
GWAS Source
CURATION
Phenotype details
Top-level summary
Marker details
Phenotype summary
Phenotype details
Panel information
Experiment details
Association results
Marker details
Phenotype summary
GWAS Central identifier
HGVPM126
Phenotype
Parkinson's disease
Used in Study
GWAS of Parkinson's disease (HGVST83)
Description
Sibling pairs that are discordant for Parkinson's Disease.
Variable type
Not supplied
Variable unit
Not supplied
Related links
Not supplied
Phenotype annotation
MeSH: Parkinson Disease (D010300)
Phenotypes inferred from OMIM
PARKINSON DISEASE (168600)
HPO: Mask-like facies (HP:0000298)
HPO: Depression (HP:0000716)
HPO: Dementia (HP:0000726)
HPO: Personality changes (HP:0000751)
HPO: Dysarthria (HP:0001260)
HPO: Parkinsonism (HP:0001300)
HPO: Tremor (HP:0001337)
HPO: Soft voice (HP:0001621)
HPO: Rigidity (HP:0002063)
HPO: Bradykinesia (HP:0002067)
HPO: Hyposmia (HP:0004409)
HPO: Short stepped shuffling gait (HP:0007311)
Summary
View
Panels
Phenotypes
Analysis experiments
Association results
The following Phenotypes are defined in this Study. What is a Phenotype
Phenotype
Description
Parkinson's disease
Sibling pairs that are discordant for Parkinson's Disease.
Parkinson's disease
Not provided
The following Sample Panels are defined in this Study. What is a Sample Panel
Sample Panel name
No. individuals
Panel composition
Geographic region
Tier 1 cohort (Discordant sibling-pairs)
886
Not provided
All resided within Minnesota or one of the surrounding four states (Wisconsin, Iowa, South Dakota, or North Dakota)
Tier 2 cohort
664
Not provided
All resided within Minnesota or one of the surrounding four states (Wisconsin, Iowa, South Dakota, or North Dakota)
The following Assayed Panels are defined in this Study. What is an Assayed Panel
Assayed Panel name
No. individuals
Selected from Sample Panel(s)
Phenotype(s) tested
Cases (Tier 1: PD)
344
Tier 1 cohort (Discordant sibling-pairs)
Parkinson's disease
Controls (Tier 1: Sibling pairs discordant for Parkinson's disease)
443
Tier 1 cohort (Discordant sibling-pairs)
Parkinson's disease
Cases (Tier 2: PD)
332
Tier 2 cohort
Parkinson's disease
Controls (unrelated unaffected)
332
Tier 2 cohort
Parkinson's disease
GWAS Central identifier
HGVST83
Study name
GWAS of Parkinson's disease
Total markers imported
195,214
Phenotype(s) tested
Parkinson's disease
Study design
Case and control
Genotype platforms
Not supplied
Abstract
Submission information
Contributor
Date Submitted
Author?
Submitter?
Source?
GWAS Central
2002-12-09
Maraganore DM et al.
2002-12-09
Author communication
Corresponding author
Date of contact
Response received
Date of response
Type of response
Dennis G. Ballinger
2011-03-17
2011-03-17
E-mail bounce
Links
Supplementary data from PMID16252231
The Study contains 2 Analysis Experiments. What is an Analysis Experiment
Expand All
Collapse All
Experiment HGVE138
Association analysis experiment (Tier 1) for Parkinson's disease
Phenotype
Parkinson's disease
Total no. markers imported
192,227
Genotype platform(s)
Analysis summary
Analysis Method
Result Set
log-additive method
Trend analysis (HGVRS137)
Assayed panels used in Experiment
Assayed panel name
No. individuals
Selected from Sample panel(s)
Cases (Tier 1: PD)
344
Tier 1 cohort (Discordant sibling-pairs)
Controls (Tier 1: Sibling pairs discordant for Parkinson's disease)
443
Tier 1 cohort (Discordant sibling-pairs)
Related links
Supplementary file 1
Individual-level data statement
Access to individual-level data must be made to the study authors
Comments
None
GWAS Central marker ID
HGVM2262419
Marker accession ID
rs3746736
Variation type
SNP
Observed alleles
5' upstream 30bp
(allele seq)
3' downstream 30bp
Genomic location
chr20:23,424,613..23,424,613 (view in genome browser: Ensembl | UCSC)
Mapped gene
CSTL1[exonic]
Marker status and Revision History
This marker is currently ACTIVE (no revision history)
Cross-references
None available
Links
OMIM
SNPedia
dbSNP
Top 50 associations for Parkinson's disease (HGVPM126) from data set HGVRS137(192,227 in database)
Experiment: Association analysis experiment (Tier 1) for Parkinson's disease (HGVE138)
Data set: Trend analysis (HGVRS137)
Rank
Identifier
Accession
Region
p-value
-log p-value
Effect size
Risk allele
Related data
Links