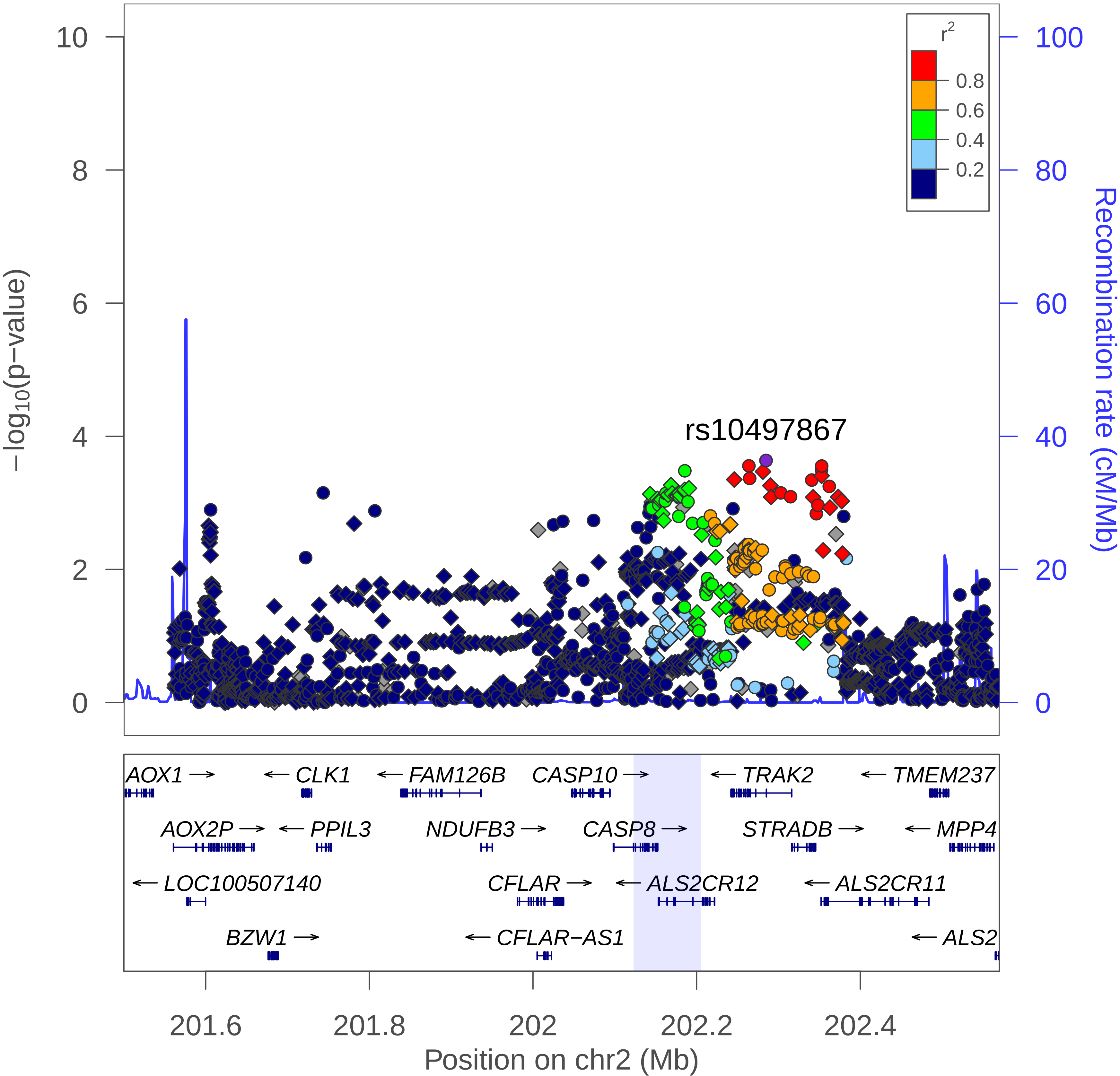
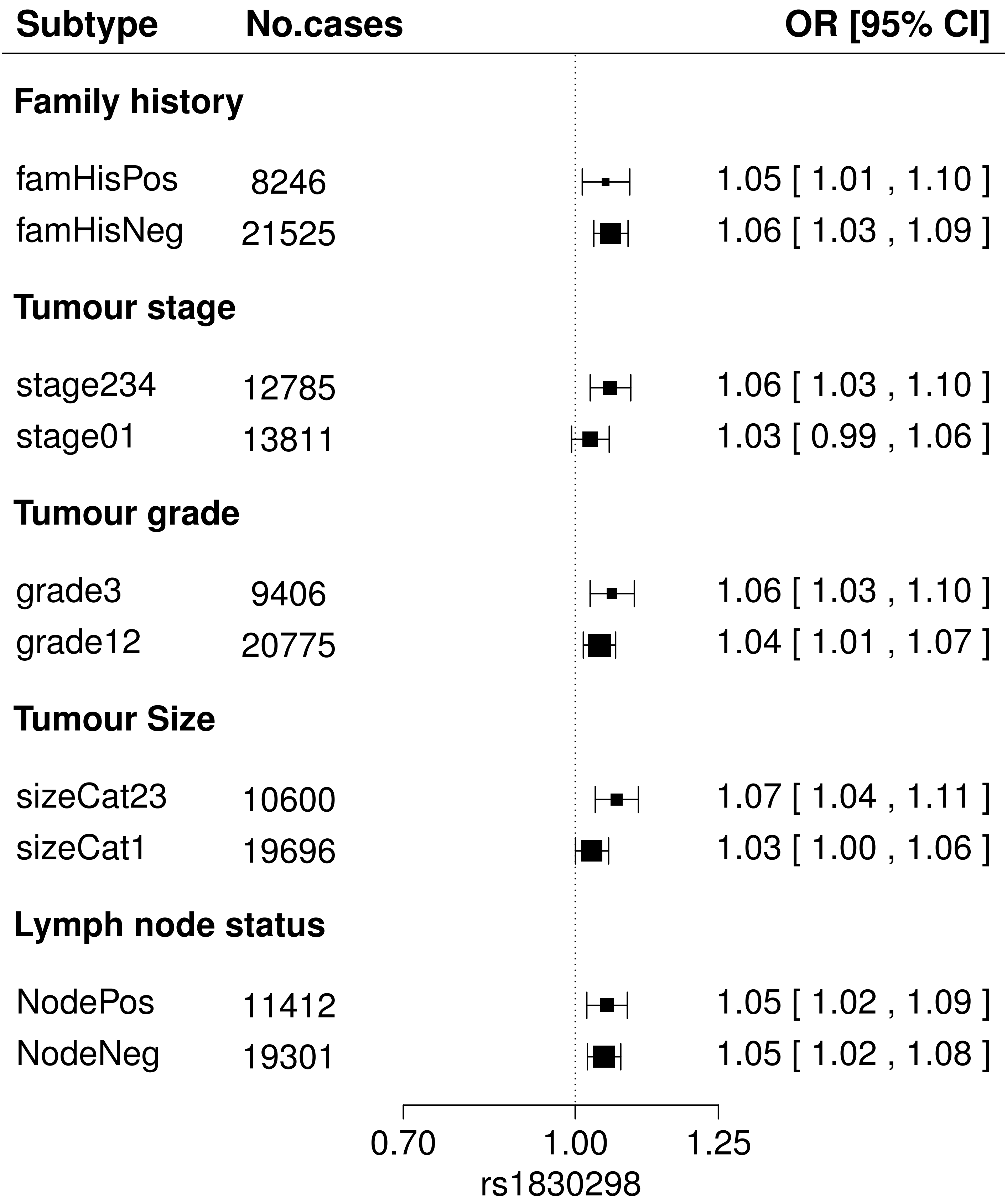
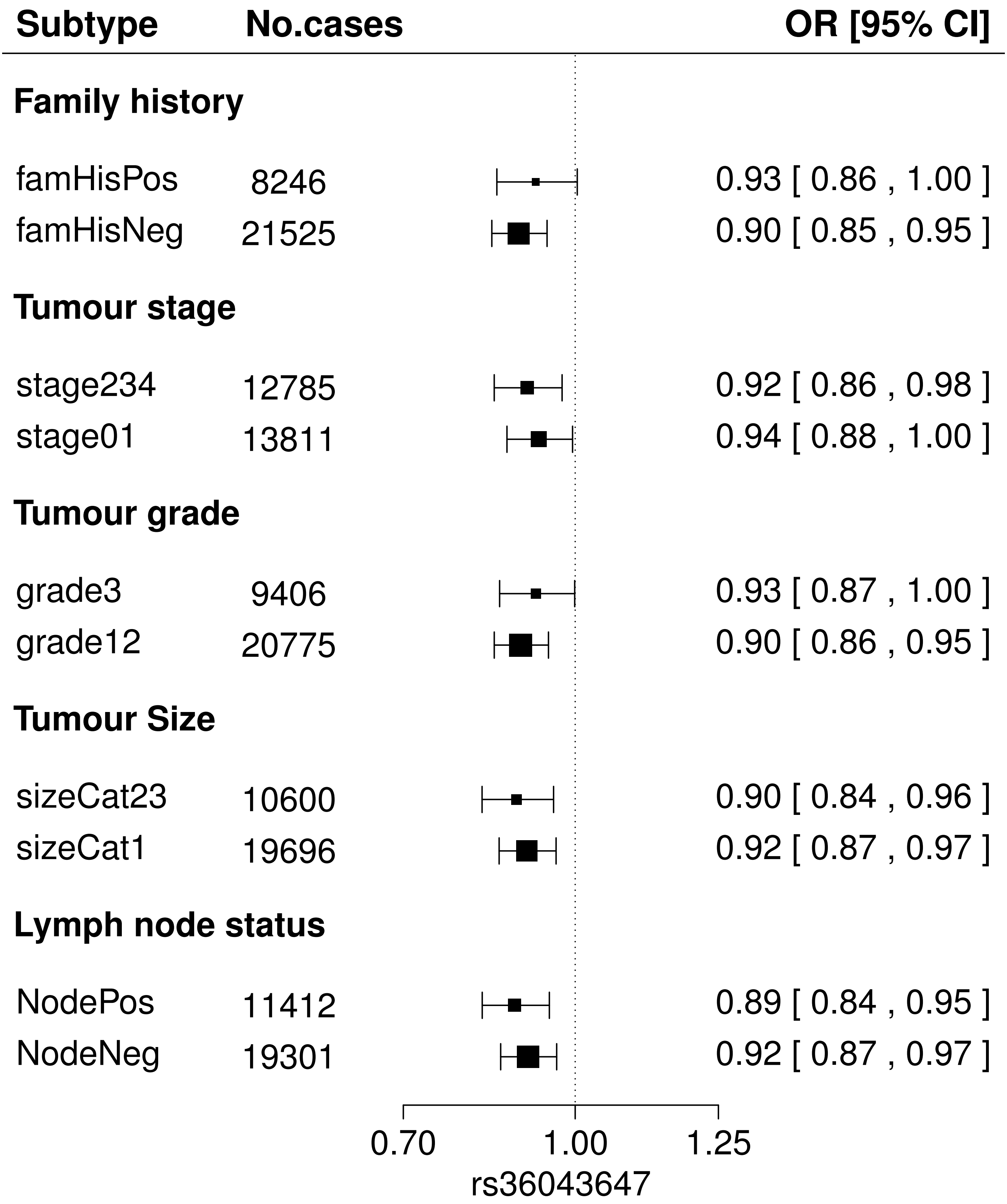
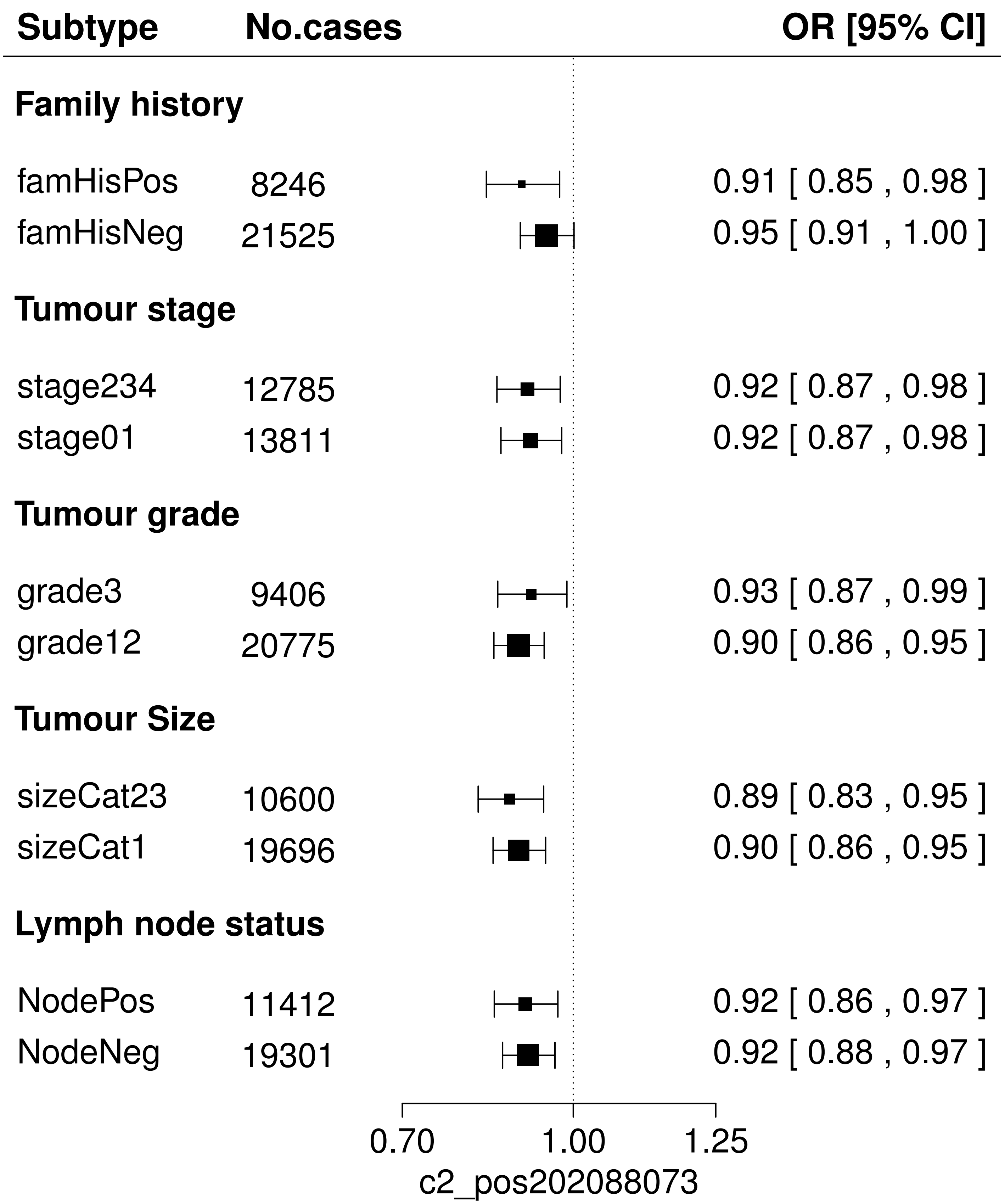
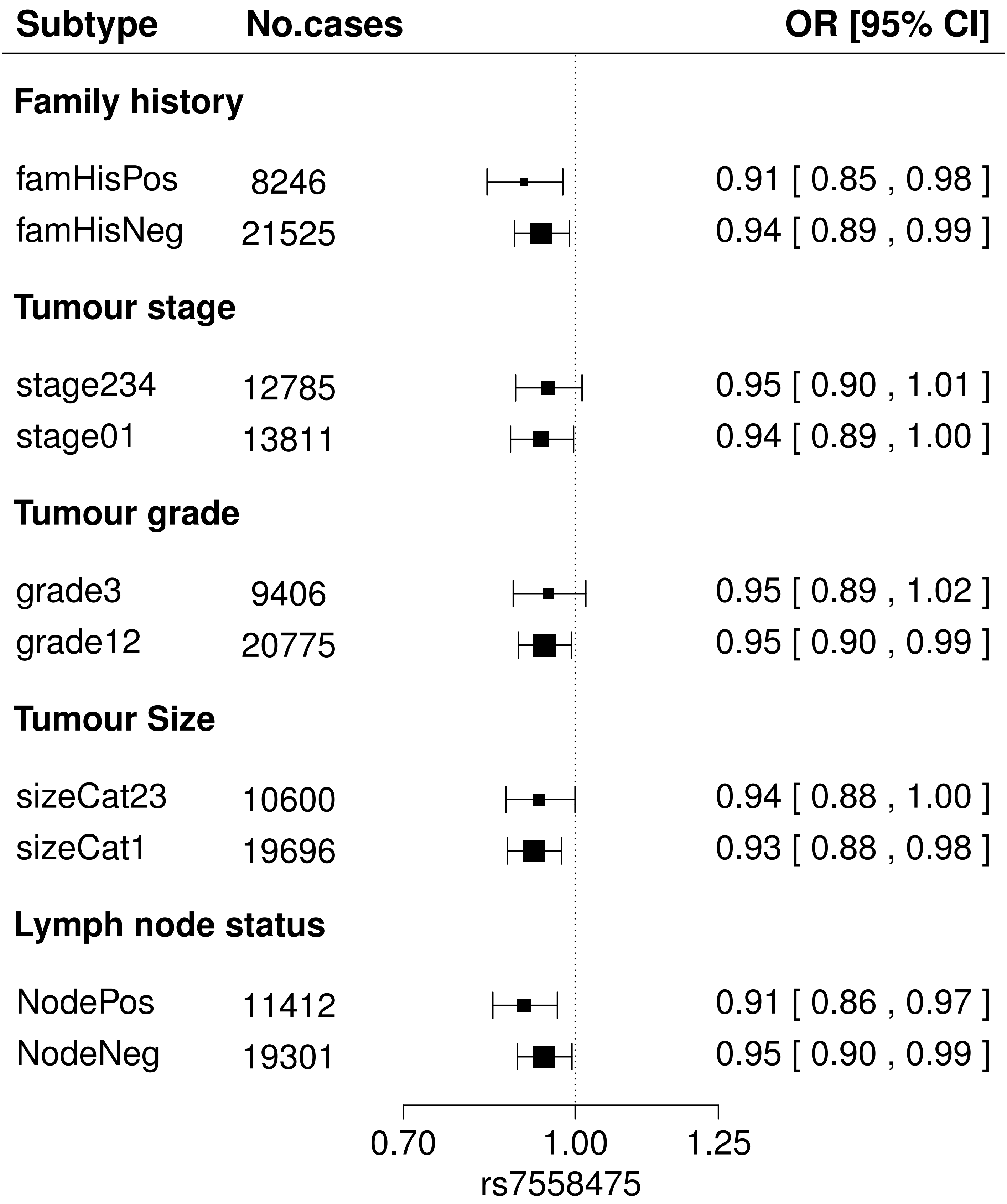


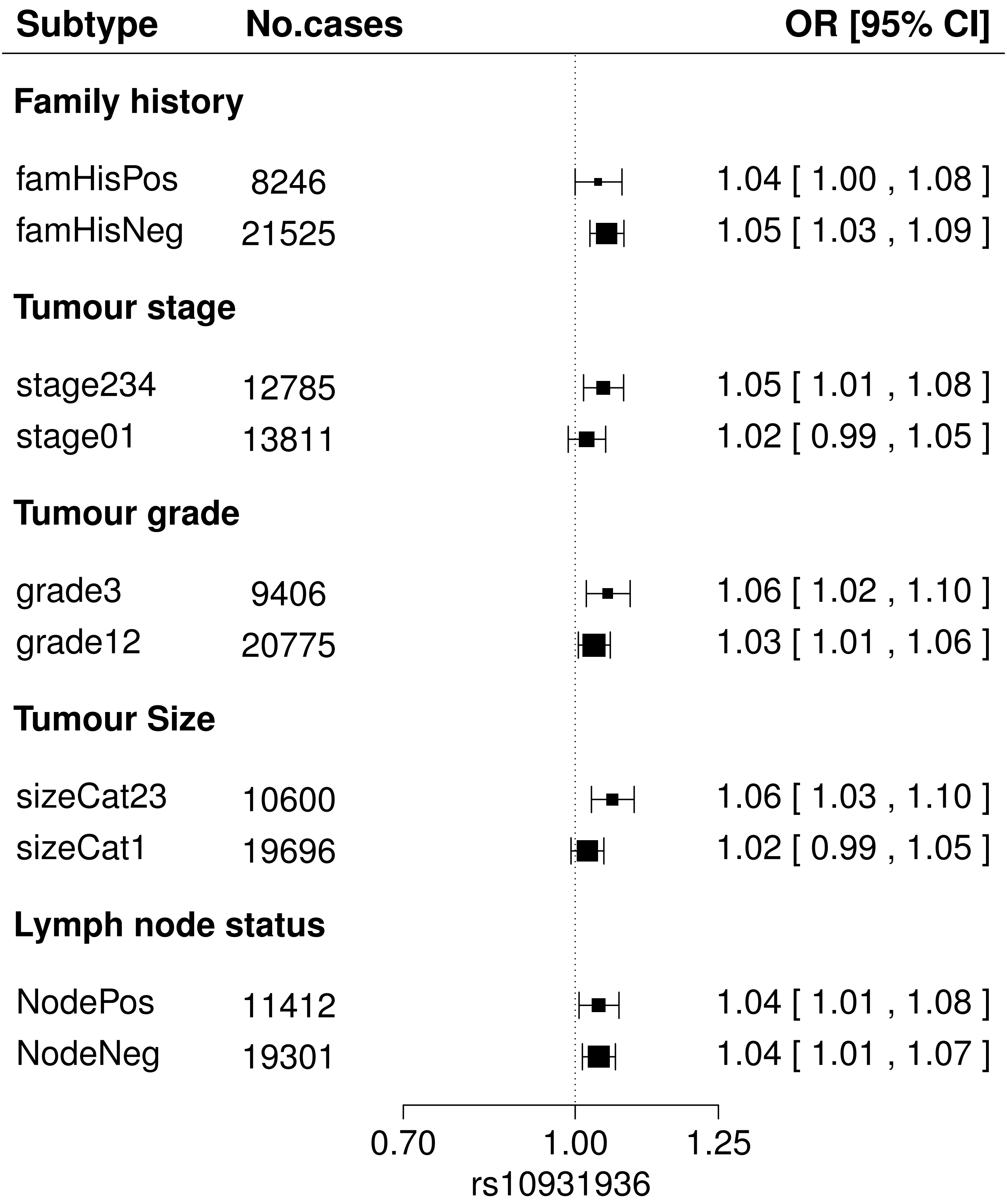
rs10497867
r^2
0.8
0.6
0.4
0.2
10
8
6
4
2
0
100
80
60
40
20
0
−log$_{10}$(p−value)
Recombination rate (cM/Mb)
AOX1 →
← CLK1
← FAM126B
