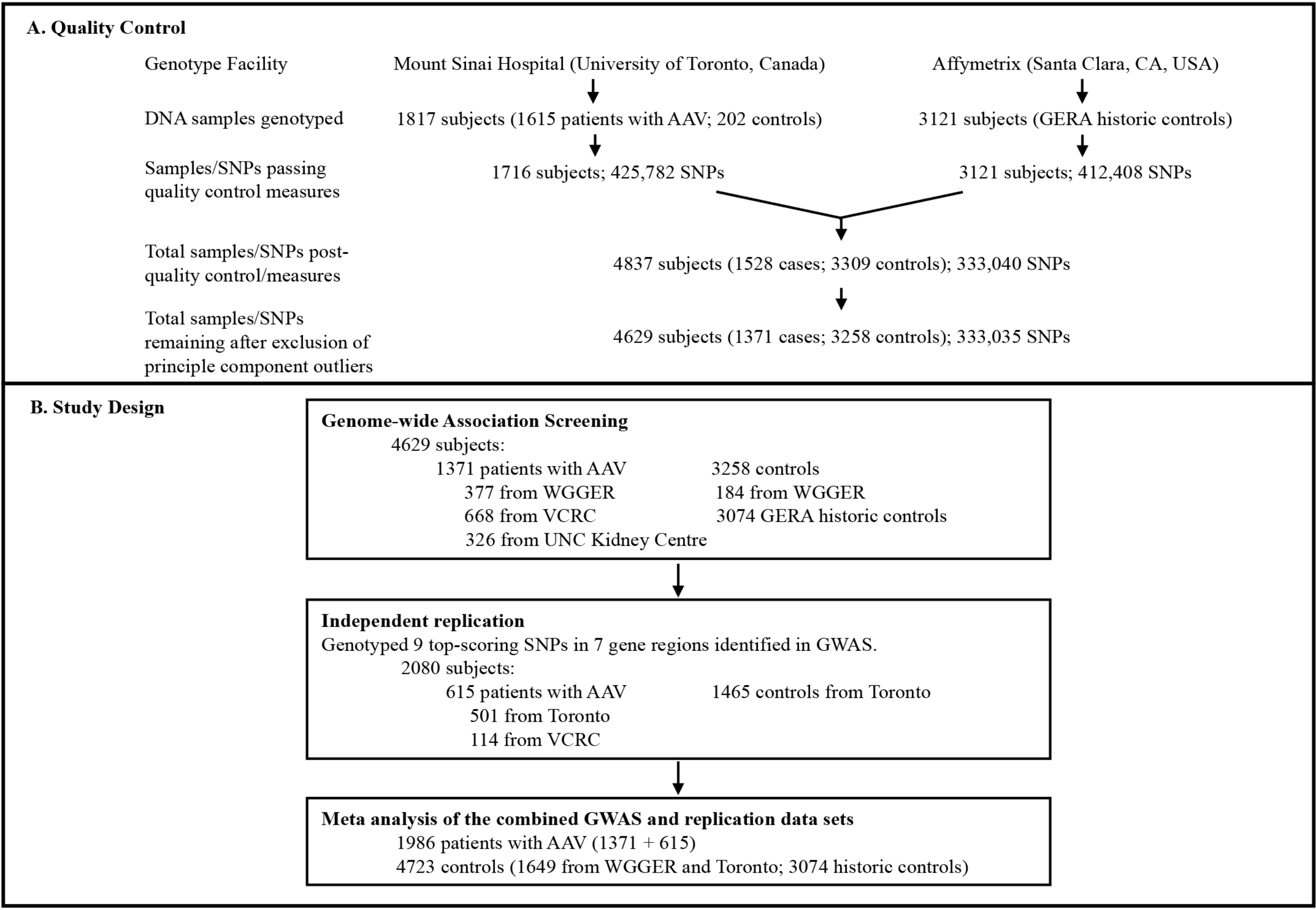


A. Quality Control

Genotype Facility	Mount Sinai Hospital (University of Toronto, Canada)	Affymetrix (Santa Clara, CA, USA)
DNA samples genotyped	1817 subjects (1615 patients with AAV; 202 controls)	3121 subjects (GERA historic controls)
Samples/SNPs passing quality control measures	1716 subjects; 425,782 SNPs	3121 subjects; 412,408 SNPs
Total samples/SNPs post-quality control/measures	4837 subjects (1528 cases; 3309 controls); 333,040 SNPs	
Total samples/SNPs remaining after exclusion of principle component outliers	4629 subjects (1371 cases; 3258 controls); 333,035 SNPs	

