

1 Case-control DBの機能追加

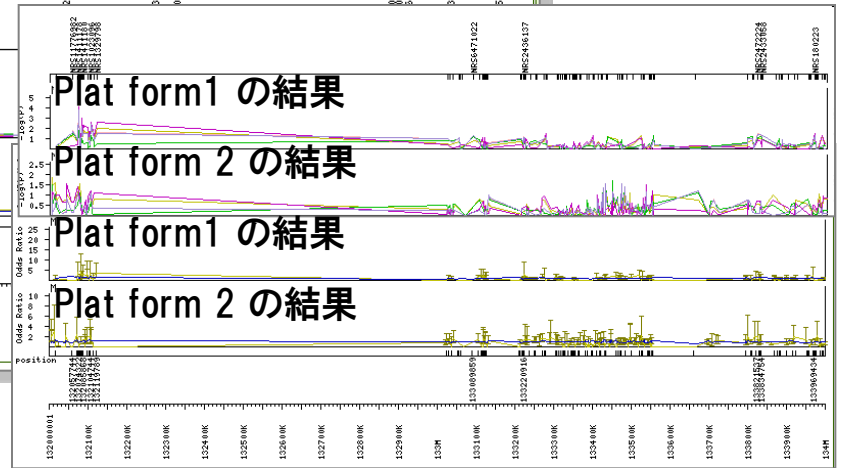
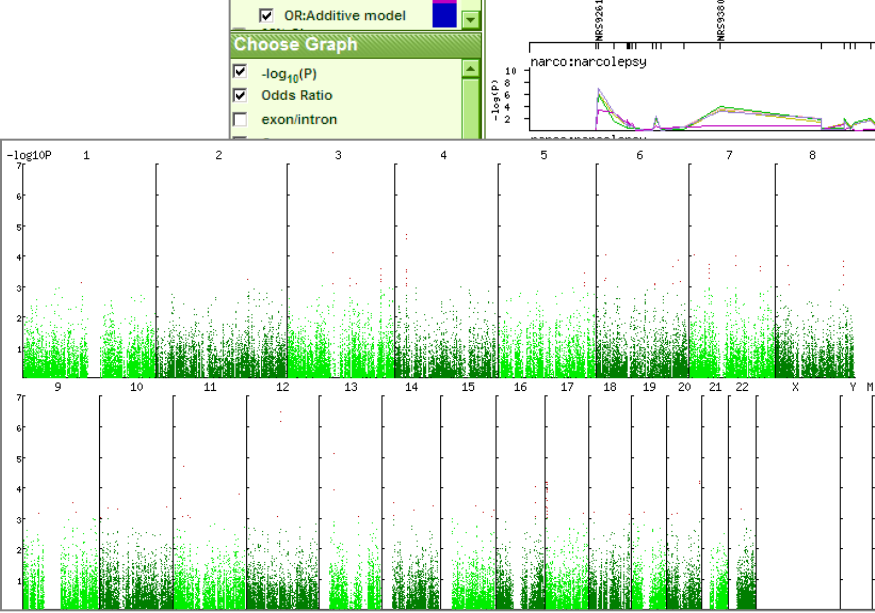
narco : narcolepsy