CASP10 →
← TRAK2
← TMEM237
AOX2P →
← PPIL3
NDUFB3 →
CASP8 →
STRADB →
← MPP4
← LOC100507140
CFLAR →
← ALS2CR12
← ALS2CR11
BZW1 →
← CFLAR−AS1
← ALS2
201.6
201.8
202
202.2
202.4
Position on chr2 (Mb)

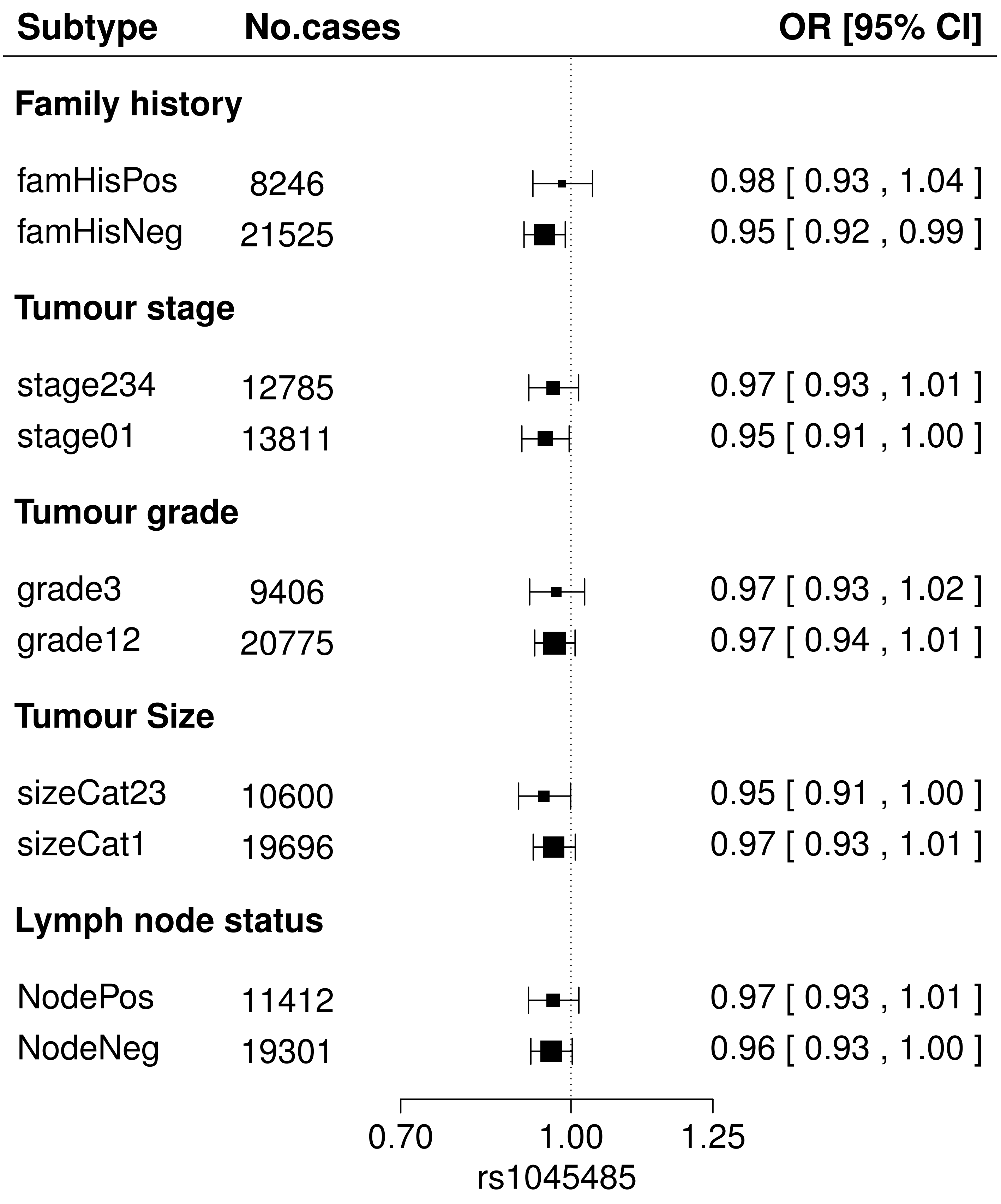

Subtype
No.cases
OR [95% CI]
Family history
famHisPos
8246
1.05 [1.01 , 1.10]
famHisNeg
21525
1.06 [1.03 , 1.09]
Tumour stage
stage234
