

# main\_genes

September 2, 2021

## 1 Enrichment and Overlap of PGC2+CLOZUK

```
[1]: import re
import os, errno
import functools
import numpy as np
import pandas as pd
from plotnine import *
from pandas_plink import read_plink
from warnings import filterwarnings
from matplotlib.cbook import mplDeprecation
from scipy.stats import fisher_exact, binom_test

filterwarnings("ignore",category=mplDeprecation)
filterwarnings('ignore', category=UserWarning, module='plotnine.*')
filterwarnings('ignore', category=DeprecationWarning, module='plotnine.*')
```

### 1.1 Config and Functions

```
[2]: config = {
    'biomart_file': '../_h/biomart.csv',
    'phenotype_file': '/ceph/projects/v4_phase3_paper/inputs/phenotypes/_m/
    ↪merged_phenotypes.csv',
    'plink_file_prefix': '/ceph/projects/v4_phase3_paper/inputs/genotypes/_m/
    ↪LIBD_Brain_TopMed',
    'gwas_snp_file': '/ceph/projects/v4_phase3_paper/inputs/sz_gwas/pgc2_clozuk/
    ↪map_phase3/_m/libd_hg38_pgc2sz_snps.tsv'
}

config_feature = {
    'de_file': '.././differential_expression/_m/genes/diffExpr_szVctl_full.
    ↪txt',
    'residual_expression_file': '.././differential_expression/_m/genes/
    ↪residualized_expression.tsv',
    'fastqtl_output_file': '.././eqtl/caudate/summary_table/_m/
    ↪Brainseq_LIBD_caudate_4features.signifpairs.txt.gz',
}
```

```
feature = "genes"
```

```
[3]: @functools.lru_cache()
def feature_map(feature):
    return {"genes": "Gene", "transcripts": "Transcript",
            "exons": "Exon", "junctions": "Junction"}[feature]

@functools.lru_cache()
def get_de_df():
    """
    Load DE analysis
    """
    return pd.read_csv(config_feature['de_file'], sep='\t', index_col=0)

@functools.lru_cache()
def get_eqtl_df():
    eqtl_df = pd.read_csv(config_feature['fastqtl_output_file'], sep='\t')
    return eqtl_df[(eqtl_df["Type"] == feature_map(feature))]

@functools.lru_cache()
def get_gwas_snps():
    return pd.read_csv(config['gwas_snp_file'], sep='\t', index_col=0,
    ↪low_memory=False)

@functools.lru_cache()
def get_integration_df():
    return get_gwas_snps().merge(get_eqtl_df(), left_on='our_snp_id',
    ↪right_on='variant_id',
                                suffixes=['_PGC2', '_eQTL'])\
    .merge(get_de_df(), left_on='gene_id',
    ↪right_index=True)

@functools.lru_cache()
def get_residual_expression_df():
    return pd.read_csv(config_feature['residual_expression_file'],
                        sep='\t', index_col=0).transpose()

@functools.lru_cache()
def get_pheno_df():
    return pd.read_csv(config['phenotype_file'], index_col=0)
```

```
[4]: def agree_direction(row):
    return [-1, 1][row['pgc2_a1_same_as_our_counted']] * np.sign(row['OR'] - 1)
    ↪ * np.sign(row['slope']) * np.sign(row['t'])

def letter_snp(number, a0, a1):
    '''
    Example:
    letter_snp(0, 'A', 'G') is 'AA'
    letter_snp(1, 'A', 'G') is 'AG'
    letter_snp(2, 'A', 'G') is 'GG'

    '''
    if np.isnan(number):
        return np.nan
    if len(a0)==1 and len(a1)==1:
        sep = ''
    else:
        sep = ' '
    return sep.join(sorted([a0]*int(number) + [a1]*(2-int(number))))

def get_gwas_snp(snp_id):
    gwas = get_gwas_snps()
    r = gwas[gwas['our_snp_id']==snp_id]
    assert len(r) == 1
    return r
```

```
[5]: @functools.lru_cache()
def get_expression_and_pheno_df():
    return pd.merge(get_pheno_df(), get_residual_expression_df(),
    ↪ left_index=True, right_index=True)

@functools.lru_cache()
def get_plink_tuple():
    '''
    Usage: (bim, fam, bed) = get_plink_tuple()
    '''
    return read_plink(config['plink_file_prefix'])

@functools.lru_cache()
def subset_bed():
    """
    This subsets the bed and bim file and returns the new subsetted
    data with shared brain_ids.
```

```

This is to speed up accessing the bed file.
"""
(bim, fam, bed) = get_plink_tuple()
brain_ids = list(set(get_expression_and_pheno_df()['BrNum'])).
↪intersection(set(fam['fid'])))
    fam_pos = list(fam[(fam["fid"].isin(brain_ids))]).
↪drop_duplicates(subset="fid").loc[:, 'i'])
    unique_snps = get_eqtl_df().variant_id.unique()
    snp_info = bim[(bim["snp"].isin(unique_snps))].copy()
    snp_pos = list(snp_info.loc[:, "i"])
    new_bed = bed[snp_pos].compute()[:, fam_pos]
    new_bim = bim[(bim["i"].isin(snp_pos))].reset_index(drop=True)
    new_bim['ii'] = new_bim.index
    return new_bed, new_bim, brain_ids

@functools.lru_cache()
def get_snp_df(snp_id):
    """
    Returns a dataframe containing the genotype on snp snp_id.
    The allele count is the same as in the plink files.

    Example:
    get_snp_df('rs653953').head(5)

        rs653953_num rs653953_letter rs653953
    Br5168          0             GG      0\nGG
    Br2582          1             AG      1\nAG
    Br2378          1             AG      1\nAG
    Br5155          2             AA      2\nAA
    Br5182          2             AA      2\nAA
    """
    bed, bim, brain_ids = subset_bed()
    snp_info = bim[bim['snp']==snp_id]
    snp_pos = snp_info.iloc[0]['ii']
    dfsnp = pd.DataFrame(bed[[snp_pos]], columns=brain_ids, index=[snp_id + '_num'])
    ↪.transpose().dropna()
    my_letter_snp = functools.partial(letter_snp, a0=snp_info.iloc[0]['a0'],
    ↪a1=snp_info.iloc[0]['a1'])
    # the 2 - in next line is to workaround a possible bug in pandas_plink? a1
    ↪and a0 inverted
    dfsnp[[snp_id + '_num']] = 2 - dfsnp[[snp_id + '_num']].astype('int')
    dfsnp[snp_id + '_letter'] = dfsnp[snp_id + '_num'].apply(my_letter_snp)
    dfsnp[snp_id] = (dfsnp[snp_id + '_num'].astype('str') + '\n' +
                     dfsnp[snp_id + '_letter'].astype('str')).astype('category')
    return dfsnp

```

```

@functools.lru_cache()
def get_gwas_ordered_snp_df(snp_id):
    """
    Returns a dataframe containing the genotype on snp snp_id.
    The allele count is the number of risk alleles according to GWAS.

    Example:
    get_gwas_ordered_snp_df('rs653953').head(5)

           rs653953_num rs653953_letter rs653953
    Br5168             2             GG      2\nGG
    Br2582             1             AG      1\nAG
    Br2378             1             AG      1\nAG
    Br5155             0             AA      0\nAA
    Br5182             0             AA      0\nAA
    """
    pgc = get_gwas_snps()
    dfsnp = get_snp_df(snp_id).copy()
    gwas_snp = get_gwas_snp(snp_id)
    if gwas_snp['pgc2_a1_same_as_our_counted'].iloc[0]:
        if gwas_snp['OR'].iloc[0] > 1:
            pass
        else:
            dfsnp[[snp_id + '_num']] = 2 - dfsnp[[snp_id + '_num']]
    else:
        if gwas_snp['OR'].iloc[0] > 1:
            dfsnp[[snp_id + '_num']] = 2 - dfsnp[[snp_id + '_num']]
        else:
            pass
    dfsnp[snp_id] = (dfsnp[snp_id + '_num'].astype('str') + '\n' +
                    dfsnp[snp_id + '_letter'].astype('str')).astype('category')
    return dfsnp

@functools.lru_cache()
def get_biomart_df():
    biomart = pd.read_csv(config['biomart_file'])
    biomart['description'] = biomart['description'].str.replace('\[Source.
↪*$',' ', regex=True)
    return biomart

@functools.lru_cache()
def get_risk_allele(snp_id):
    gwas_snp = get_gwas_snp(snp_id)

```

```

if gwas_snp['OR'].iloc[0] > 1:
    ra = gwas_snp['A1'].iloc[0]
else:
    ra = gwas_snp['A2'].iloc[0]

return ra

```

```

[6]: def get_gene_symbol(gene_id, biomart=get_biomart_df()):
    ensge = re.sub('\.+$', '', gene_id)
    ggg = biomart[biomart['ensembl_gene_id']==ensge]
    if ggg.shape[0]==0:
        return '', ''
    gs = ggg['external_gene_name'].values[0]
    de = ggg['description'].values[0]
    if type(de)!=str:
        de = ''
    de = re.sub('\[Source:.*$', '', de)
    return gs, de

def save_plot(p, fn):
    for ext in ['png', 'pdf', 'svg']:
        p.save(fn + '.' + ext)

def get_snp_gene_pheno_df(snp_id, gene_id, snp_df_func):
    pheno_columns = list(get_pheno_df().columns)
    expr_df = get_expression_and_pheno_df()[pheno_columns + [gene_id]]
    snp_df = snp_df_func(snp_id)
    return expr_df.merge(snp_df, left_on='BrNum', right_index=True)

def simple_snp_expression_pheno_plot_impl(snp_id, gene_id, snp_df_func,
    pheno_var):
    df = get_snp_gene_pheno_df(snp_id, gene_id, snp_df_func)
    df['Dx'] = df.Dx.astype('category').cat.rename_categories({'Control':
    'CTL', 'Schizo': 'SZ'})
    y0 = df[gene_id].quantile(.01) - 0.26
    y1 = df[gene_id].quantile(.99) + 0.26
    pjd = position_jitterdodge(jitter_width=0.27)
    p = ggplot(df, aes(x=snp_id, y=gene_id, fill=pheno_var)) \
    + geom_boxplot(alpha=0.4, outlier_alpha=0) \
    + geom_jitter(position=pjd, stroke=0, alpha=0.6) + ylim(y0, y1) \
    + labs(y='Residualized expression', fill='Diagnosis') \
    + theme_bw(base_size=20)\
    + theme(legend_title=element_text(face='bold'),
        panel_grid_major=element_blank(),

```

```

        panel_grid_minor=element_blank())
    return p

def simple_gwas_ordered_snp_expression_pheno_plot(snp_id, gene_id, pheno_var):
    return simple_snp_expression_pheno_plot_impl(snp_id, gene_id,
    ↪get_gwas_ordered_snp_df, pheno_var)

def gwas_annotation(snp_id):
    return 'SZ GWAS pvalue: %.1e' % get_gwas_snp(snp_id).iloc[0]['P']

def eqtl_annotation(snp_id, gene_id):
    r = get_eqtl_df()[
        (get_eqtl_df()['variant_id']==snp_id) &
        (get_eqtl_df()['gene_id']==gene_id)]
    assert len(r)==1
    return 'eQTL nominal p-value: %.1e' % r.iloc[0]['pval_nominal']

def de_annotation(gene_id):
    g = get_de_df()[
        (get_de_df()['gencodeID'] == gene_id)]
    return 'DE adj.P.Val: %.3f' % g.iloc[0]['adj.P.Val']

def risk_allele_annotation(snp_id):
    return 'SZ risk allele: %s' % get_risk_allele(snp_id)

def gwas_annotated_eqtl_pheno_plot(snp_id, gene_id, pheno_var):
    p = simple_gwas_ordered_snp_expression_pheno_plot(snp_id, gene_id,
    ↪pheno_var)
    gene_symbol, gene_description = get_gene_symbol(gene_id)
    title = "\n".join([gene_symbol, gene_id,
                        gwas_annotation(snp_id),
                        risk_allele_annotation(snp_id),
                        eqtl_annotation(snp_id, gene_id),
                        de_annotation(gene_id)])

    p += ggtitle(title)
    return p

```

## 1.2 Genes

```
[7]: try:
      os.makedirs(feature)
    except OSError as e:
        if e.errno != errno.EEXIST:
            raise
```

### 1.2.1 Enrichment

#### Integrate DEG with PGC2+CLOZUK SNPs

```
[8]: dft = get_integration_df()
     dft.shape
```

/home/jbenja13/.local/lib/python3.9/site-packages/numpy/lib/arraysetops.py:583:  
FutureWarning: elementwise comparison failed; returning scalar instead, but in  
the future will perform elementwise comparison

```
[8]: (1616724, 52)
```

```
[9]: dft['agree_direction'] = dft.apply(agree_direction, axis=1)
     agreement = {1: 'Yes', -1: 'No', 0: 0}
     dft.agree_direction = [agreement[item] for item in dft['agree_direction']]
     ## Enrichment test
     table = [[np.sum((dft['P']<5e-8) & ((dft['adj.P.Val']<.05))),
               np.sum((dft['P']<5e-8) & ((dft['adj.P.Val']>=.05))),
               np.sum((dft['P']>=5e-8) & ((dft['adj.P.Val']<.05))),
               np.sum((dft['P']>=5e-8) & ((dft['adj.P.Val']>=.05)))]]
     print(table)
     fisher_exact(table)
```

```
[[3020, 33767], [168656, 1411281]]
```

```
[9]: (0.74838687753065, 1.3150145933372704e-55)
```

```
[10]: dft1 = dft[(dft['P']<5e-8) & ((dft['adj.P.Val']<.05))]
     df = dft1.groupby('agree_direction').size().reset_index()
     df
```

```
[10]:  agree_direction    0
     0                No   688
     1                Yes  2332
```

```
[11]: binom_test(df[0].iloc[1], df[0].sum())
```

```
[11]: 2.4968994585676953e-207
```

```
[12]: dft2 = dft[(dft['P']<=5e-8) & (dft['adj.P.Val'] < 0.05)].copy()
     dft2.groupby("gene_id").first().reset_index().shape
```



