
NR237380
NR237381
NR237382
NR237383
NR237384
NR237385
NR237386
NR237387
NR237388
NR237389
NR237390
NR237391
NR237392
NR237393
NR237394
NR237395
NR237396
NR237397
NR237398
NR237399
NR237400
NR237401
NR237402
NR237403
NR237404
NR237405
NR237406
NR237407
NR237408
NR237409
NR237410
NR237411
NR237412
NR237413
NR237414
NR237415
NR237416
NR237417
NR237418
NR237419
NR237420
NR237421
NR237422
NR237423
NR237424
NR237425
NR237426
NR237427
NR237428
NR237429
NR237430
NR237431
NR237432
NR237433
NR237434
NR237435
NR237436
NR237437
NR237438
NR237439
NR237440
NR237441
NR237442
NR237443
NR237444
NR237445
NR237446
NR237447
NR237448
NR237449
NR237450
NR237451
NR237452
NR237453
NR237454
NR237455
NR237456
NR237457
NR237458
NR237459
NR237460
NR237461
NR237462
NR237463
NR237464
NR237465
NR237466
NR237467
NR237468
NR237469
NR237470
NR237471
NR237472
NR237473
NR237474
NR237475
NR237476
NR237477
NR237478
NR237479
NR237480
NR237481
NR237482
NR237483
NR237484
NR237485
NR237486
NR237487
NR237488
NR237489
NR237490
NR237491
NR237492
NR237493
NR237494
NR237495
NR237496
NR237497
NR237498
NR237499
NR237500
NR237501
NR237502
NR237503
NR237504
NR237505
NR237506
NR237507
NR237508
NR237509
NR237510
NR237511
NR237512
NR237513
NR237514
NR237515
NR237516
NR237517
NR237518
NR237519
NR237520
NR237521
NR237522
NR237523
NR237524
NR237525
NR237526
NR237527
NR237528
NR237529
NR237530
NR237531
NR237532
NR237533
NR237534
NR237535
NR237536
NR237537
NR237538
NR237539
NR237540
NR237541
NR237542
NR237543
NR237544
NR237545
NR237546
NR237547
NR237548
NR237549
NR237550
NR237551
NR237552
NR237553
NR237554
NR237555
NR237556
NR237557
NR237558
NR237559
NR237560
NR237561
NR237562
NR237563
NR237564
NR237565
NR237566
NR237567
NR237568
NR237569
NR237570
NR237571
NR237572
NR237573
NR237574
NR237575
NR237576
NR237577
NR237578
NR237579
NR237580
NR237581
NR237582
NR237583
NR237584
NR237585
NR237586
NR237587
NR237588
NR237589
NR237590
NR237591
NR237592
NR237593
NR237594
NR237595
NR237596
NR237597
NR237598
NR237599
NR237600
NR237601
NR237602
NR237603
NR237604
NR237605
NR237606
NR237607
NR237608
NR237609
NR237610
NR237611
NR237612
NR237613
NR237614
NR237615
NR237616
NR237617
NR237618
NR237619
NR237620
NR237621
NR237622
NR237623
NR237624
NR237625
NR237626
NR237627
NR237628
NR237629
NR237630
NR237631
NR237632
NR237633
NR237634
NR237635
NR237636
NR237637
NR237638
NR237639
NR237640
NR237641
NR237642
NR237643
NR237644
NR237645
NR237646
NR237647
NR237648
NR237649
NR237650
NR237651
NR237652
NR237653
NR237654
NR237655
NR237656
NR237657
NR237658
NR237659
NR237660
NR237661
NR237662
NR237663
NR237664
NR237665
NR237666
NR237667
NR237668
NR237669
NR237670
NR237671
NR237672
NR237673
NR237674
NR237675
NR237676
NR237677
NR237678
NR237679
NR237680
NR237681
NR237682
NR237683
NR237684
NR237685
NR237686
NR237687
NR237688
NR237689