12785
1.06 [1.03 , 1.10]
stage01
13811
1.03 [0.99 , 1.06]
Tumour grade
grade3
9406
1.06 [1.03 , 1.10]
grade12
20775
1.04 [1.01 , 1.07]
Tumour Size
sizeCat23
10600
1.07 [1.04 , 1.11]
sizeCat1
19696
1.03 [1.00 , 1.06]
Lymph node status
NodePos
11412
1.05 [1.02 , 1.09]
NodeNeg
19301
1.05 [1.02 , 1.08]
0.70
1.00
1.25
rs1830298

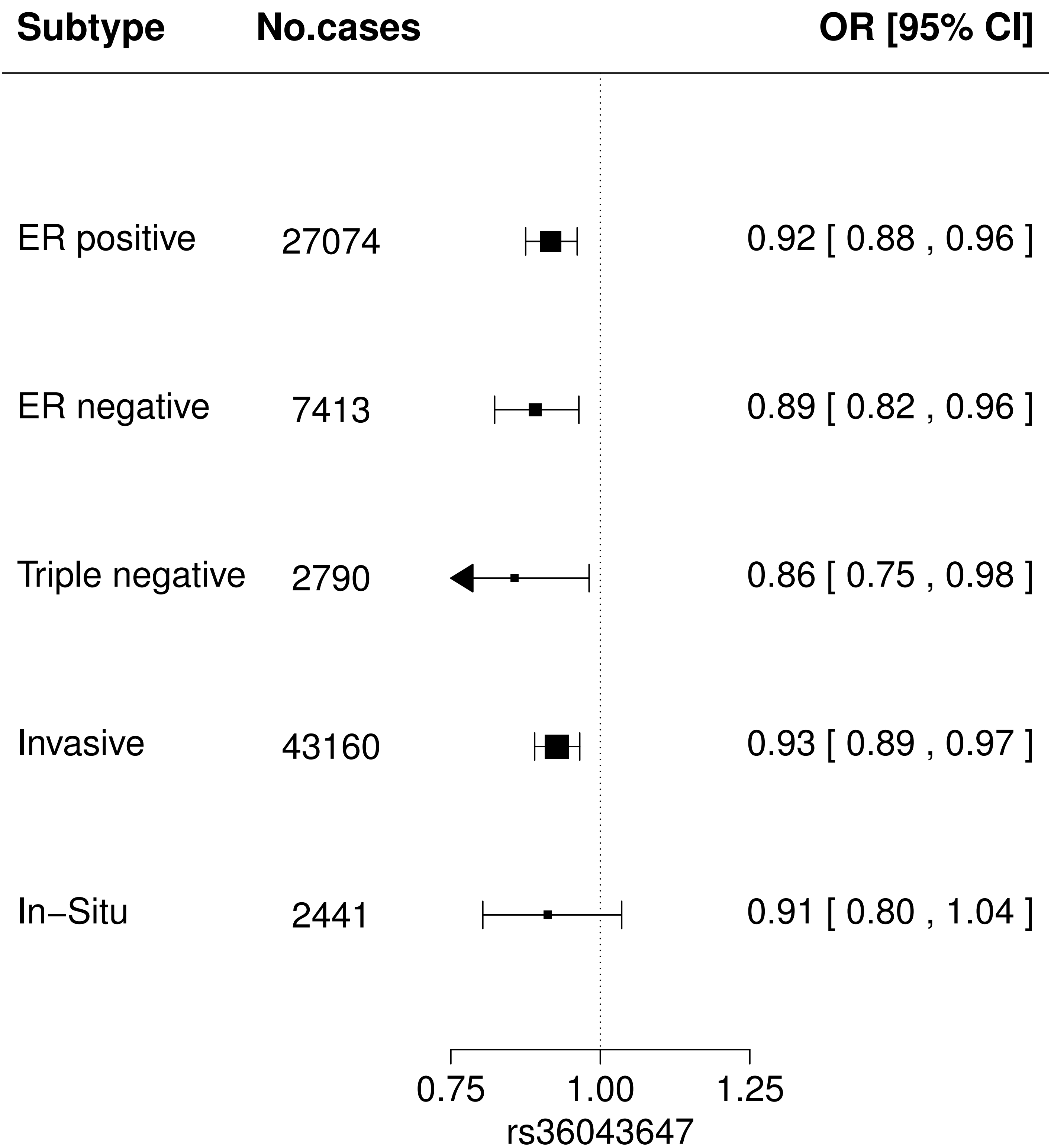

Subtype