[12]: (35, 53)

```
[13]: dft2['risk_allele'] = dft2['our_snp_id'].apply(get_risk_allele)
```

```
[14]: direction = {-1: 'Down', 1: 'Up'}
boolean_conv = {True: 1, False: -1}
dft2.pgc2_a1_same_as_our_counted = [boolean_conv[item] for item in np.
    →dft2['pgc2_a1_same_as_our_counted']]
dft2['eqtl_gwas_dir'] = [direction[item] for item in np.
    →sign(dft2['pgc2_a1_same_as_our_counted']) * np.sign(dft2['slope']) * np.
    →sign(dft2['OR'] - 1)]
dft2['de_dir'] = [direction[item] for item in np.sign(dft2['t'])]
dft2['eqtl_slope'] = np.sign(dft2['pgc2_a1_same_as_our_counted']) * np.
    →sign(dft2['OR'] - 1) * dft2['slope']
dft3 = dft2[['gene_id', 'Symbol', 'variant_id', 'A1', 'A2', 'risk_allele', 'OR',
    'P', 'pval_nominal', 'adj.P.Val', 'logFC', 't', 'eqtl_slope',
    'de_dir', 'eqtl_gwas_dir', 'agree_direction']]
dft3['Symbol'].fillna(dft3['gene_id'], inplace=True)
dft3.to_csv('%s/integration_by_symbol.txt' % feature, sep='\t', index=False)
```

/home/jbenja13/.local/lib/python3.9/site-packages/pandas/core/series.py:4463:

SettingWithCopyWarning:

A value is trying to be set on a copy of a slice from a DataFrame

See the caveats in the documentation: [https://pandas.pydata.org/pandas-docs/stable/user\\_guide/indexing.html#returning-a-view-versus-a-copy](https://pandas.pydata.org/pandas-docs/stable/user_guide/indexing.html#returning-a-view-versus-a-copy)

```
[15]: #dft3 = pd.read_csv("../m/genes/integration_by_symbol.txt", sep='\t')
#dft3['Symbol'].fillna(dft3['gene_id'], inplace=True)
df2 = dft3.groupby(['gene_id']).first().reset_index().sort_values('P')
df2.groupby(['agree_direction']).size()
```

```
[15]: agree_direction
No      13
Yes     22
dtype: int64
```

```
[16]: df2.set_index('Symbol').rename(columns={'t': 'de_t', 'P': 'GWAS_P',
    →'pval_nominal': 'eQTL_pvalue',
    'adj.P.Val': 'de_FDR'})
```

```
[16]:
```

	gene_id	variant_id	A1	A2	\
Symbol					
ZSCAN9	ENSG00000137185.11	chr6:27731058:C:T	C	T	
HCG4	ENSG00000176998.4	chr6:29298706:G:A	G	A	
BRD2	ENSG00000204256.12	chr6:32669525:G:A	G	A	
FLOT1	ENSG00000137312.14	chr6:30406434:A:G	A	G	
HCG11	ENSG00000228223.2	chr6:26466161:G:A	G	A	

BAG6	ENSG00000204463.12	chr6:31222761:G:A	G	A
NELFE	ENSG00000204356.12	chr6:31868665:A:G	A	G
ZNF204P	ENSG00000204789.4	chr6:27343879:A:G	A	G
NGEF	ENSG00000066248.14	chr2:232843683:G:A	G	A
PRRC2A	ENSG00000204469.12	chr6:31379292:C:T	C	T
PLCH2	ENSG00000149527.17	chr1:2440958:A:G	A	G
IP6K3	ENSG00000161896.11	chr6:33541236:A:G	A	G
DNAJC19	ENSG00000205981.6	chr3:181087982:C:G	C	G
LINC01470	ENSG00000249484.8	chr5:152727548:A:G	A	G
CNNM2	ENSG00000148842.17	chr10:102825368:C:A	C	A
ZNF391	ENSG00000124613.8	chr6:26990749:T:C	T	C
PLPP5	ENSG00000147535.16	chr8:38263203:T:C	T	C
PTN	ENSG00000105894.11	chr7:137363413:T:C	T	C
TRMT61A	ENSG00000166166.12	chr14:103541799:CAGG:C	CAGG	C
PCCB	ENSG00000114054.13	chr3:136088767:G:T	G	T
PPTC7	ENSG00000196850.5	chr12:110126854:A:C	A	C
TSNARE1	ENSG00000171045.14	chr8:142261784:C:T	C	T
HIRIP3	ENSG00000149929.15	chr16:29959536:C:G	C	G
PPM1M	ENSG00000164088.17	chr3:52253452:G:T	G	T
VRK2	ENSG00000028116.16	chr2:57874972:T:A	T	A
ZC3H7B	ENSG00000100403.11	chr22:41357599:G:A	G	A
REEP2	ENSG00000132563.15	chr5:138362610:C:T	C	T
KDM3B	ENSG00000120733.13	chr5:138362610:C:T	C	T
SREBF2	ENSG00000198911.11	chr22:41830391:C:T	C	T
BNIP3L	ENSG00000104765.15	chr8:26368096:G:A	G	A
ENSG00000228944.1	ENSG00000228944.1	chr7:24695385:G:C	G	C
PHF1	ENSG00000112511.17	chr6:33343202:G:A	G	A
C4A	ENSG00000244731.7	chr6:31482137:T:C	T	C
ZNF14	ENSG00000105708.8	chr19:19623068:T:C	T	C
ENSG00000253553.5	ENSG00000253553.5	chr8:88271460:A:G	A	G

	risk_allele	OR	GWAS_P	eQTL_pvalue \
Symbol				
ZSCAN9	C	1.26220	1.210000e-39	8.946610e-04
HCG4	G	1.26360	2.500000e-39	4.443110e-05
BRD2	G	1.21970	6.820000e-30	1.131810e-05
FLOT1	A	1.23630	2.800000e-27	6.289760e-04
HCG11	A	0.91432	1.020000e-14	1.374100e-05
BAG6	A	0.92265	6.790000e-14	8.199240e-05
NELFE	A	1.07930	1.660000e-13	7.305820e-04
ZNF204P	G	0.93306	4.310000e-13	1.058040e-03
NGEF	A	0.91758	7.130000e-13	2.415450e-04
PRRC2A	C	1.08610	7.930000e-12	4.021460e-04
PLCH2	G	0.92873	4.630000e-11	2.811440e-09
IP6K3	G	0.93912	2.350000e-10	1.762100e-04
DNAJC19	C	1.07600	7.190000e-10	4.418510e-04
LINC01470	A	1.06910	8.730000e-10	5.630250e-14

CNNM2	C	1.06040	1.120000e-09	2.413790e-11
ZNF391	C	0.94335	1.550000e-09	1.247880e-03
PLPP5	T	1.06970	2.490000e-09	3.233620e-04
PTN	C	0.94234	2.770000e-09	2.497190e-04
TRMT61A	C	0.93885	4.218000e-09	1.193570e-04
PCCB	T	0.94493	5.050000e-09	2.185890e-14
PPTC7	A	1.06290	5.810000e-09	7.078260e-04
TSNARE1	C	1.07400	1.150000e-08	4.723740e-05
HIRIP3	C	1.05670	1.190000e-08	6.256180e-04
PPM1M	G	1.05600	1.350000e-08	6.597780e-04
VRK2	A	0.91021	1.370000e-08	6.451330e-04
ZC3H7B	A	0.94102	1.760000e-08	1.177050e-07
REEP2	C	1.05600	1.860000e-08	3.010360e-07
KDM3B	C	1.05600	1.860000e-08	1.455140e-04
SREBF2	C	1.05530	2.290000e-08	9.115040e-05
BNIP3L	A	0.93463	2.300000e-08	1.403510e-04
ENSG00000228944.1	C	0.91974	2.310000e-08	1.514450e-04
PHF1	A	0.94764	4.060000e-08	1.541960e-04
C4A	T	1.06010	4.120000e-08	1.330480e-04
ZNF14	T	1.06130	4.300000e-08	3.436760e-06
ENSG00000253553.5	A	1.06200	4.410000e-08	3.018090e-04

Symbol	de_FDR	logFC	de_t	eqtl_slope	de_dir \
ZSCAN9	2.262693e-02	-0.077130	-3.126621	-0.307218	Down
HCG4	1.090935e-02	0.231187	3.425733	0.374783	Up
BRD2	1.718619e-02	-0.051562	-3.239197	-0.404029	Down
FLOT1	1.347849e-02	-0.047316	-3.337230	-0.193544	Down
HCG11	1.181958e-03	0.114043	4.193600	0.211170	Up
BAG6	1.241493e-02	-0.044002	-3.373864	0.148294	Down
NELFE	2.507792e-02	-0.046326	-3.081897	0.090267	Down
ZNF204P	1.603175e-02	0.095817	3.267501	0.089913	Up
NGEF	9.144005e-03	0.104229	3.493550	-0.144167	Up
PRRC2A	1.283462e-03	-0.066383	-4.167993	-0.131287	Down
PLCH2	1.909382e-04	-0.169308	-4.727576	-0.174917	Down
IP6K3	2.435252e-03	-0.233018	-3.959823	0.131471	Down
DNAJC19	2.188006e-02	0.042839	3.140810	0.098382	Up
LINC01470	1.143030e-02	0.406227	3.407085	0.488819	Up
CNNM2	3.899296e-03	0.053701	3.799656	-0.169331	Up
ZNF391	2.054017e-02	0.082216	3.166580	-0.145940	Up
PLPP5	2.838093e-02	-0.073766	-3.026111	-0.209961	Down
PTN	7.972676e-03	-0.086066	-3.545516	0.122412	Down
TRMT61A	4.958693e-02	-0.058611	-2.775075	-0.127611	Down
PCCB	3.397384e-02	-0.058710	-2.946922	-0.246348	Down
PPTC7	3.514250e-03	0.090141	3.837368	-0.119268	Up
TSNARE1	4.121874e-03	-0.076791	-3.780711	0.177779	Down
HIRIP3	2.093547e-04	-0.091615	-4.698442	0.139753	Down

PPM1M	8.758407e-06	-0.136491	-5.484394	-0.116511	Down
VRK2	1.076112e-02	-0.127117	-3.431328	0.150703	Down
ZC3H7B	1.255326e-02	-0.055238	-3.367649	0.258939	Down
REEP2	6.655155e-09	0.137644	6.993625	0.145249	Up
KDM3B	1.518040e-02	-0.031591	-3.287837	-0.167676	Down
SREBF2	9.947151e-04	-0.075761	-4.243388	-0.177463	Down
BNIP3L	1.065324e-02	-0.063722	-3.435641	-0.101083	Down
ENSG00000228944.1	2.256942e-02	0.278474	3.127826	0.412318	Up
PHF1	2.281934e-02	-0.046170	-3.123441	-0.161077	Down
C4A	7.880375e-03	0.358141	3.550690	0.178003	Up
ZNF14	1.653178e-02	0.074769	3.254277	0.231031	Up
ENSG00000253553.5	2.909648e-02	0.202146	3.016220	-0.220670	Up

eqtl\_gwas\_dir agree\_direction

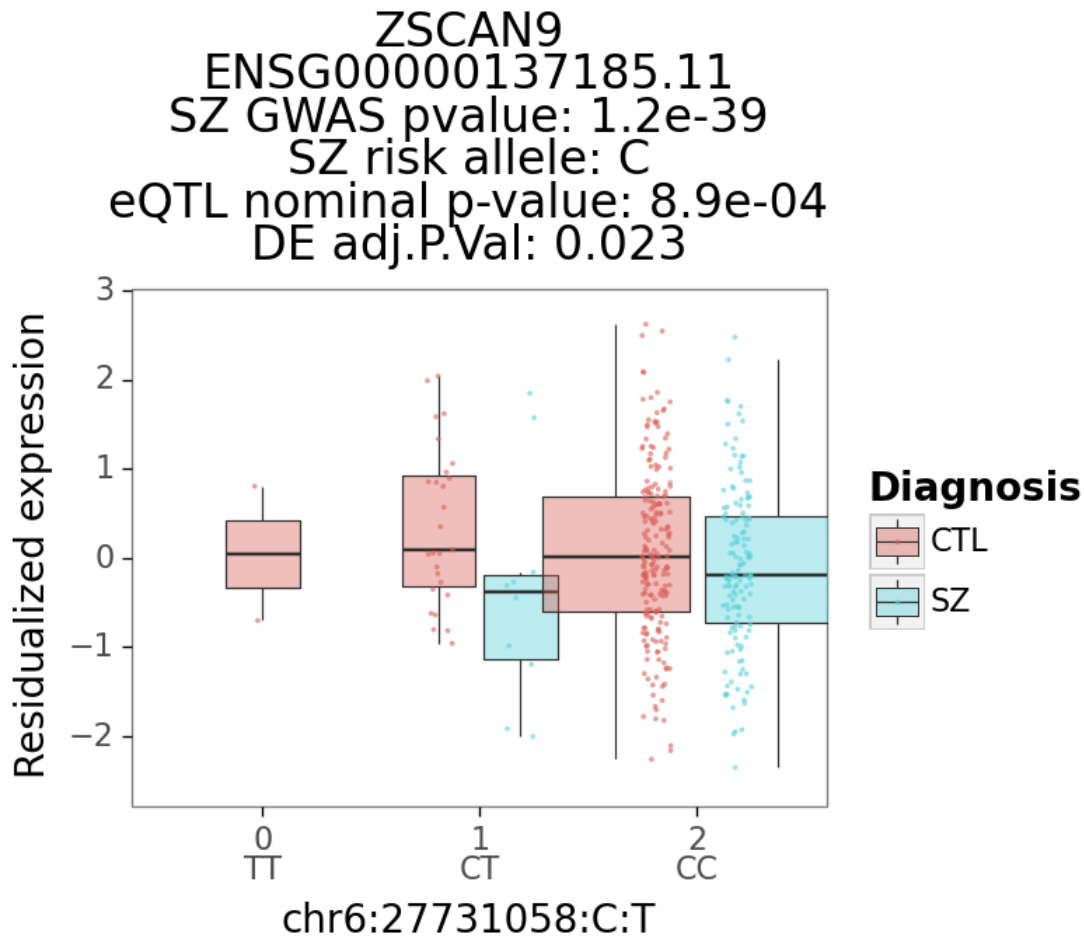
Symbol		
ZSCAN9	Down	Yes
HCG4	Up	Yes
BRD2	Down	Yes
FLOT1	Down	Yes
HCG11	Up	Yes
BAG6	Up	No
NELFE	Up	No
ZNF204P	Up	Yes
NGEF	Down	No
PRRC2A	Down	Yes
PLCH2	Down	Yes
IP6K3	Up	No
DNAJC19	Up	Yes
LINC01470	Up	Yes
CNNM2	Down	No
ZNF391	Down	No
PLPP5	Down	Yes
PTN	Up	No
TRMT61A	Down	Yes
PCCB	Down	Yes
PPTC7	Down	No
TSNARE1	Up	No
HIRIP3	Up	No
PPM1M	Down	Yes
VRK2	Up	No
ZC3H7B	Up	No
REEP2	Up	Yes
KDM3B	Down	Yes
SREBF2	Down	Yes
BNIP3L	Down	Yes
ENSG00000228944.1	Up	Yes
PHF1	Down	Yes