B. Study Design

Genome-wide Association Screening

4629 subjects:

1371 patients with AAV	3258 controls
377 from WGGER	184 from WGGER
668 from VCRC	3074 GERA historic controls
326 from UNC Kidney Centre	

Independent replication

Genotyped 9 top-scoring SNPs in 7 gene regions identified in GWAS.

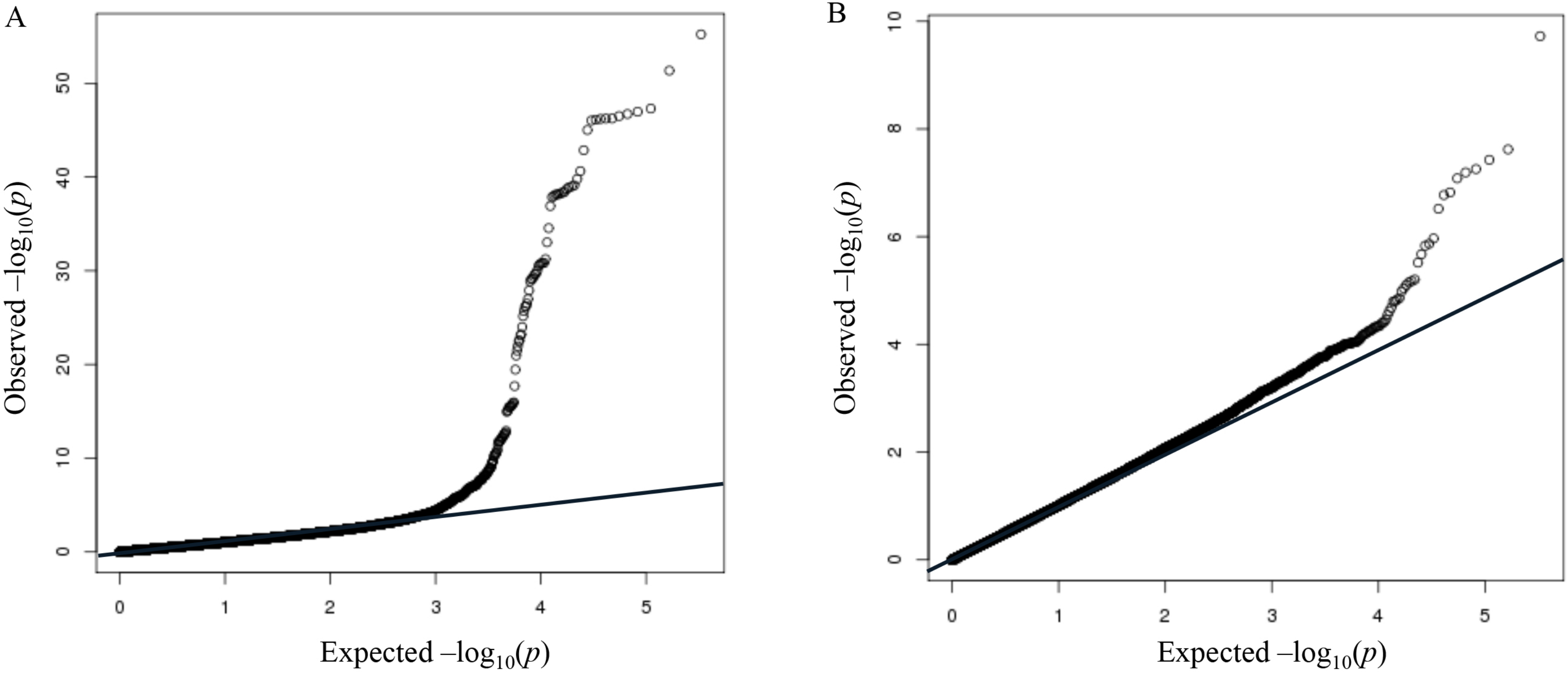
2080 subjects:

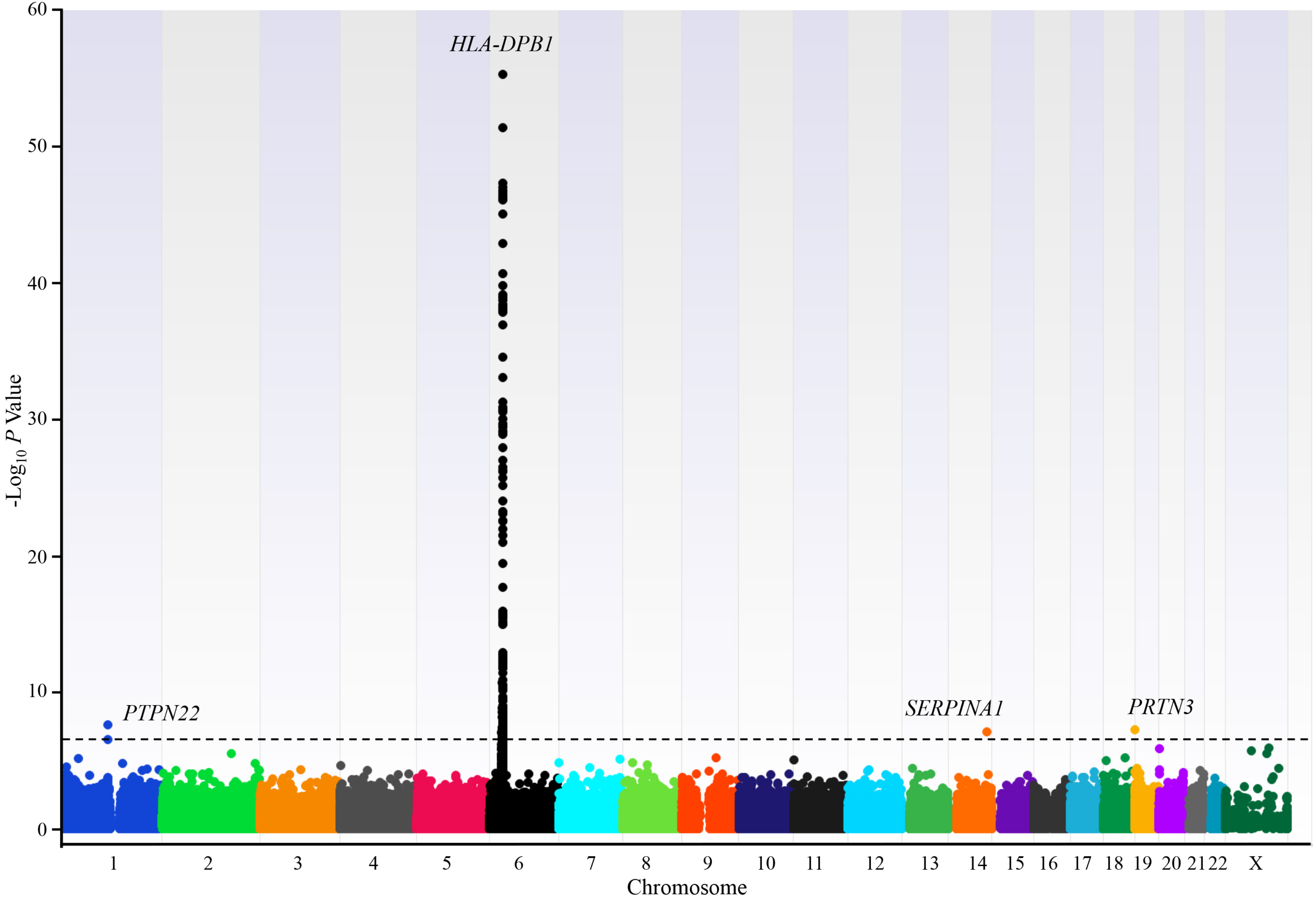
615 patients with AAV	1465 controls from Toronto
501 from Toronto	
114 from VCRC	