NR237690
NR237691
NR237692
NR237693
NR237694
NR237695
NR237696
NR237697
NR237698
NR237699
NR237700
NR237701
NR237702
NR237703
NR237704
NR237705
NR237706
NR237707
NR237708
NR237709
NR237710
NR237711
NR237712
NR237713
NR237714
NR237715
NR237716
NR237717
NR237718
NR237719
NR237720
NR237721
NR237722
NR237723
NR237724
NR237725
NR237726
NR237727
NR237728
NR237729
NR237730
NR237731
NR237732
NR237733
NR237734
NR237735
NR237736
NR237737
NR237738
NR237739
NR237740
NR237741
NR237742
NR237743
NR237744
NR237745
NR237746
NR237747
NR237748
NR237749
NR237750
NR237751
NR237752
NR237753
NR237754
NR237755
NR237756
NR237757
NR237758
NR237759
NR237760
NR237761
NR237762
NR237763
NR237764
NR237765
NR237766
NR237767
NR237768
NR237769
NR237770
NR237771
NR237772
NR237773
NR237774
NR237775
NR237776
NR237777
NR237778
NR237779
NR237780
NR237781
NR237782
NR237783
NR237784
NR237785
NR237786
NR237787
NR237788
NR237789
NR237790
NR237791
NR237792
NR237793
NR237794
NR237795
NR237796
NR237797
NR237798
NR237799
NR237800
NR237801
NR237802
NR237803
NR237804
NR237805
NR237806
NR237807
NR237808
NR237809
NR237810
NR237811
NR237812
NR237813
NR237814
NR237815
NR237816
NR237817
NR237818
NR237819
NR237820
NR237821
NR237822
NR237823
NR237824
NR237825
NR237826
NR237827
NR237828
NR237829
NR237830
NR237831
NR237832
NR237833
NR237834
NR237835
NR237836
NR237837
NR237838
NR237839
NR237840
NR237841
NR237842
NR237843
NR237844
NR237845
NR237846
NR237847
NR237848
NR237849
NR237850
NR237851
NR237852
NR237853
NR237854
NR237855
NR237856
NR237857
NR237858
NR237859
NR237860
NR237861
NR237862
NR237863
NR237864
NR237865
NR237866
NR237867
NR237868
NR237869
NR237870
NR237871
NR237872
NR237873
NR237874
NR237875
NR237876
NR237877
NR237878
NR237879
NR237880
NR237881
NR237882
NR237883
NR237884
NR237885
NR237886
NR237887
NR237888
NR237889
NR237890
NR237891
NR237892
NR237893
NR237894
NR237895
NR237896
NR237897
NR237898
NR237899
NR237900
NR237901
NR237902
NR237903
NR237904
NR237905
NR237906
NR237907
NR237908
NR237909
NR237910
NR237911
NR237912
NR237913
NR237914
NR237915
NR237916
NR237917
NR237918
NR237919
NR237920
NR237921
NR237922
NR237923
NR237924
NR237925
NR237926
NR237927
NR237928
NR237929
NR237930
NR237931
NR237932
NR237933
NR237934
NR237935
NR237936
NR237937
NR237938
NR237939
NR237940
NR237941
NR237942
NR237943
NR237944
NR237945
NR237946
NR237947
NR237948
NR237949
NR237950
NR237951
NR237952
NR237953
NR237954
NR237955
NR237956
NR237957
NR237958
NR237959
NR237960
NR237961
NR237962
NR237963
NR237964
NR237965
NR237966
NR237967
NR237968
NR237969
NR237970
NR237971
NR237972
NR237973
NR237974
NR237975
NR237976
NR237977
NR237978
NR237979
NR237980
NR237981
NR237982
NR237983
NR237984
NR237985
NR237986
NR237987
NR237988
NR237989
NR237990
NR237991
NR237992
NR237993
NR237994
NR237995
NR237996
NR237997
NR237998
NR237999
NR238000