C4A	Up	Yes
ZNF14	Up	Yes
ENSG00000253553.5	Down	No

### 1.2.2 Plot with PGC2 risk allele

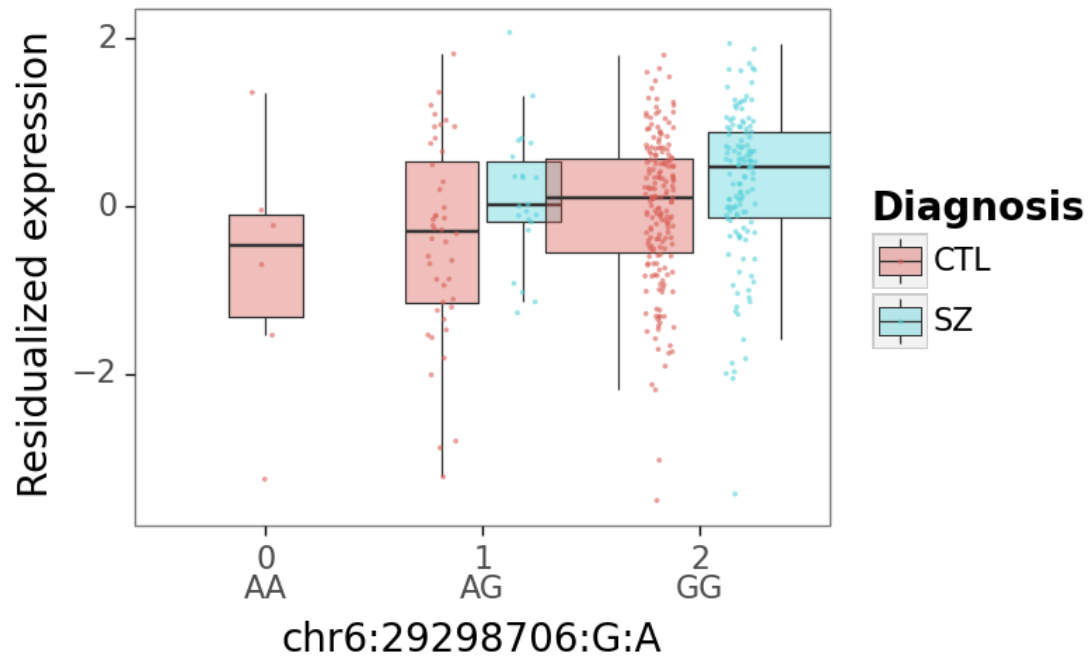
```
[17]: for xx in range(df2.shape[0]):
      gg = gwas_annotated_eqtl_pheno_plot(df2.iloc[xx, :].variant_id, df2.
      → iloc[xx, :].gene_id, 'Dx')
      print(gg)
      label = '%s/eqtl_gwas_%s' % (feature, df2.iloc[xx, :].Symbol)
      save_plot(gg, label)
```

Mapping files: 100% | 3/3 [00:25<00:00, 8.53s/it]



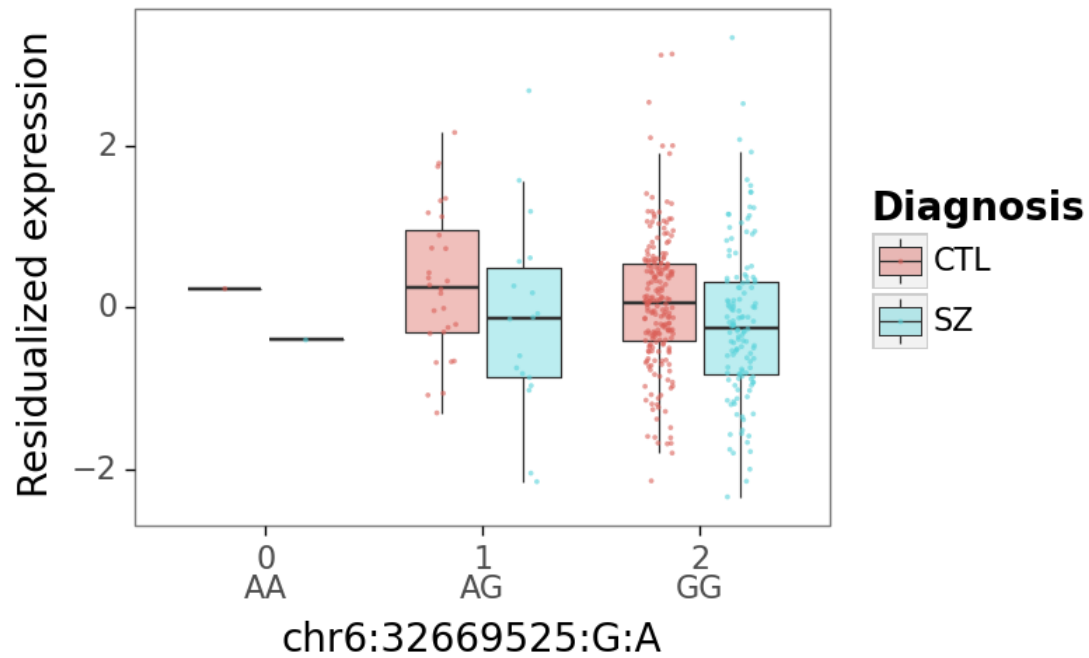
<ggplot: (8779647847280)>

HCG4  
 ENSG00000176998.4  
 SZ GWAS pvalue: 2.5e-39  
 SZ risk allele: G  
 eQTL nominal p-value: 4.4e-05  
 DE adj.P.Val: 0.011



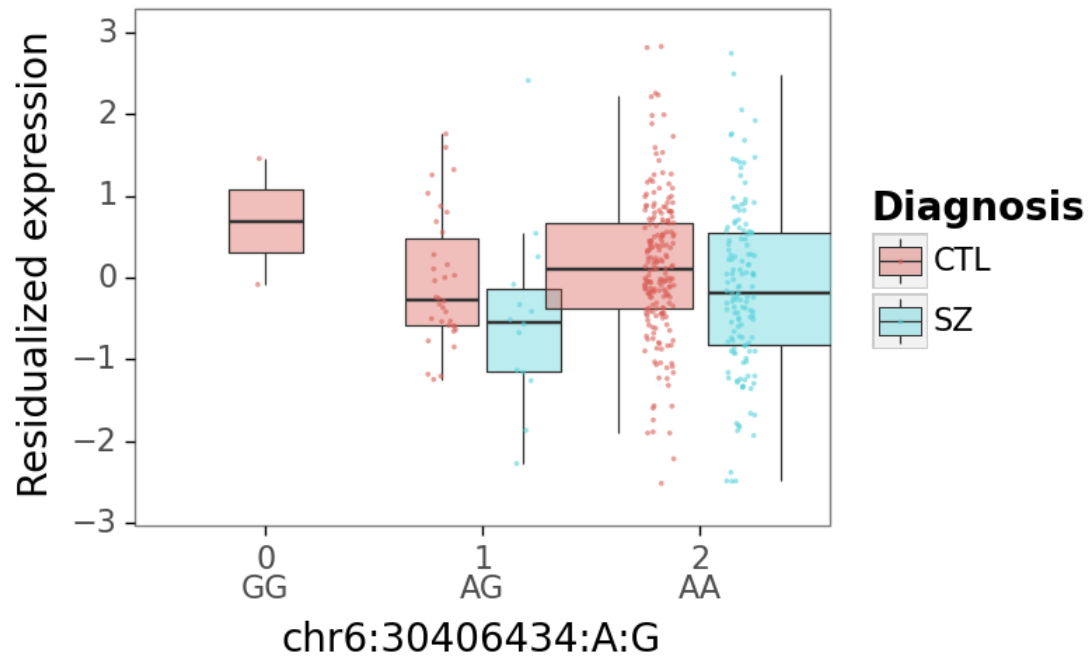
<ggplot: (8779648926365)>

BRD2  
 ENSG00000204256.12  
 SZ GWAS pvalue: 6.8e-30  
 SZ risk allele: G  
 eQTL nominal p-value: 1.1e-05  
 DE adj.P.Val: 0.017



<ggplot: (8779649122193)>

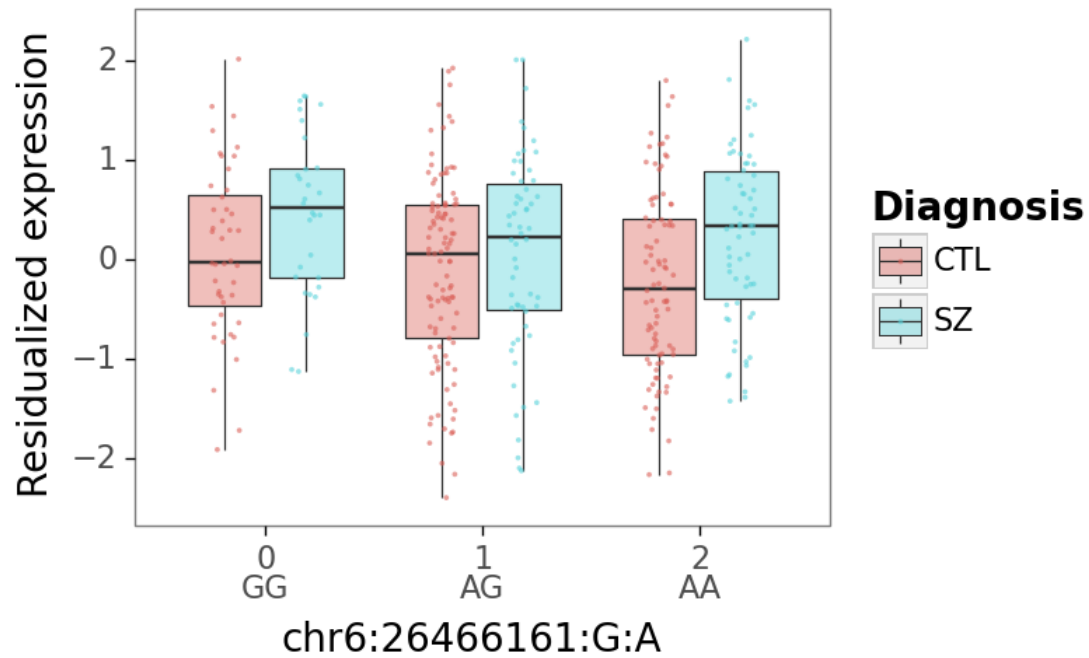
FLOT1  
ENSG00000137312.14  
SZ GWAS pvalue: 2.8e-27  
SZ risk allele: A  
eQTL nominal p-value: 6.3e-04  
DE adj.P.Val: 0.013



<ggplot: (8779673197017)>

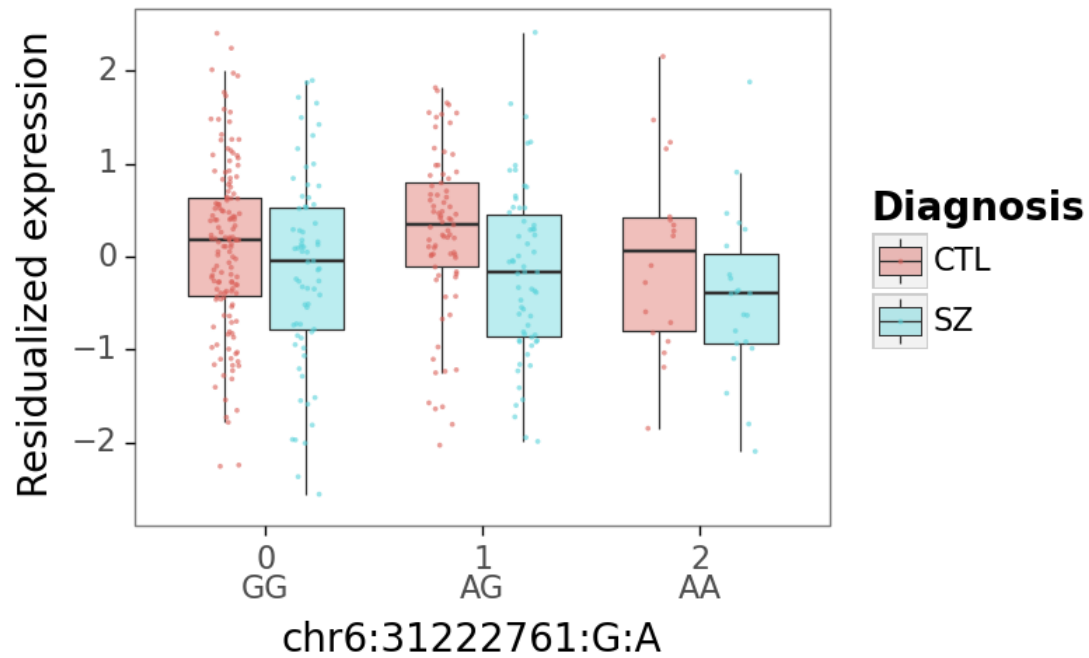


HCG11  
ENSG00000228223.2  
SZ GWAS pvalue: 1.0e-14  
SZ risk allele: A  
eQTL nominal p-value: 1.4e-05  
DE adj.P.Val: 0.001



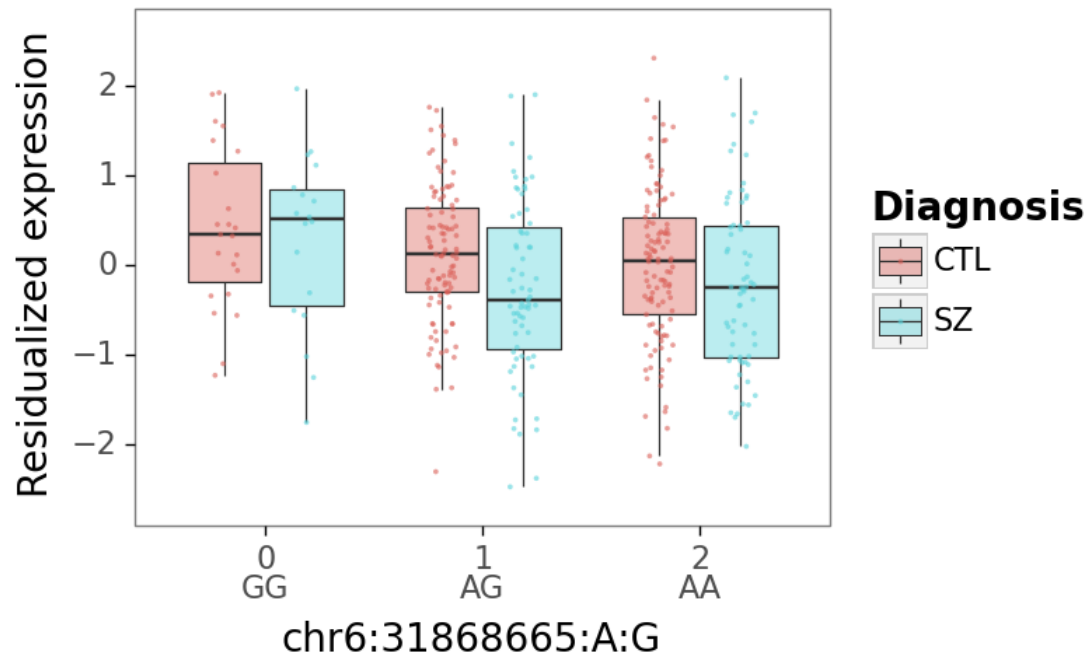
<ggplot: (8779673470455)>

BAG6  
 ENSG00000204463.12  
 SZ GWAS pvalue: 6.8e-14  
 SZ risk allele: A  
 eQTL nominal p-value: 8.2e-05  
 DE adj.P.Val: 0.012