Meta analysis of the combined GWAS and replication data sets

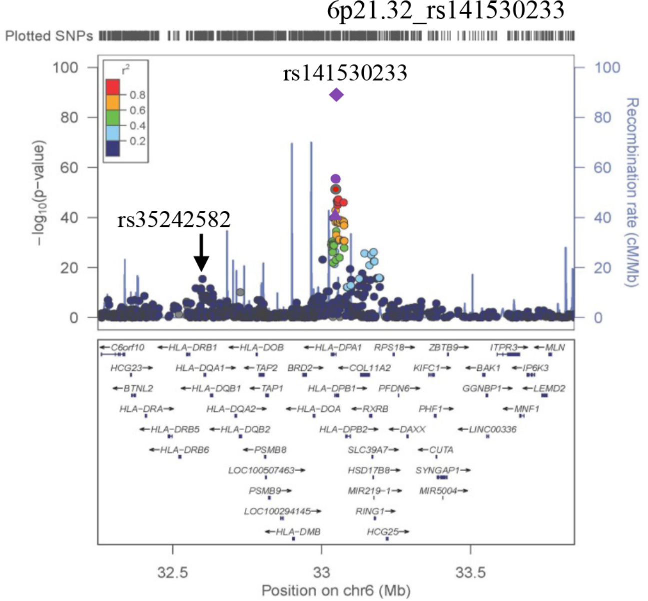
1986 patients with AAV (1371 + 615)

4723 controls (1649 from WGGER and Toronto; 3074 historic controls)

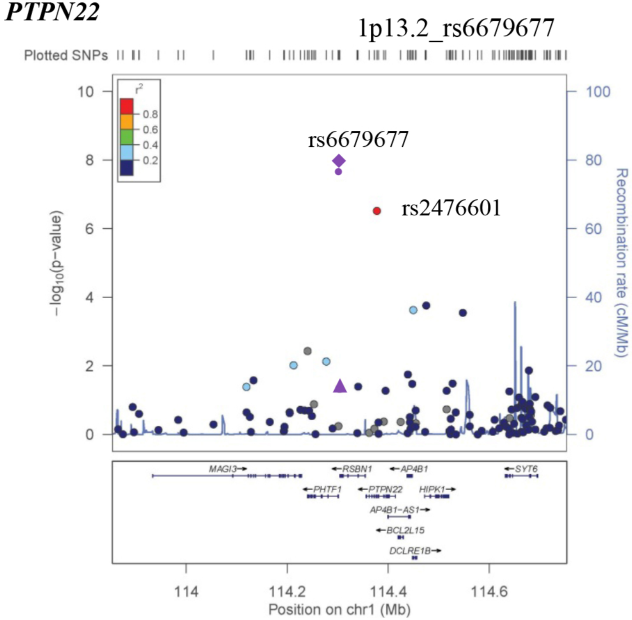

A
Observed –log10(p)
Expected –log10(p)
0
10
20
30
40
50
0
1
2
3
4
5
B
Observed –log10(p)
Expected –log10(p)
0
2
4
6
8
10
0
1
2
3
4
5


HLA-DPB1
PTPN22
SERPINA1
PRTN3
60
50
40
30
20
10
0
$-\mathrm{Log}_{10}$ P Value
1
2
3
4
5
6
7
8
9
10
11
12
13
14
15
16
17
18
19
20
21
22
X
Chromosome