複数の計算データを一度に閲覧

クリックすると領域の図

高さが頻度
色の濃さがCNV数

各パターンのgenotype

DGVのデータ

Var_Num	Sub number	Region	Variation Type	Frequency	Genotype pattern	GDV-ID:region	Gene Name
var.1259	1	Chr16 21441805-21493293		14/198			
var.1259	1.1	Chr16 21441805-21493293	CNV	2/198	NR8150335:4- NR8183170:4- NR8636204:4- NR8150336:4- NR8574661:4		
var.1259	1.1.1	Chr16 21441805-21493293	CNV	1/198	NR8150335:0A4G- NR8183170:4C0T- NR8636204:3G1T- NR8150336:0A4G- NR8574661:0A4G		
var.1259	1.1.2	Chr16 21441805-21493293	CNV	1/198	NR8150335:0A4G- NR8183170:2C2T- NR8636204:2G2T- NR8150336:2A2G- NR8574661:1A3G		
var.1259	1.2	Chr16 21441805-21493293	CNV	7/198	NR8150335:3- NR8183170:3- NR8636204:3- NR8150336:3- NR8574661:3		
var.1259	1.2.1	Chr16 21441805-21493293	CNV	1/198	NR8150335:0A3G- NR8183170:1C2T- NR8636204:1G2T- NR8150336:0A3G- NR8574661:3A0G		
var.1259	1.2.2	Chr16 21441805-21493293	CNV	1/198	NR8150335:0A3G- NR8183170:2C1T- NR8636204:2G1T-		

- 頻度情報、統計解析結果については一般公開
- 統合データベースプロジェクト ヒトゲノム多型データ共有方針を、「特定ゲノム」の方針を参考にして、中核機関と共同で作成済み
- データ受け入れ時のデータ利用の申請書とデータの利用に関する同意書とセキュリティ手順書を作成済み
- 検討委員会を招集し、年度末までに決定予定


2-1 今年度のGWAS-DB予定

- ・ SNP間相互作用の蓄積、グラフ表示機能。
- ・ SNPごとのシグナル強度のグラフ表示などのGWAS-DBのインターフェースの拡張。
- ・ 子宮内膜症(厚生科研)についてのケースコントロールデータをDBに搭載予定。
- ・ 外部の機関が産出したデータの受け入れのためのレポジトリシステムの構築。

3-1 リシークエンスDB進捗と今年度の予定

- ・ パーキンソン病のリシークエンスDBの構築とALSリシークエンスDBのインターフェースの改良
- ・ 目的:
 - 網羅的なmutationの情報とそれに付随する臨床情報を搭載し、臨床現場にも役立つDBの構築を目的
- ・ 内容:
 - 疾患関連遺伝子に関する患者の独自のリシークエンスデータと付随する臨床情報
 - 論文から収集したmutation情報と臨床情報(症状の種類、発症年齢など)
 - その他
 - Mutationの遺伝子機能情報、立体構造情報など
- ・ 現在: ALSは構築完了、パーキンソン病は構築中
- ・ 今年度の予定: パーキンソン病の実験データの追加

3-2 リシークエンスDBの画面例



About PD

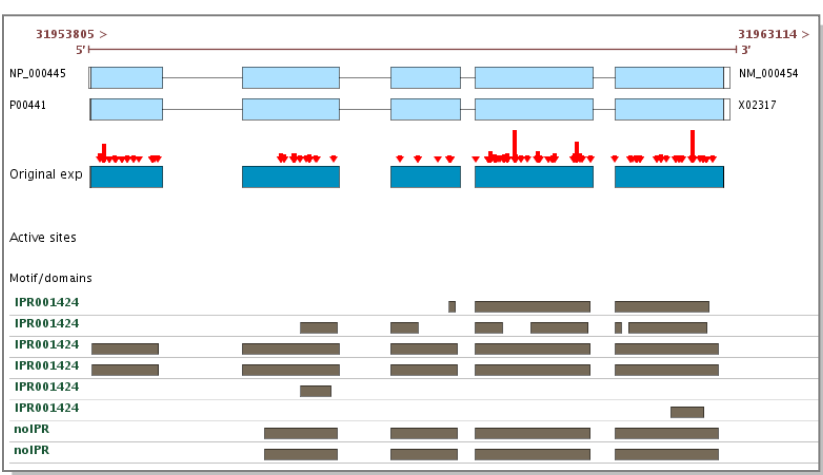
Parkinson's disease (PD) is a progressive degenerative disorder of the central nervous system. The mechanisms to cause PD are not fully clarified. The aim of this database is to collect mutations related to PD and their clinical information exhaustively in order to deepen our understanding of PD. Both our original data and extracted data from published papers are accumulated in this database.