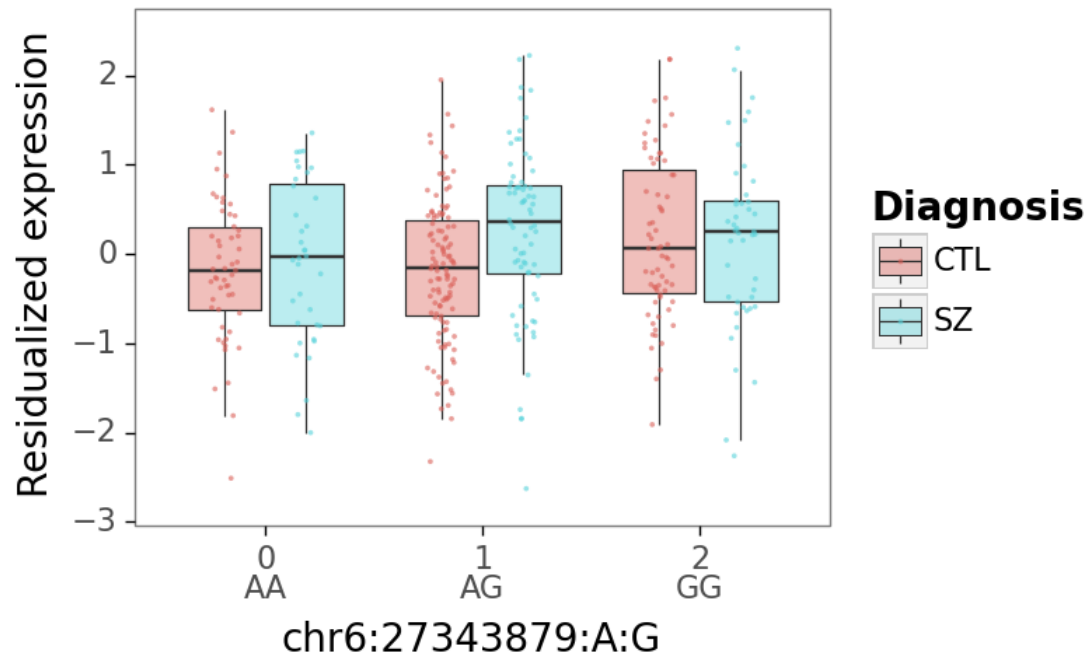
<ggplot: (8779647655783)>

NELFE  
ENSG00000204356.12  
SZ GWAS pvalue: 1.7e-13  
SZ risk allele: A  
eQTL nominal p-value: 7.3e-04  
DE adj.P.Val: 0.025



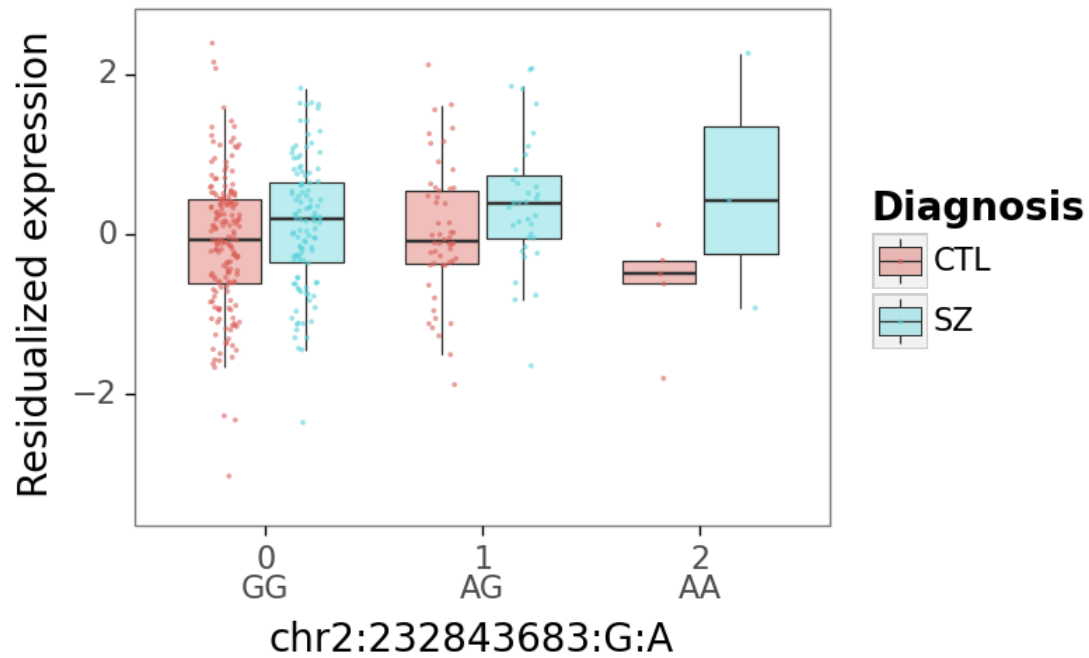
<ggplot: (8779616699468)>

ZNF204P  
ENSG00000204789.4  
SZ GWAS pvalue: 4.3e-13  
SZ risk allele: G  
eQTL nominal p-value: 1.1e-03  
DE adj.P.Val: 0.016



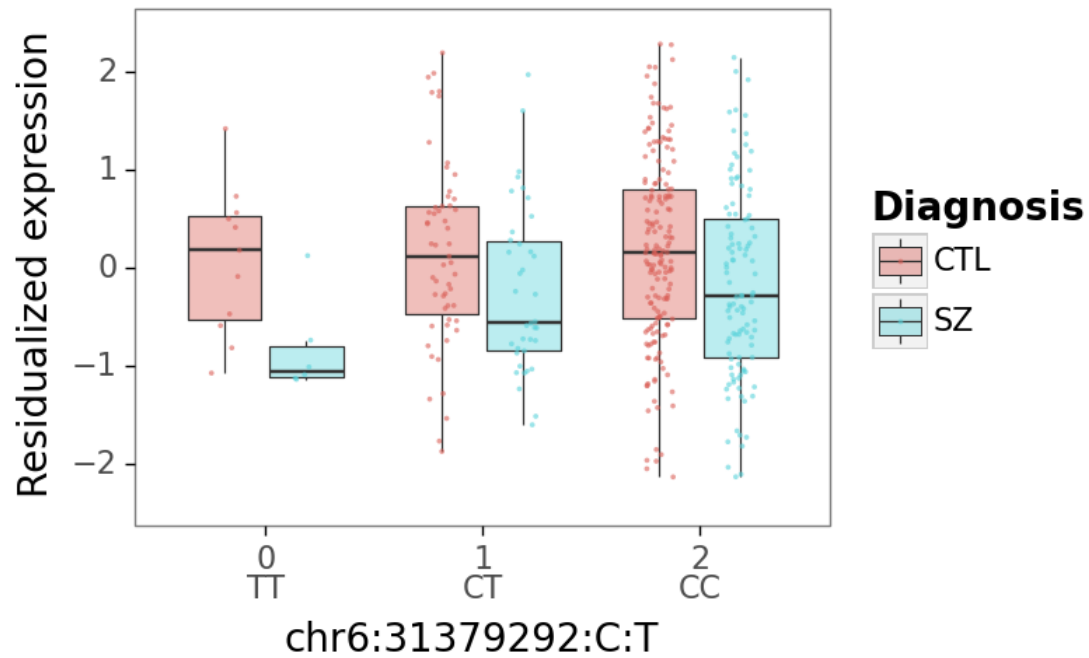
<ggplot: (8779647377445)>

NGEF  
 ENSG00000066248.14  
 SZ GWAS pvalue:  $7.1e-13$   
 SZ risk allele: A  
 eQTL nominal p-value:  $2.4e-04$   
 DE adj.P.Val: 0.009



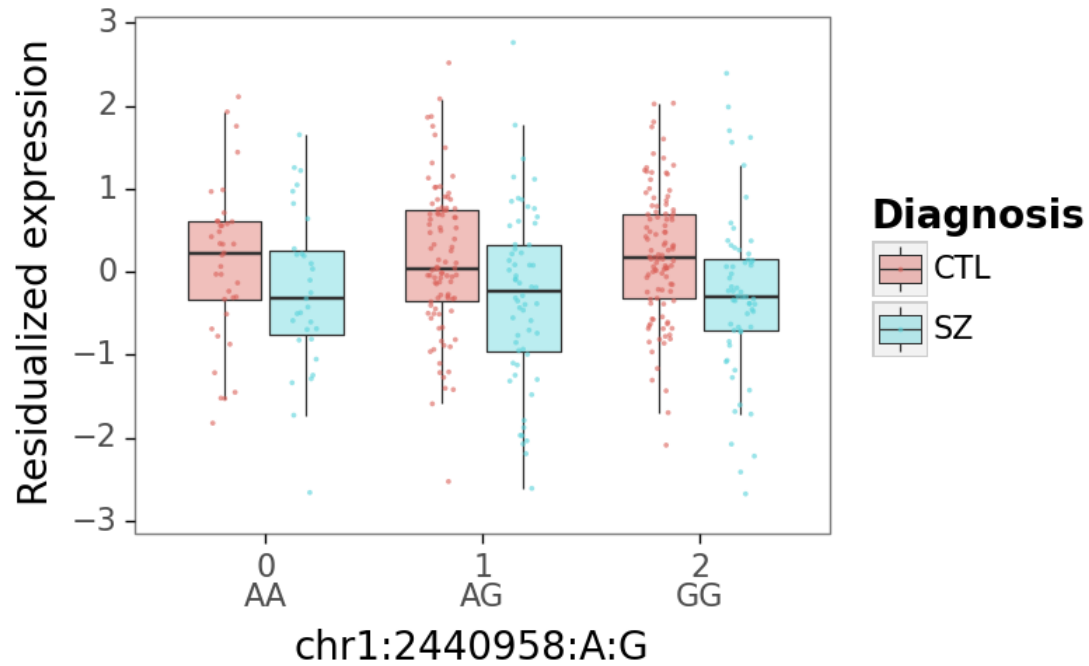
<ggplot: (8779674186150)>

PRRC2A  
 ENSG00000204469.12  
 SZ GWAS pvalue: 7.9e-12  
 SZ risk allele: C  
 eQTL nominal p-value: 4.0e-04  
 DE adj.P.Val: 0.001



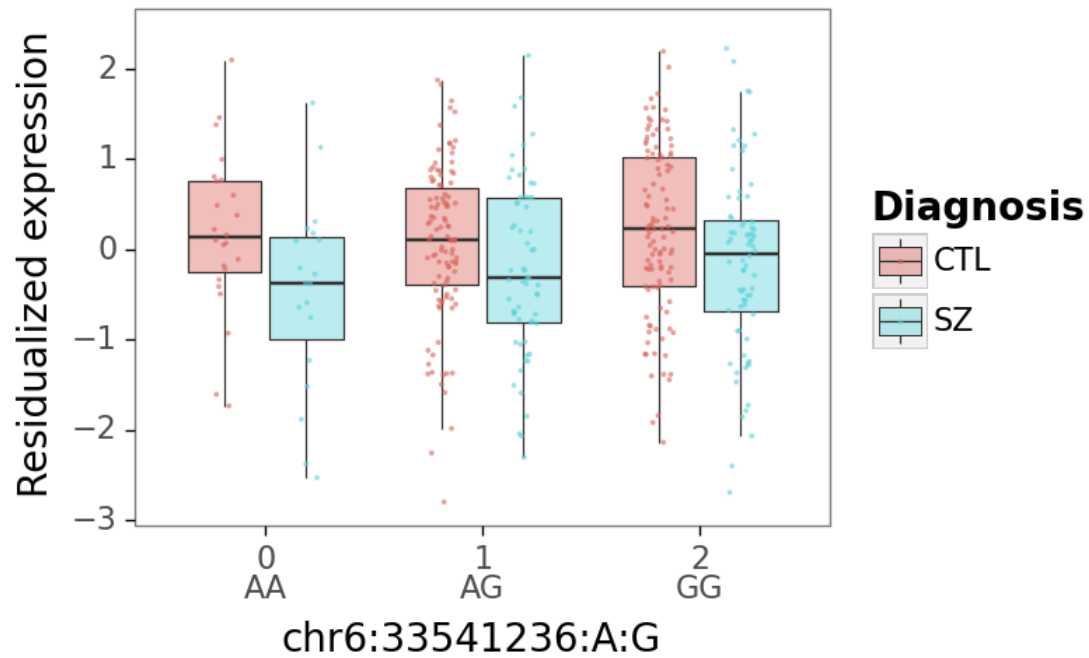
<ggplot: (8779648727333)>

PLCH2  
 ENSG00000149527.17  
 SZ GWAS pvalue: 4.6e-11  
 SZ risk allele: G  
 eQTL nominal p-value: 2.8e-09  
 DE adj.P.Val: 0.000