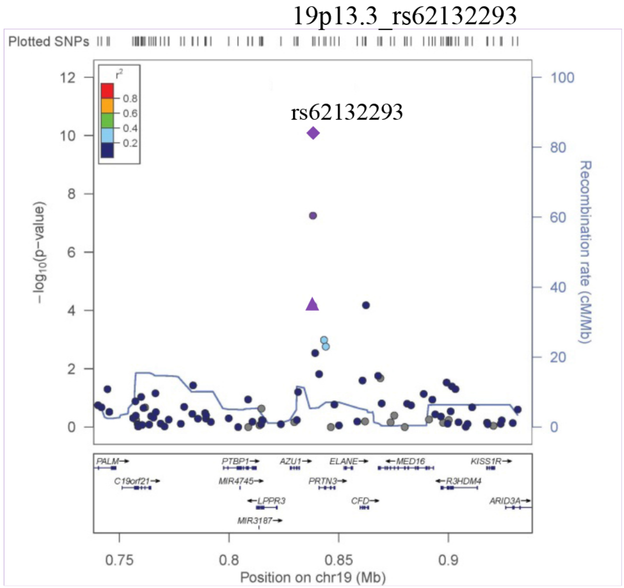
***HLA-DP* and *DQ* regions**



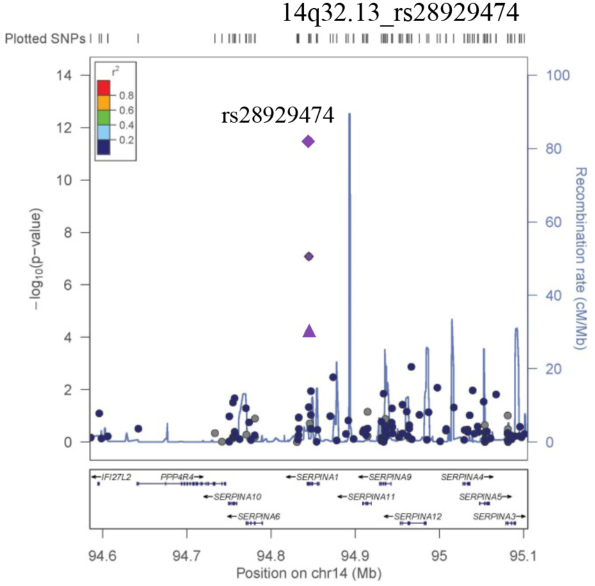
PTPN22

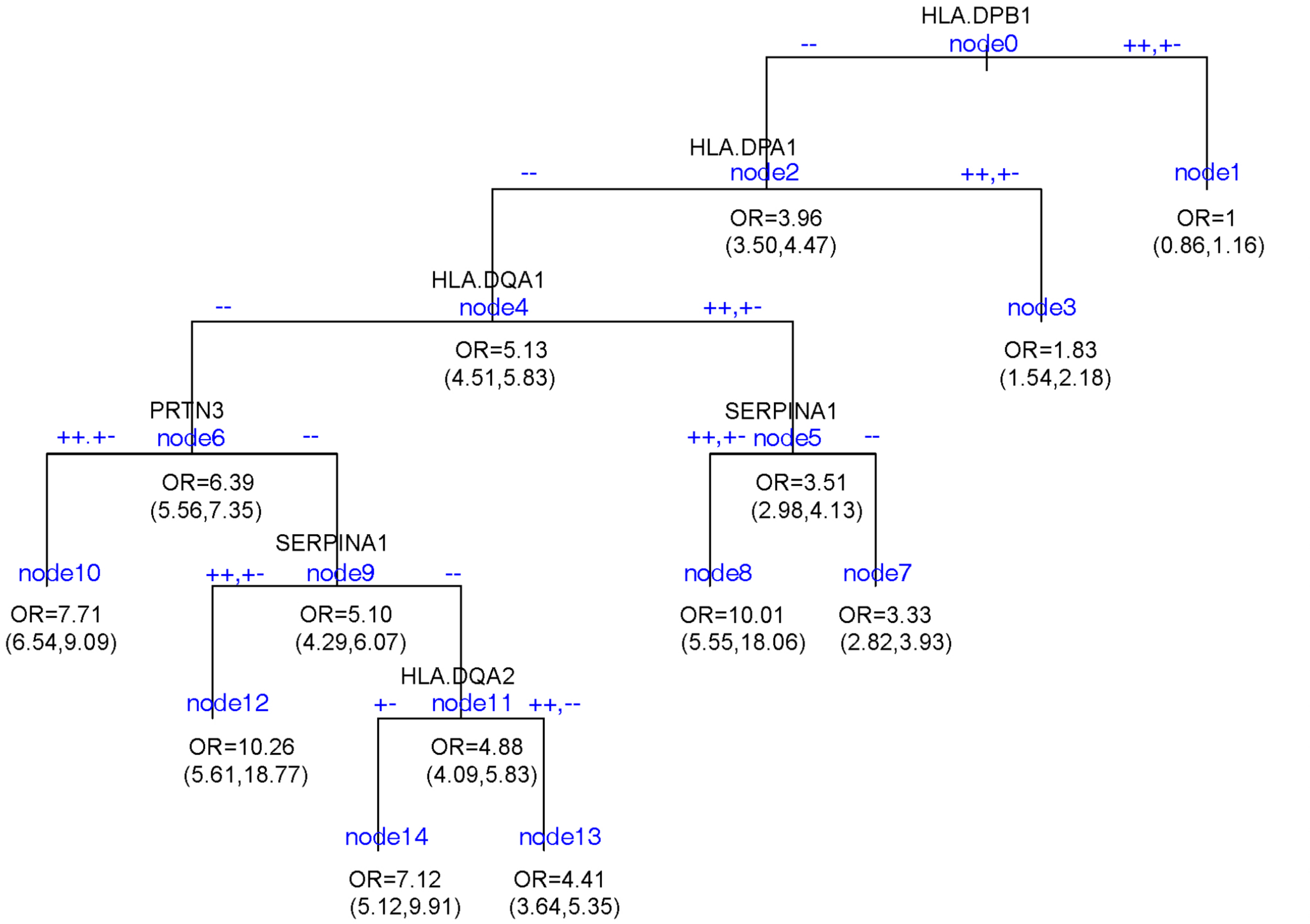


PRTN3

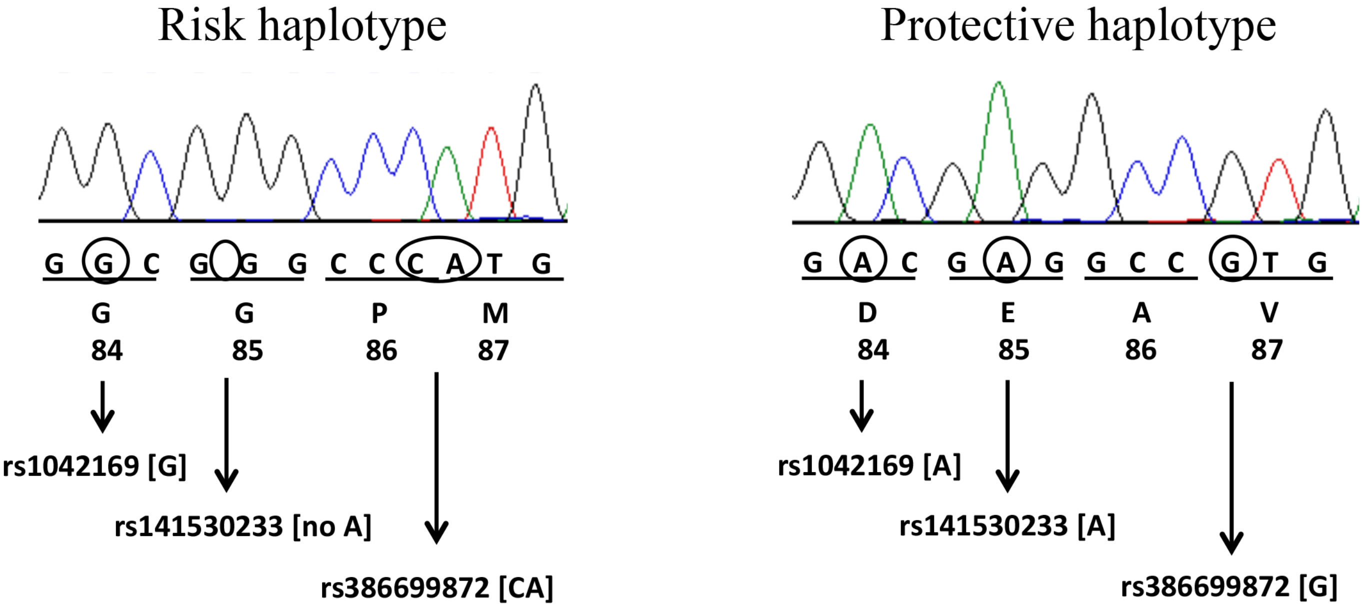


SERPINA1





Node	N. Cases	N. Controls
0	Parent Node	
1	416	2418
2	1570	2305
3	257	818
4	1313	1487
5	392	649
6	921	838
7	361	631
8	31	18
9	372	424
10	549	414
11	342	407
12	30	17
13	255	336
14	87	71



Risk haplotype

Protective haplotype