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



2

CNV databaseの画面例

Select Var Type: CNV Select Study: affy6.0_normal

Chromosome 1 Region 3400001 4300000 Show 900Kbp go

BOXの高さが個体数
BOXの色がCNV数

Var-Num	Sub number	Region	Variation Type	Frequency	Genotype pattern
var.21	3	Chr1 3493933-3529341		5/198	
var.21	3.1	Chr1 3493933-3529341	CNV	5/198	NRS12139206.4 - NRS2794327.4 - NRS2821025.4 - NRS4364818.4
var.21	3.1.1	Chr1 3493933-3529341	CNV	1/198	NRS12139206.4C0T - NRS2794327.1C3T - NRS2821025.0A4G - NRS4364818.4
var.21	3.1.2	Chr1 3493933-3529341	CNV	1/198	NRS12139206.2C2T - NRS2794327.2C2T - NRS2821025.0A4G - NRS4364818.4
var.21	3.1.3	Chr1 3493933-3529341	CNV	1/198	NRS12139206.4C0T - NRS2794327.3C1T - NRS2821025.0A4G - NRS4364818.4
var.21	3.1.4	Chr1 3493933-3529341	CNV	1/198	NRS12139206.0C4T - NRS2794327.1C3T - NRS2821025.0A4G - NRS4364818.4
var.21	3.1.5	Chr1 3493933-3529341	CNV	1/198	NRS12139206.4C0T - NRS2794327.2C2T - NRS2821025.0A4G - NRS4364818.4
var.21	4	Chr1 3493933-3529341		3/198	
var.21	4.1	Chr1 3493933-3529341	CNV	1/198	NRS12139206.4 - NRS2794327.4 - NRS2821025.4 - NRS4364818.4
var.21	4.1.1	Chr1 3493933-3529341	CNV	1/198	NRS12139206.2C2T - NRS2794327.2C2T - NRS2821025.0A4G - NRS4364818.4
var.21	4.2	Chr1 3493933-3529341	CNV	2/198	NRS12139206.3 - NRS2794327.3 - NRS2821025.3 - NRS4364818.4
var.21	4.2.1	Chr1 3493933-3529341	CNV	1/198	NRS12139206.3C0T - NRS2794327.3C0T - NRS2821025.0A3G - NRS4364818.4
var.21	4.2.2	Chr1 3493933-3529341	CNV	1/198	NRS12139206.3C0T - NRS2794327.3C0T - NRS2821025.0A3G - NRS4364818.4
var.22	2	Chr1 3576310-3618537		3/198	
var.22	2.1	Chr1 3576310-3618537	CNV	1/198	NRS12027041.4 - NRS3765730.4 - NRS751035.4 - NRS17379833.4
var.22	2.1.1	Chr1 3576310-3618537	CNV	1/198	NRS12027041.2C2G - NRS3765730.2A2G - NRS751035.1A3G - NRS17379833.4
var.22	2.2	Chr1 3576310-3618537	CNV	2/198	NRS12027041.3 - NRS3765730.3 - NRS751035.3 - NRS17379833.4
var.22	2.2.1	Chr1 3576310-3618537	CNV	1/198	NRS12027041.2C1G - NRS3765730.0A3G - NRS751035.0A3G - NRS17379833.4
var.22	2.2.2	Chr1 3576310-3618537	CNV	1/198	NRS12027041.1C2G - NRS3765730.0A3G - NRS751035.0A3G - NRS17379833.0A3T - NRS17379833.4
var.22	3	Chr1 3576310-3618537		8/198	
var.22	3.1	Chr1 3576310-3618537	CNV	5/198	NRS4276857.4 - NRS732903.4 - NRS3765696.4 - NRS12027041.4 - NRS3765730.4 - NRS17379833.4
var.22	3.1.1	Chr1 3576310-3618537	CNV	2/198	NRS4276857.2C2T - NRS732903.0A4G - NRS3765696.0C4T - NRS12027041.0C4G - NRS17379833.4

Chr1 3400001 4300000

Gene/contig

CNV

既知のCNV

PD mutation

Top Mutation Search Help

Causative genes

- [PARK7](#)
- [LRRK2](#)
- [GBA](#)
- [HTRA2](#)
- [NR4A2](#)
- [PARK2](#)
- [PINK1](#)
- [SNCA](#)
- [SNCG](#)
- [UCHL1](#)
- [SNCAIP](#)

Related genes

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

Link

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 database is to collect mutations related to PD and their clinical information exhaustively in order to deepen our understanding of PD. Both our original extracted data from published papers are accumulated in this database.

Causative genes

- [PARK7](#) Parkinson disease (autosomal recessive, early onset) 7
- [LRRK2](#) leucine-rich repeat kinase 2
- [GBA](#) glucosidase, beta; acid (includes glucosylceramidase)
- [HTRA2](#) HtrA serine peptidase 2
- [NR4A2](#) nuclear receptor subfamily 4, class A, member 2
- [PARK2](#) Parkinson disease (autosomal recessive, late onset) 2
- [PINK1](#) PTEN induced putative kinase 1
- [SNCA](#) synuclein, alpha (non A4) variant

7944379 > 7967925 >

5' 3'

NP_009193 NM_007262

Q99497 D61380

Original exp

Active sites

Motif/domains

- IPR002818
- IPR006287
- noIPR
- noIPR
- noIPR
- noIPR

a.a.: NP_009193

PARK7 Parkinson disease (autosomal recessive, early onset) 7

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

DNA change	mRNA Accession No.	Genomic position	r.s ID	Amino Acid change	Structure	Protein Accession No.	homo/hetero	Population	Ref
ATGtoATA	NM_007262	chr1 07945505A		M2E		NP_009193	Homo	Ashkenazi Jewish	
GAGtoGAC	NM_007262	chr1 07946072C		E64D		NP_009193	Homo	Australian	
253_322del	NM_007262	chr1 7963541_7963610del		V95fsX10		NP_009193	Hetero	Ashkenazi Jewish	
CGGtoCAG	NM_007262	chr1 07953581A		R98Q		NP_009193	Hetero		
CGGtoCAG	NM_007262	chr1 07953581A		R98Q		NP_009193	Hetero	Egyptian	
CGGtoCAG	NM_007262	chr1 07953581A		R98Q		NP_009193	Hetero	Egyptian	