<ggplot: (8779648500149)>

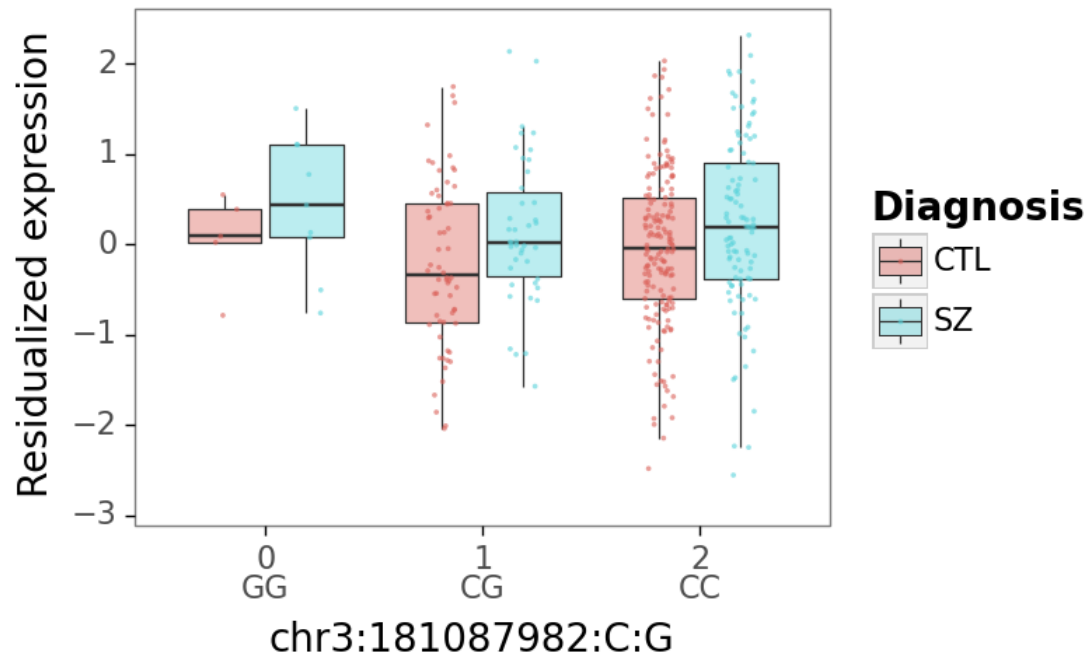
IP6K3  
 ENSG00000161896.11  
 SZ GWAS pvalue: 2.4e-10  
 SZ risk allele: G  
 eQTL nominal p-value: 1.8e-04  
 DE adj.P.Val: 0.002



<ggplot: (8779648120369)>

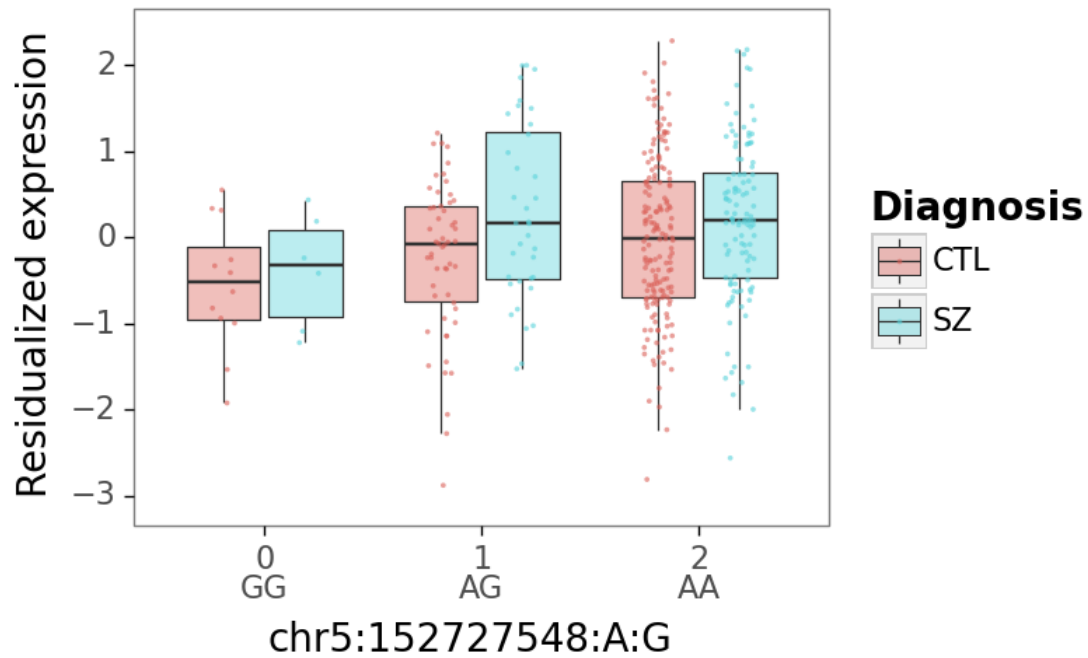


DNAJC19  
 ENSG00000205981.6  
 SZ GWAS pvalue:  $7.2 \times 10^{-10}$   
 SZ risk allele: C  
 eQTL nominal p-value:  $4.4 \times 10^{-4}$   
 DE adj.P.Val: 0.022



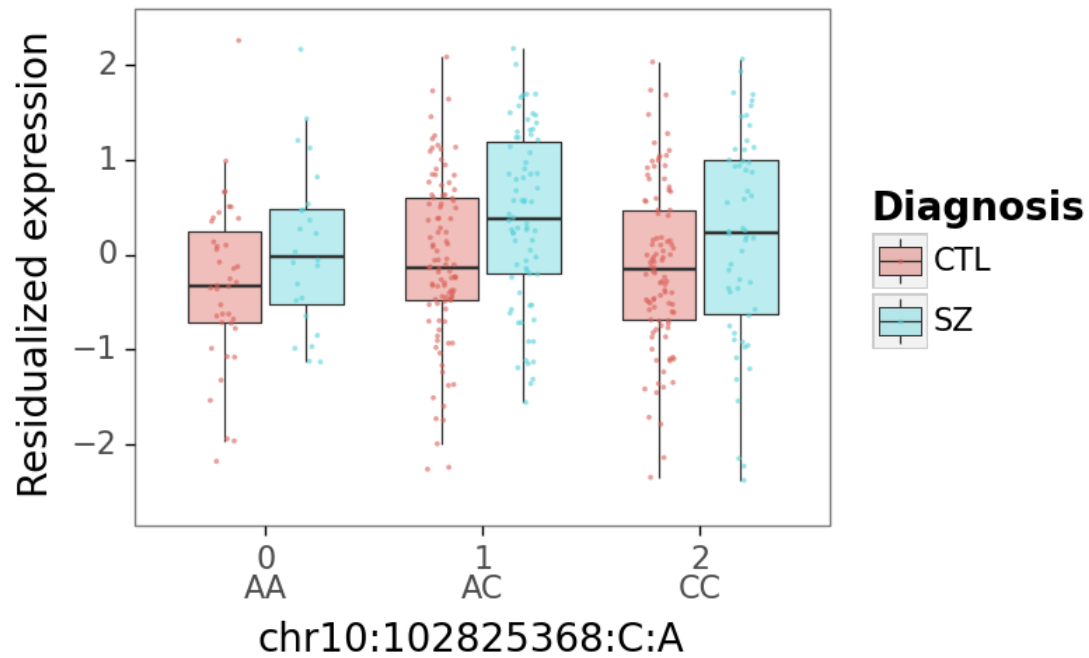
<ggplot: (8779615827688)>

LINC01470  
ENSG00000249484.8  
SZ GWAS pvalue: 8.7e-10  
SZ risk allele: A  
eQTL nominal p-value: 5.6e-14  
DE adj.P.Val: 0.011



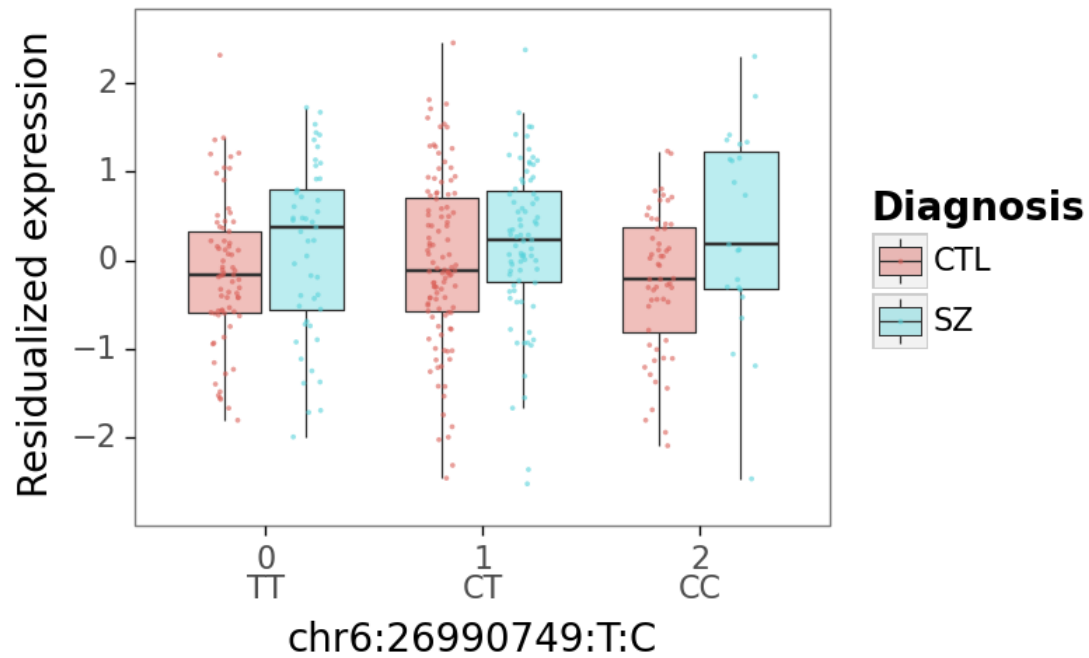
<ggplot: (8779648580172)>

CNNM2  
ENSG00000148842.17  
SZ GWAS pvalue: 1.1e-09  
SZ risk allele: C  
eQTL nominal p-value: 2.4e-11  
DE adj.P.Val: 0.004



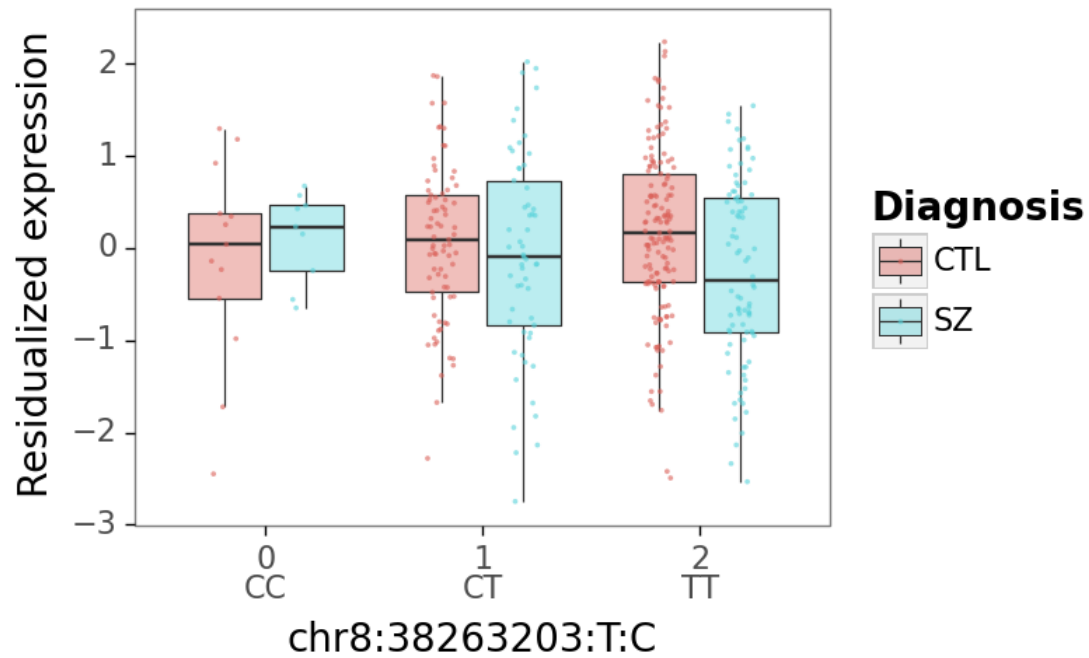
<ggplot: (8779674563202)>

ZNF391  
 ENSG00000124613.8  
 SZ GWAS pvalue: 1.6e-09  
 SZ risk allele: C  
 eQTL nominal p-value: 1.2e-03  
 DE adj.P.Val: 0.021



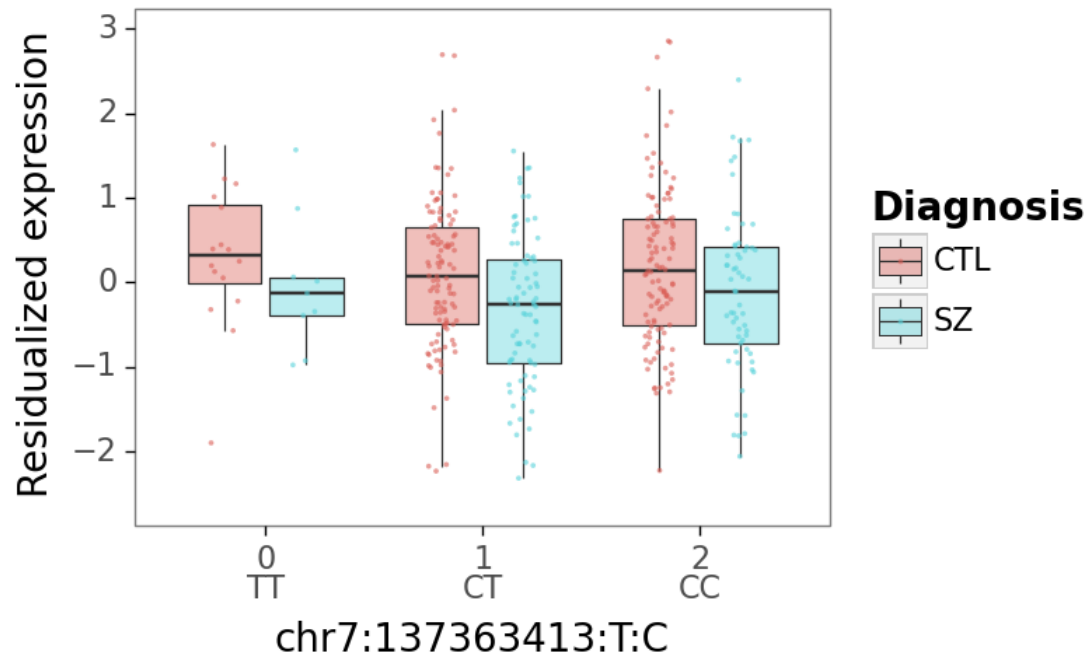
<ggplot: (8779647503433)>

PLPP5  
 ENSG00000147535.16  
 SZ GWAS pvalue: 2.5e-09  
 SZ risk allele: T  
 eQTL nominal p-value: 3.2e-04  
 DE adj.P.Val: 0.028