No.cases
OR [95% CI]
Family history
famHisPos
8246
0.93 [0.86 , 1.00]
famHisNeg
21525
0.90 [0.85 , 0.95]
Tumour stage
stage234
12785
0.92 [0.86 , 0.98]
stage01
13811
0.94 [0.88 , 1.00]
Tumour grade
grade3
9406
0.93 [0.87 , 1.00]
grade12
20775
0.90 [0.86 , 0.95]
Tumour Size
sizeCat23
10600
0.90 [0.84 , 0.96]
sizeCat1
19696
0.92 [0.87 , 0.97]
Lymph node status
NodePos
11412
0.89 [0.84 , 0.95]
NodeNeg
19301
0.92 [0.87 , 0.97]
0.70
1.00
1.25
rs36043647


Subtype
No.cases
OR [95% CI]
Family history
famHisPos 8246 0.91 [0.85 , 0.98]
famHisNeg 21525 0.95 [0.91 , 1.00]
Tumour stage
stage234 12785 0.92 [0.87 , 0.98]
stage01 13811 0.92 [0.87 , 0.98]
Tumour grade
grade3 9406 0.93 [0.87 , 0.99]
grade12 20775 0.90 [0.86 , 0.95]
Tumour Size
sizeCat23 10600 0.89 [0.83 , 0.95]