Causative genes

- [PARK7](#)
- [LRRK2](#)
- [GBA](#)
- [HTRA2](#)
- [NF4A2](#)
- [PARK2](#)
- [PINK1](#)
- [SNCA](#)
- [SNCG](#)
- [UCHL1](#)
- [SNCAIP](#)

Related genes

- [GPR37](#)
- [MTX1](#)
- [SNCB](#)



31953805 > 31963114 >

5'- 3'

NP_000445 NM_000454

P00441 X02317

Original exp

Active sites

Motif/domains

IPR001424

IPR001424

IPR001424

IPR001424

IPR001424

noIPR

noIPR


aa.No.	1	11
NM_000454	G C T A G C G A G T T A T G G C G A C G A A G G C C G T G T G C G T G C T G A A G G G C G A C G G C C C A G T G C A G	G ## ## G# ## # #
NP_000445	M A T K A V C V L K G D G P V Q	

Exon No.	1	start
#1253	GCCNN	GCGNNGNCGACGAAAGGCN
#2464	GCTAG	CGSAGTTATGCGCAGAAAGCCGTG
#3008	GCCTA	GCGAGTTATGCGCAGAAAGCCGTG
#3271	GCCTA	GCGAGTTATGCGCAGAAAGCCGTG
#3280	GCCTA	GCGAGTTATGCGCAGAAAGCCGTG
#3356	GCCTA	GCGAGTTATGCGCAGAAAGCCGTG
#3388	GCCTA	GCGAGTTATGCGCAGAAAGCCGTG
#3631	GCCTA	GCGAGTTATGCGCAGAAAGCCGTG
#3641	GCCTA	GCGAGTTATGCGCAGAAAGCCGTG
#3655	GCCTA	GCGAGTTATGCGCAGAAAGCCGTG
#3680	GCCTA	GCGAGTTATGCGCAGAAAGCCGTG
#3711	GCCTA	GCGAGTTATGCGCAGAAAGCCGTG
#3721	GCCTA	GCGAGTTATGCGCAGAAAGCCGTG
#3757	GCCTA	GCGAGTTATGCGCAGAAAGCCGTG
#3760	GCCTA	GCGAGTTATGCGCAGAAAGCCGTG
#3787	GCCTA	GCGAGTTATGCGCAGAAAGCCGTG
#3806	GCCTA	GCGAGTTATGCGCAGAAAGCCGTG
#3812	GCCTA	GCGAGTTATGCGCAGAAAGCCGTG
#3813	GCCTA	GCGAGTTATGCGCAGAAAGCCGTG

PARK7 Parkinson disease (autosomal recessive, early onset) 7

Switch: [Simple mode](#) / [Detail mode](#) / [Experimental Data only](#)

DNA change	mRNA Accession No.	Genomic position	rs ID	Amino Acid change	Structure	Protein Accession No.	homolo
AT0aATA	NM_007262	chr1 07945555A		MSI		NP_009193	His
GAGaGAC	NM_007262	chr1 07946072C		E64D		NP_009193	His
263_323aa	NM_007262	chr1 795351...7953810aa		V86Ixx1D		NP_009193	His
CGaNCAD	NM_007262	chr1 07955581A		R89G		NP_009193	His
CGaNCAD	NM_007262	chr1 07955581A		R89G		NP_009193	His
CGaNCAD	NM_007262	chr1 07955581A		R89G		NP_009193	His Eggsfin



4

来年度の計画

- ・ CNVの検出手法の改良とCNVのcase-control DBの構築
- ・ より多くのGWASデータの受け入れ、登録と公開
- ・ 一次データと二次データの関係性の表示、発現データなどの他実験データなどとの重ね合わせ表示など、GWAS DBの拡張
- ・ SNP相互作用部分の計算手法の開発
- ・ 新たに定める倫理規定、及び手順に従ったデータの預け入れと再配布サービスの開始
- ・ 海外他機関との連携の検討
- ・ パーキンソン病リシーケンスDBの仕上げ
- ・ 副腎白質ジストロフィーリシーケンスDB、痙性対麻痺リシーケンスDBの構築