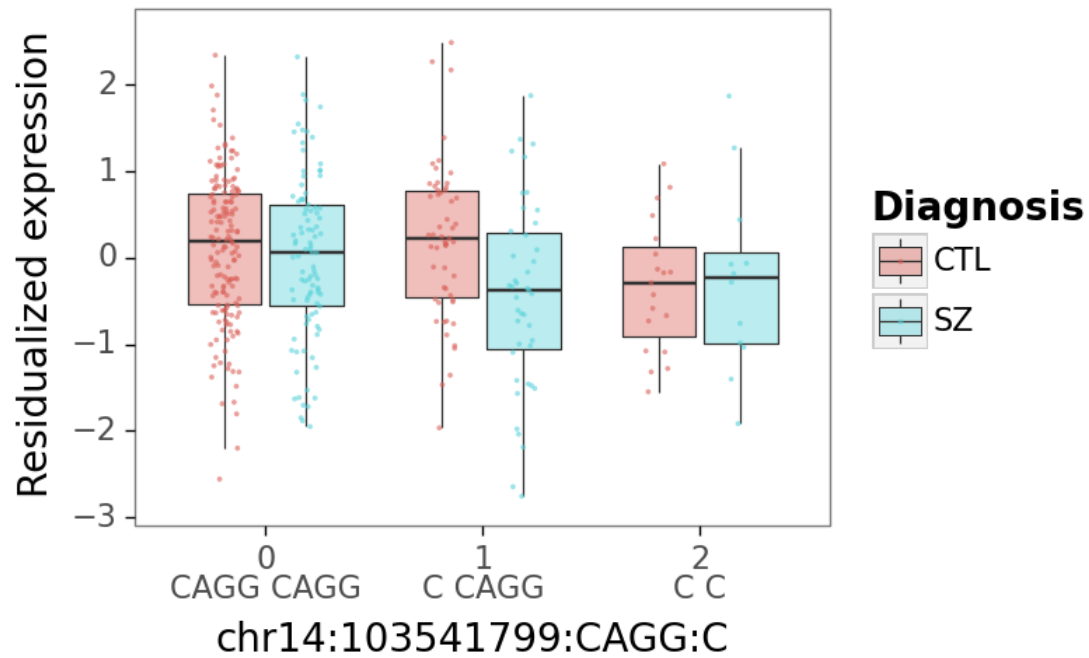
<ggplot: (8779674832619)>

PTN  
 ENSG00000105894.11  
 SZ GWAS pvalue: 2.8e-09  
 SZ risk allele: C  
 eQTL nominal p-value: 2.5e-04  
 DE adj.P.Val: 0.008



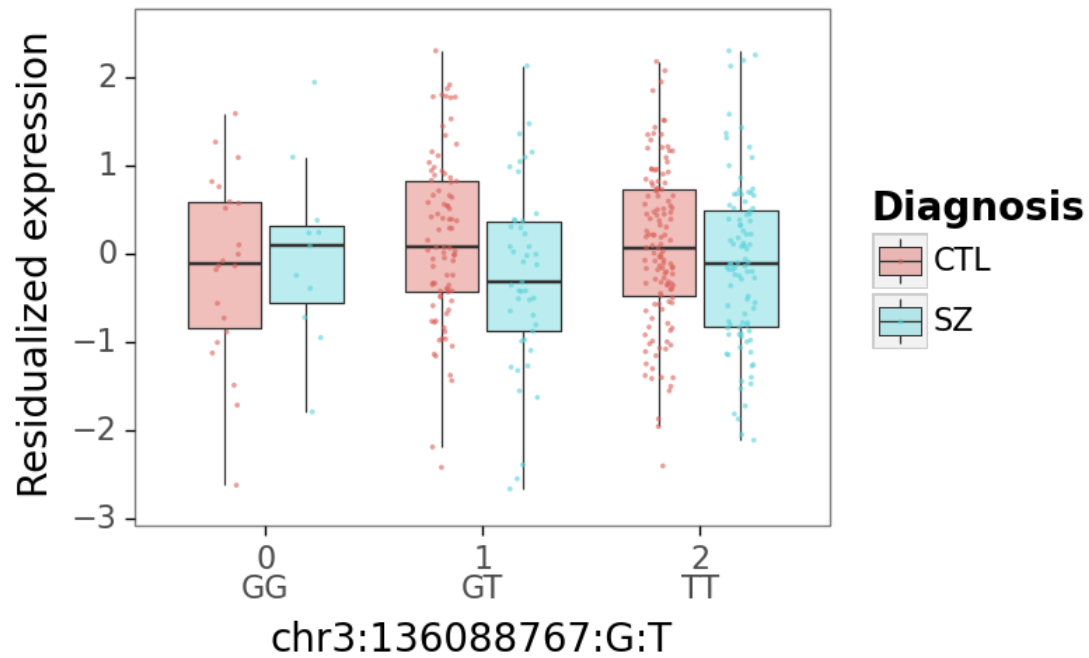
<ggplot: (8779673702925)>

TRMT61A  
 ENSG00000166166.12  
 SZ GWAS pvalue: 4.2e-09  
 SZ risk allele: C  
 eQTL nominal p-value: 1.2e-04  
 DE adj.P.Val: 0.050



<ggplot: (8779624369770)>

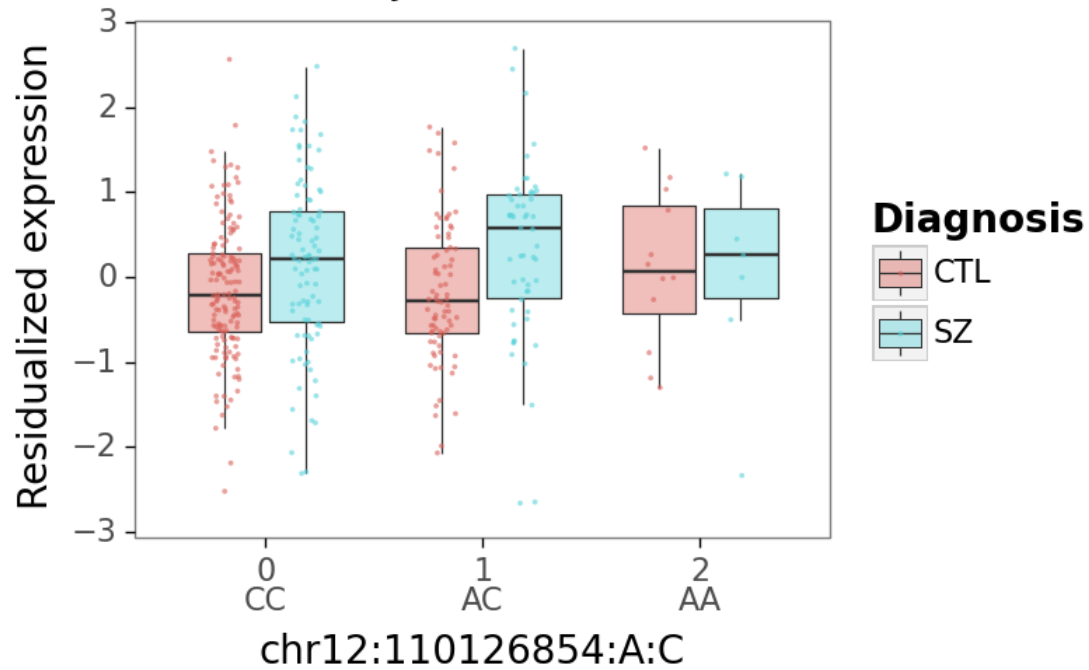
PCCB  
 ENSG00000114054.13  
 SZ GWAS pvalue: 5.0e-09  
 SZ risk allele: T  
 eQTL nominal p-value: 2.2e-14  
 DE adj.P.Val: 0.034



<ggplot: (8779627186728)>

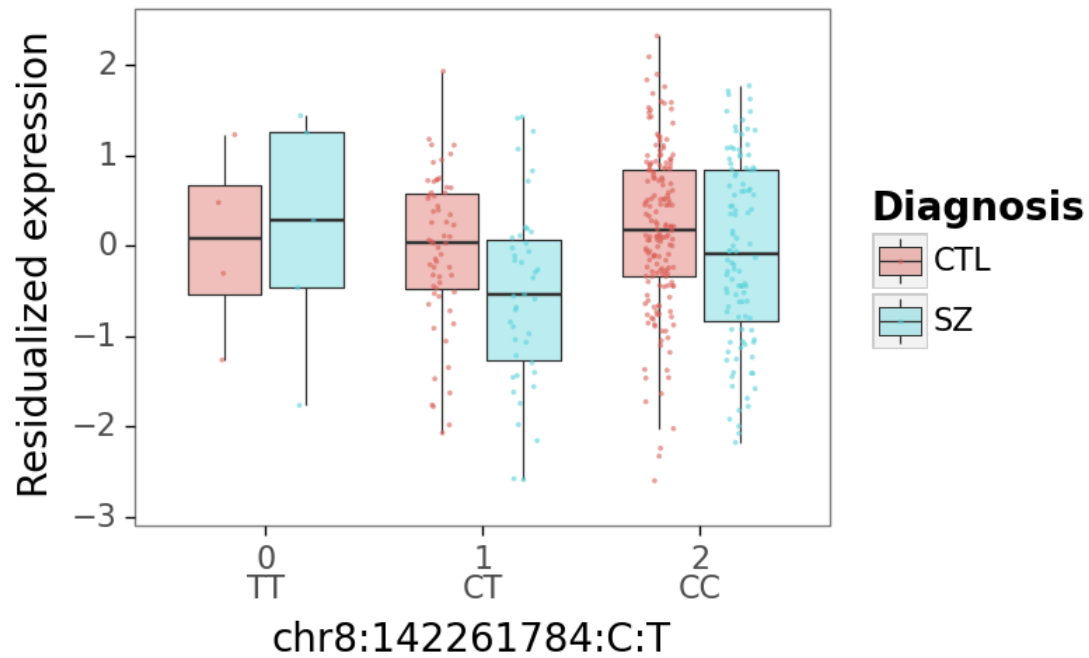


PPTC7  
 ENSG00000196850.5  
 SZ GWAS pvalue: 5.8e-09  
 SZ risk allele: A  
 eQTL nominal p-value: 7.1e-04  
 DE adj.P.Val: 0.004



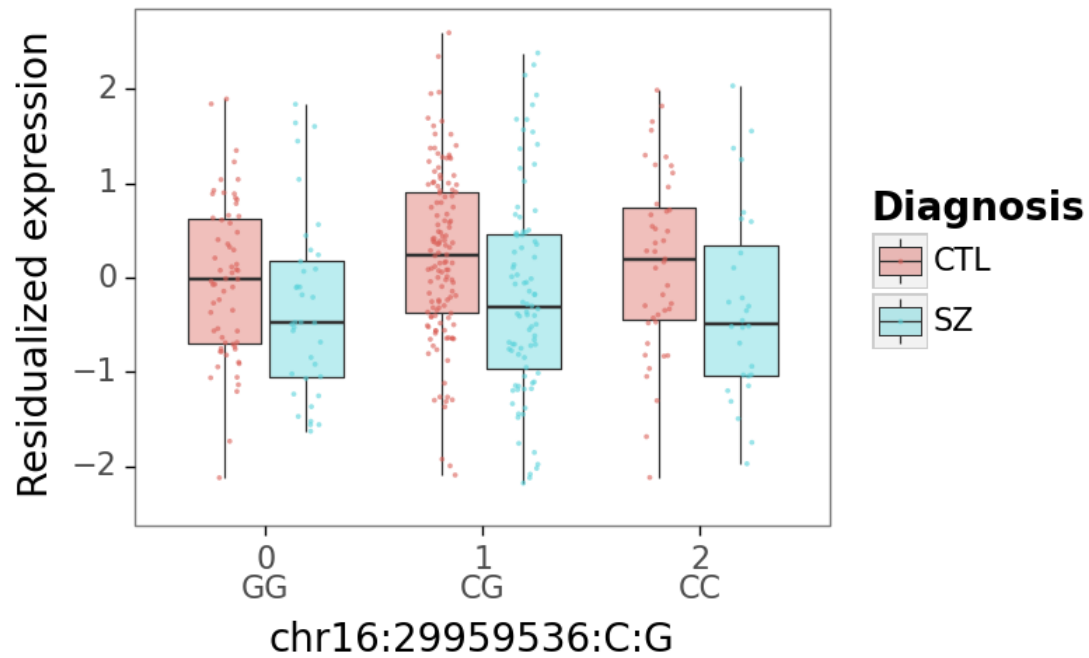
<ggplot: (8779617139191)>

TSNARE1  
ENSG00000171045.14  
SZ GWAS pvalue: 1.2e-08  
SZ risk allele: C  
eQTL nominal p-value: 4.7e-05  
DE adj.P.Val: 0.004



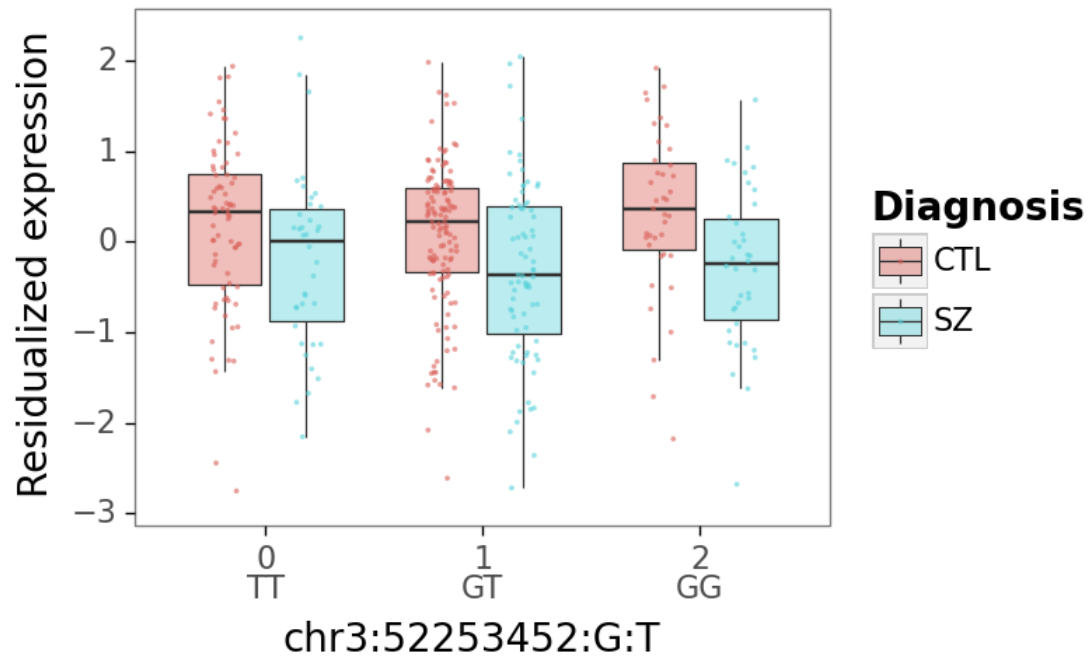
<ggplot: (8779616807331)>

HIRIP3  
ENSG00000149929.15  
SZ GWAS pvalue: 1.2e-08  
SZ risk allele: C  
eQTL nominal p-value: 6.3e-04  
DE adj.P.Val: 0.000



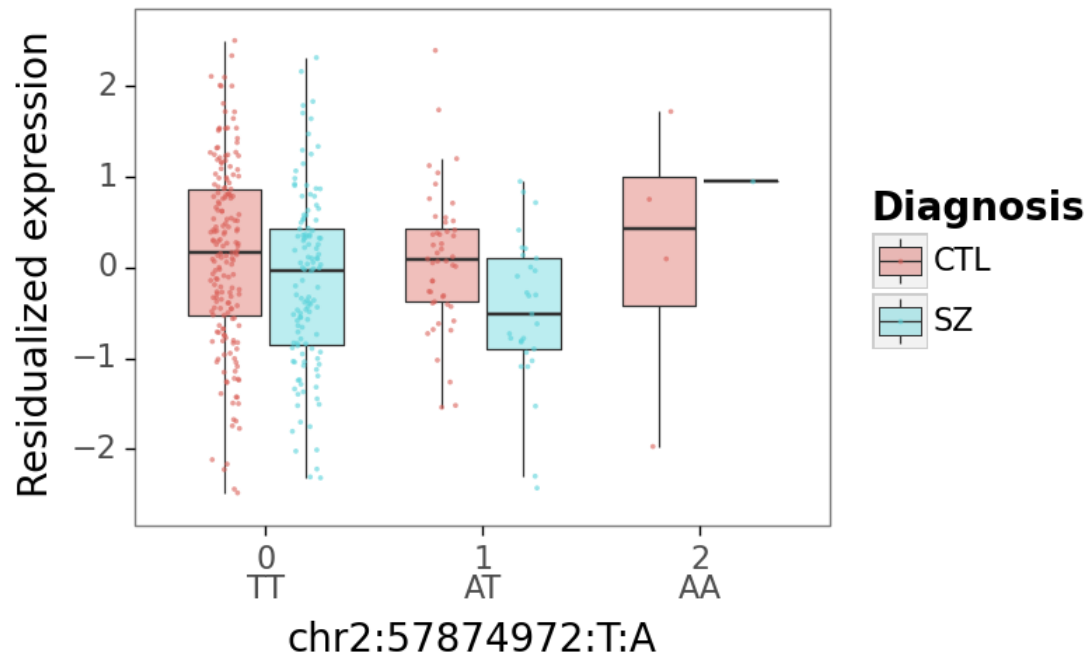
<ggplot: (8779648335812)>

PPM1M  
ENSG00000164088.17  
SZ GWAS pvalue: 1.4e-08  
SZ risk allele: G  
eQTL nominal p-value: 6.6e-04  
DE adj.P.Val: 0.000



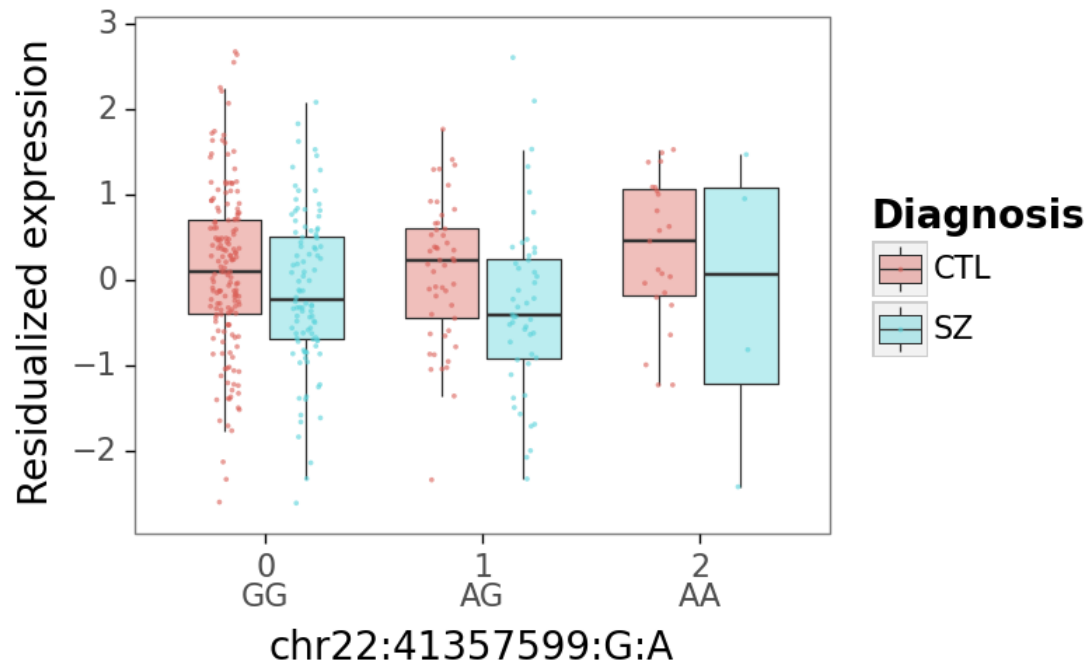
<ggplot: (8779672707127)>

VRK2  
 ENSG00000028116.16  
 SZ GWAS pvalue: 1.4e-08  
 SZ risk allele: A  
 eQTL nominal p-value: 6.5e-04  
 DE adj.P.Val: 0.011



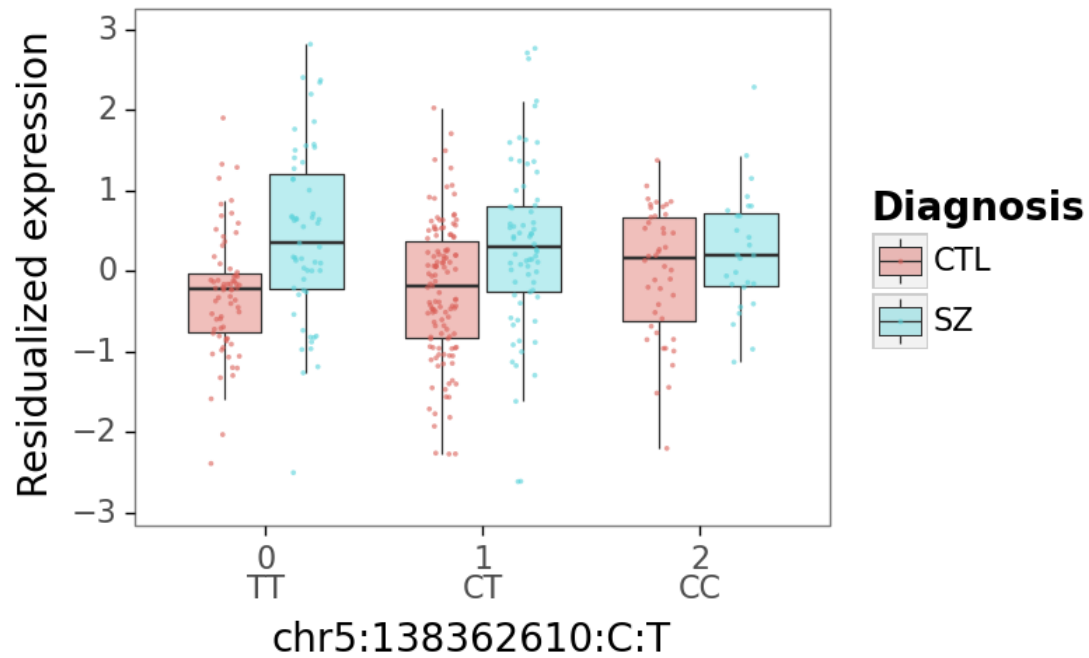
<ggplot: (8779674024744)>

ZC3H7B  
ENSG00000100403.11  
SZ GWAS pvalue: 1.8e-08  
SZ risk allele: A  
eQTL nominal p-value: 1.2e-07  
DE adj.P.Val: 0.013



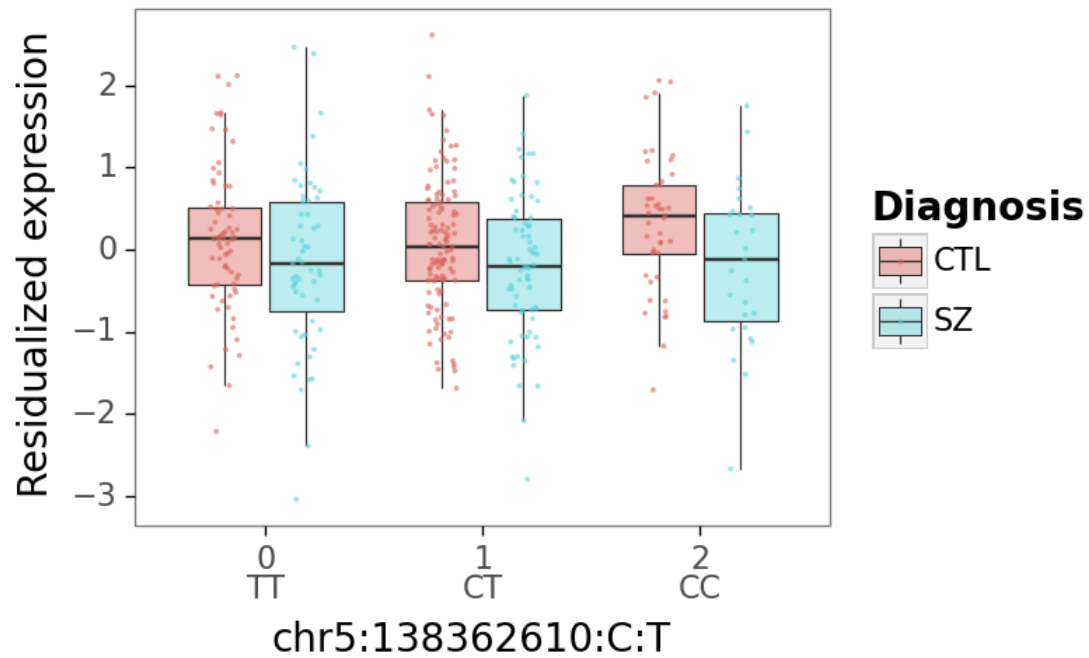
<ggplot: (8779673346380)>

REEP2  
 ENSG00000132563.15  
 SZ GWAS pvalue: 1.9e-08  
 SZ risk allele: C  
 eQTL nominal p-value: 3.0e-07  
 DE adj.P.Val: 0.000



<ggplot: (8779649073511)>

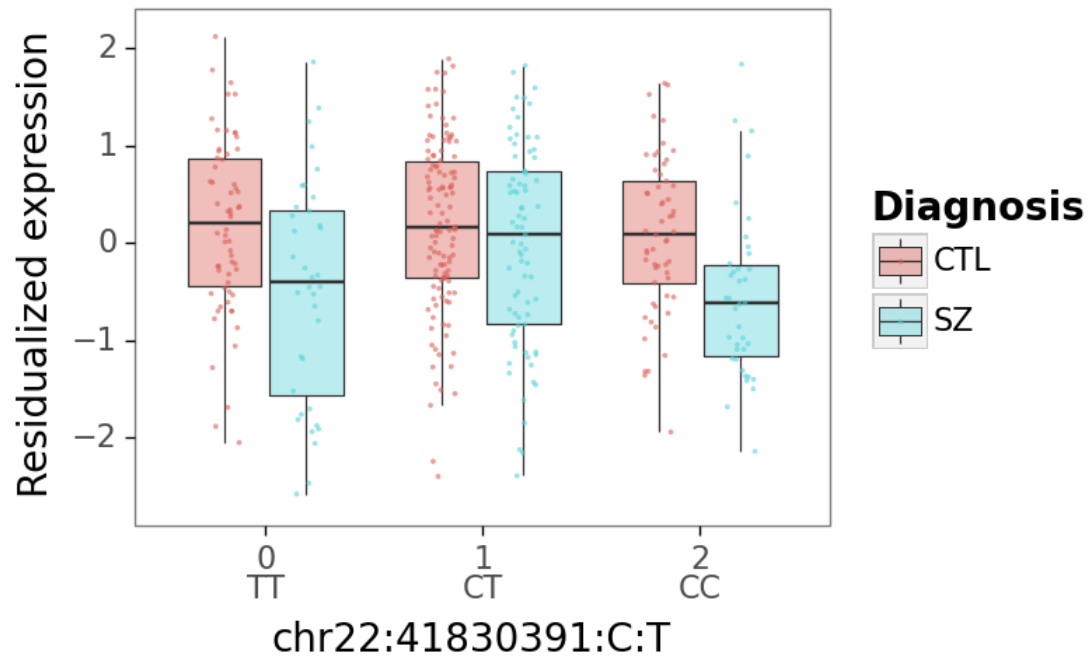
KDM3B  
ENSG00000120733.13  
SZ GWAS pvalue: 1.9e-08  
SZ risk allele: C  
eQTL nominal p-value: 1.5e-04  
DE adj.P.Val: 0.015



<ggplot: (8779647828902)>

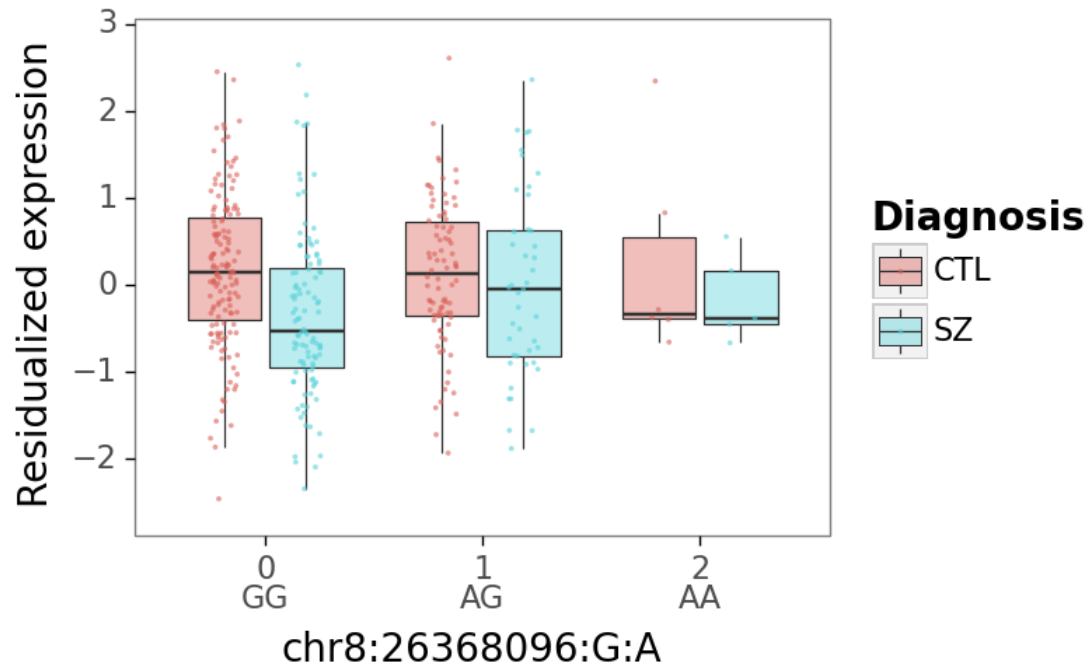


SREBF2  
 ENSG00000198911.11  
 SZ GWAS pvalue: 2.3e-08  
 SZ risk allele: C  
 eQTL nominal p-value: 9.1e-05  
 DE adj.P.Val: 0.001



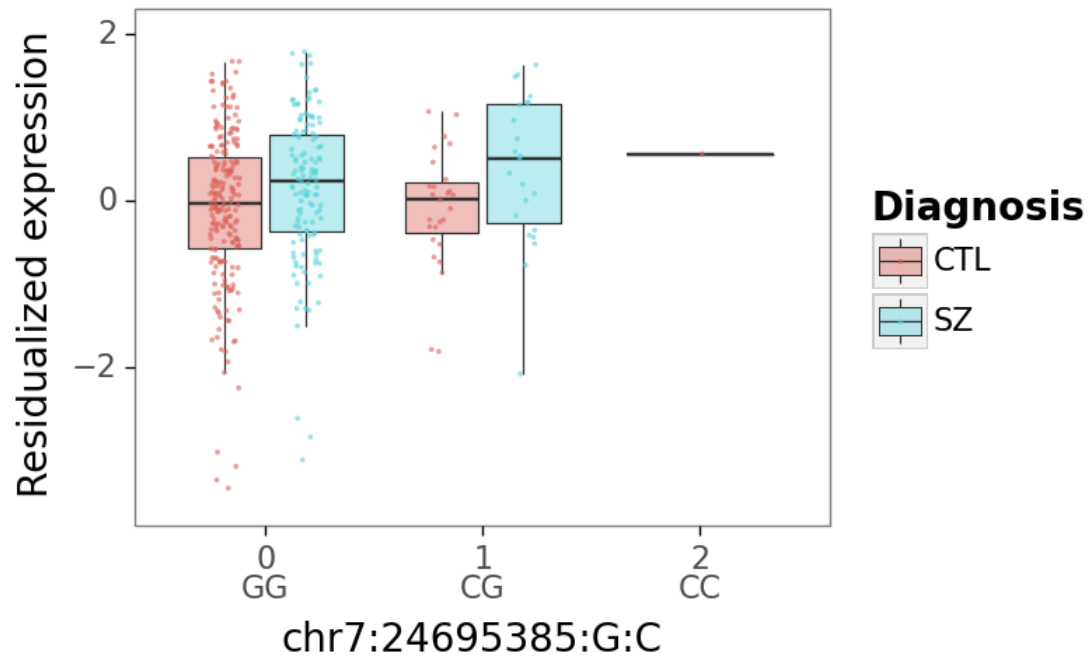
<ggplot: (8779647337539)>

BNIP3L  
 ENSG00000104765.15  
 SZ GWAS pvalue: 2.3e-08  
 SZ risk allele: A  
 eQTL nominal p-value: 1.4e-04  
 DE adj.P.Val: 0.011



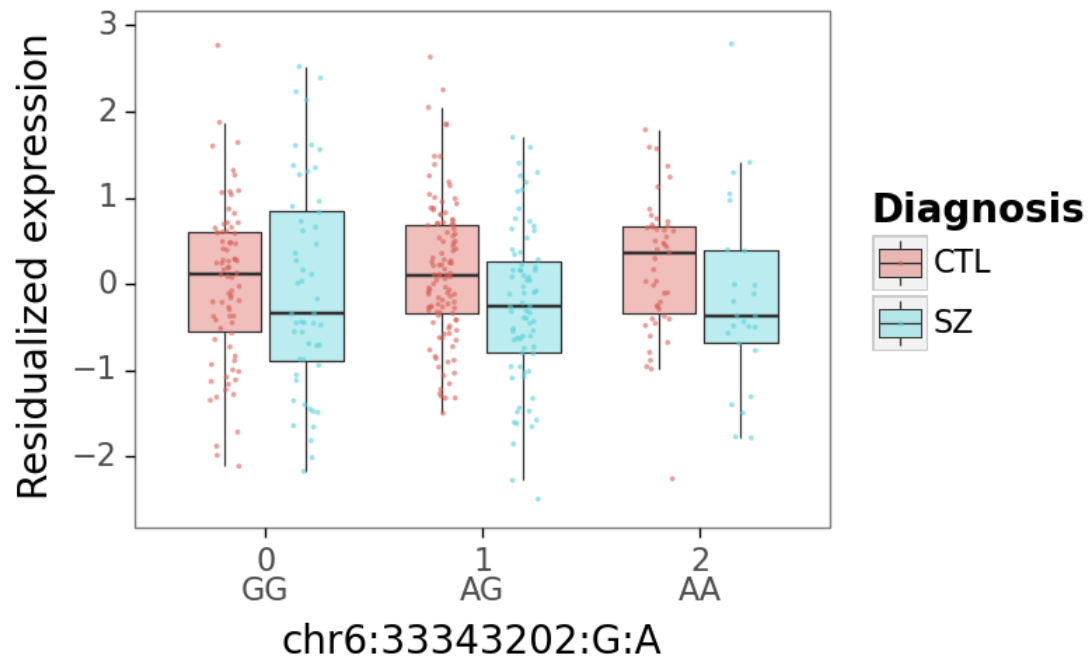
<ggplot: (8779672754827)>

AC004485.1  
 ENSG00000228944.1  
 SZ GWAS pvalue: 2.3e-08  
 SZ risk allele: C  
 eQTL nominal p-value: 1.5e-04  
 DE adj.P.Val: 0.023



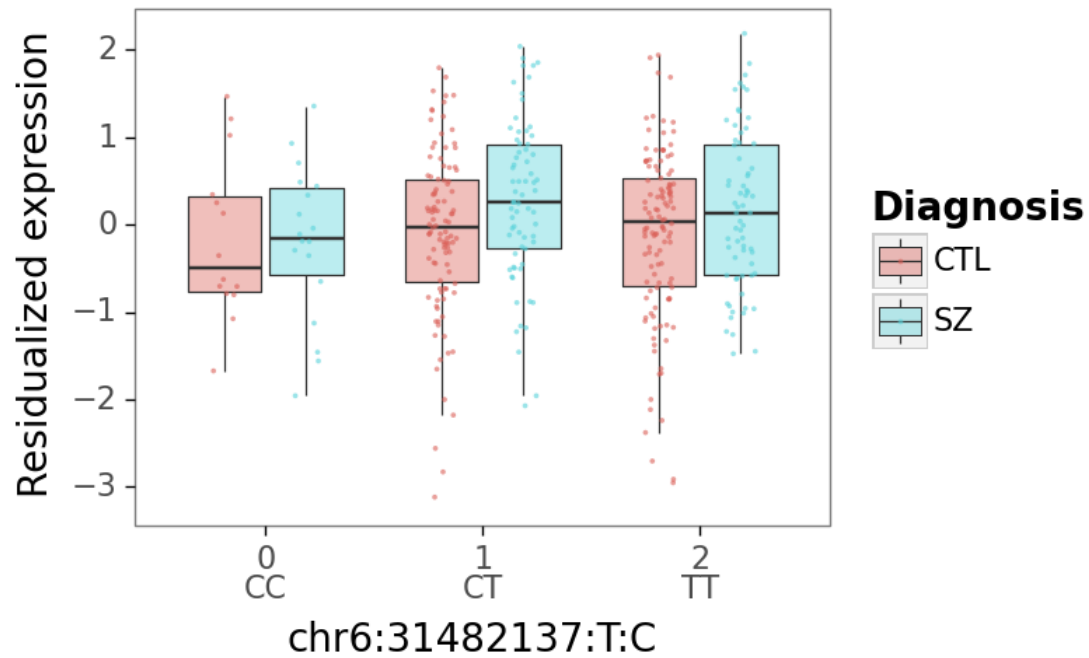
<ggplot: (8779673699669)>

PHF1  
 ENSG00000112511.17  
 SZ GWAS pvalue: 4.1e-08  
 SZ risk allele: A  
 eQTL nominal p-value: 1.5e-04  
 DE adj.P.Val: 0.023



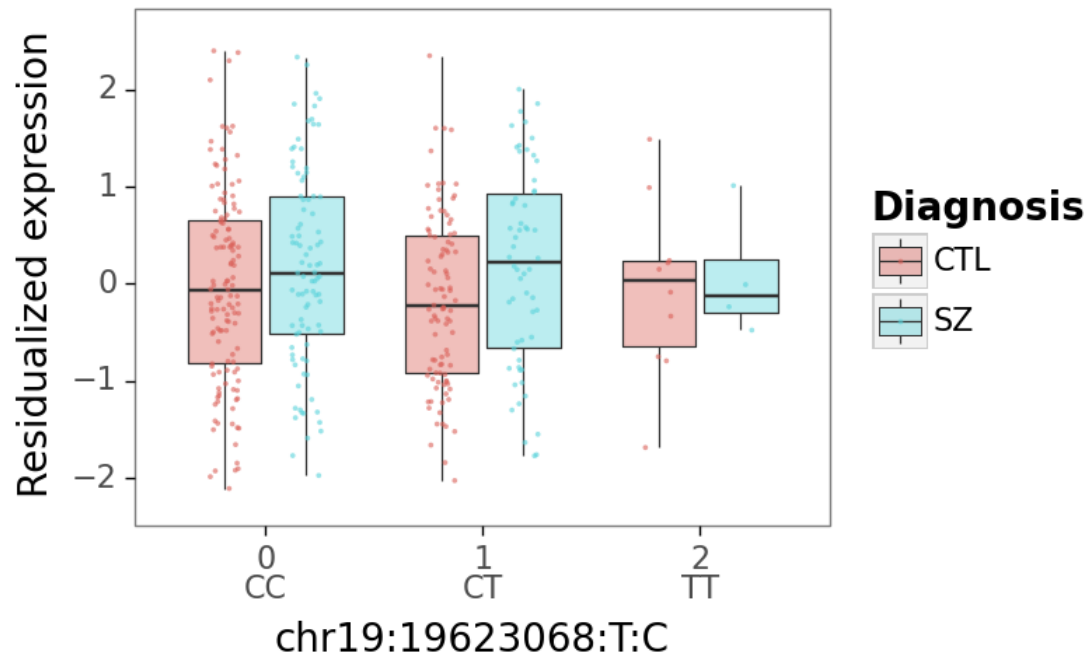
<ggplot: (8779673791501)>

C4A  
 ENSG00000244731.7  
 SZ GWAS pvalue: 4.1e-08  
 SZ risk allele: T  
 eQTL nominal p-value: 1.3e-04  
 DE adj.P.Val: 0.008



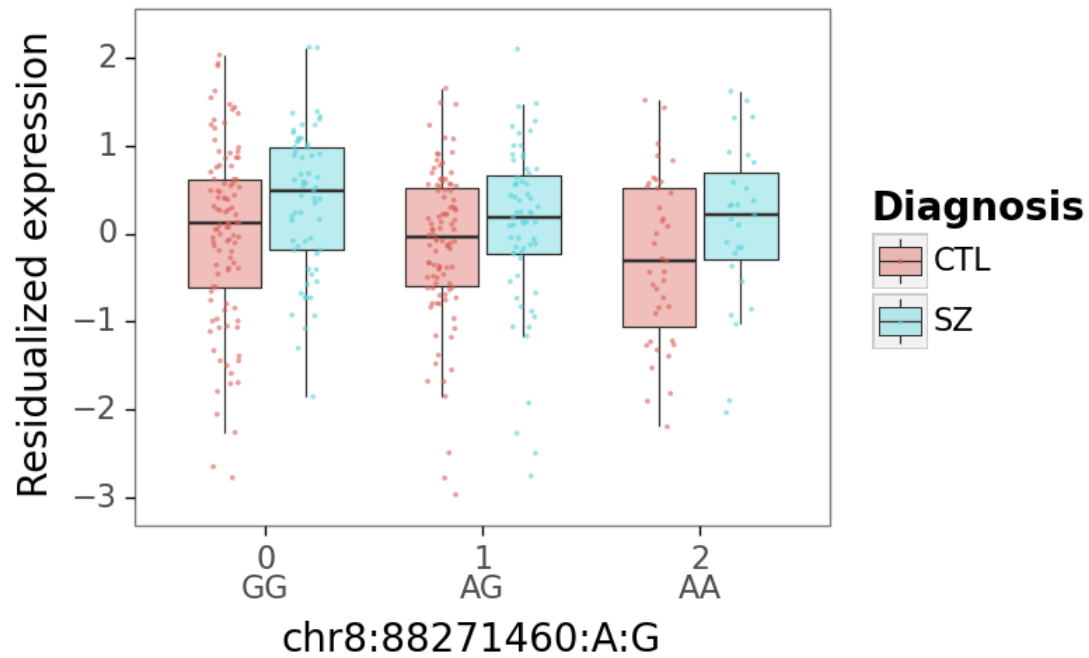
<ggplot: (8779674284858)>

ZNF14  
ENSG00000105708.8  
SZ GWAS pvalue: 4.3e-08  
SZ risk allele: T  
eQTL nominal p-value: 3.4e-06  
DE adj.P.Val: 0.017



<ggplot: (8779616381639)>

AC090568.2  
ENSG00000253553.5  
SZ GWAS pvalue: 4.4e-08  
SZ risk allele: A  
eQTL nominal p-value: 3.0e-04  
DE adj.P.Val: 0.029



<ggplot: (8779673353409)>