sizeCat1 19696 0.90 [0.86 , 0.95]
Lymph node status
NodePos 11412 0.92 [0.86 , 0.97]
NodeNeg 19301 0.92 [0.88 , 0.97]
0.70
1.00
1.25
c2_pos202088073


Subtype
No.cases
OR [95% CI]
Family history
famHisPos
8246
0.91 [0.85 , 0.98]
famHisNeg
21525
0.94 [0.89 , 0.99]
Tumour stage
stage234
12785
0.95 [0.90 , 1.01]
stage01
13811
0.94 [0.89 , 1.00]
Tumour grade
grade3
9406
0.95 [0.89 , 1.02]
grade12
20775
0.95 [0.90 , 0.99]
Tumour Size
sizeCat23
10600
0.94 [0.88 , 1.00]
sizeCat1
19696
0.93 [0.88 , 0.98]
Lymph node status
NodePos
11412
0.91 [0.86 , 0.97]
NodeNeg
19301
0.95 [0.90 , 0.99]
0.70
1.00
1.25
rs7558475

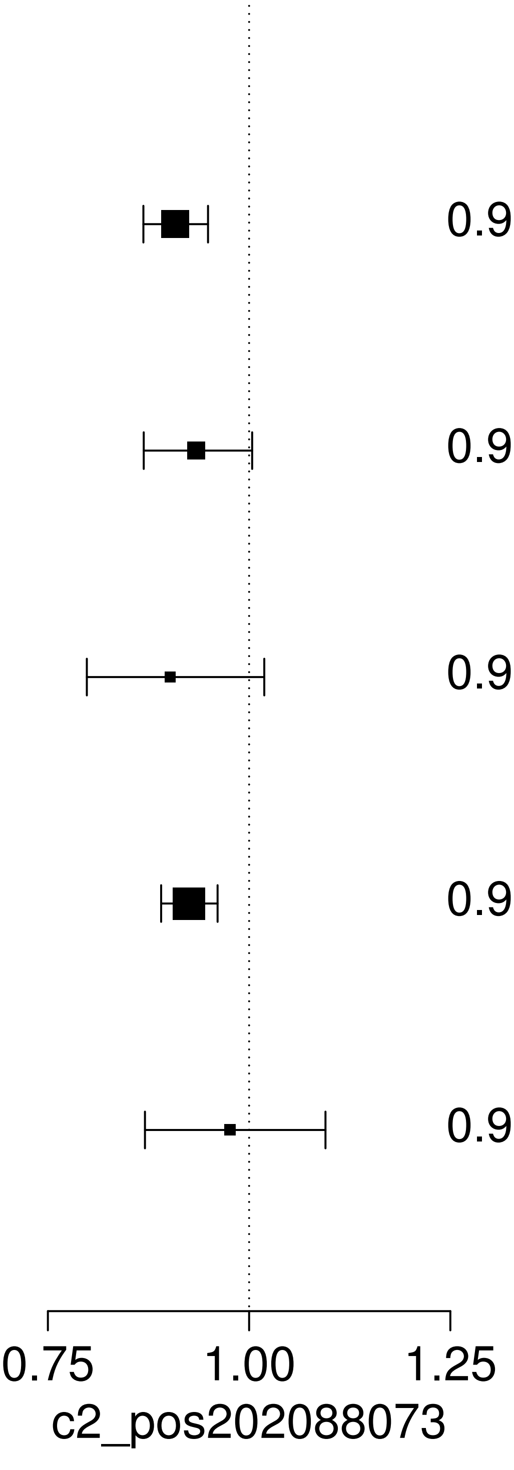

Subtype
No.cases
OR [95% CI]
Family history
famHisPos
8246
1.04 [1.00 , 1.08]
famHisNeg
21525
1.05 [1.03 , 1.09]
Tumour stage
stage234
12785
1.05 [1.01 , 1.08]
stage01
13811
1.02 [0.99 , 1.05]
Tumour grade
grade3
9406
1.06 [1.02 , 1.10]
grade12
20775
1.03 [1.01 , 1.06]
Tumour Size
sizeCat23
10600
1.06 [1.03 , 1.10]
sizeCat1
19696
1.02 [0.99 , 1.05]
Lymph node status
NodePos
11412
1.04 [1.01 , 1.08]
NodeNeg
19301
1.04 [1.01 , 1.07]
0.70
1.00
1.25
rs10931936

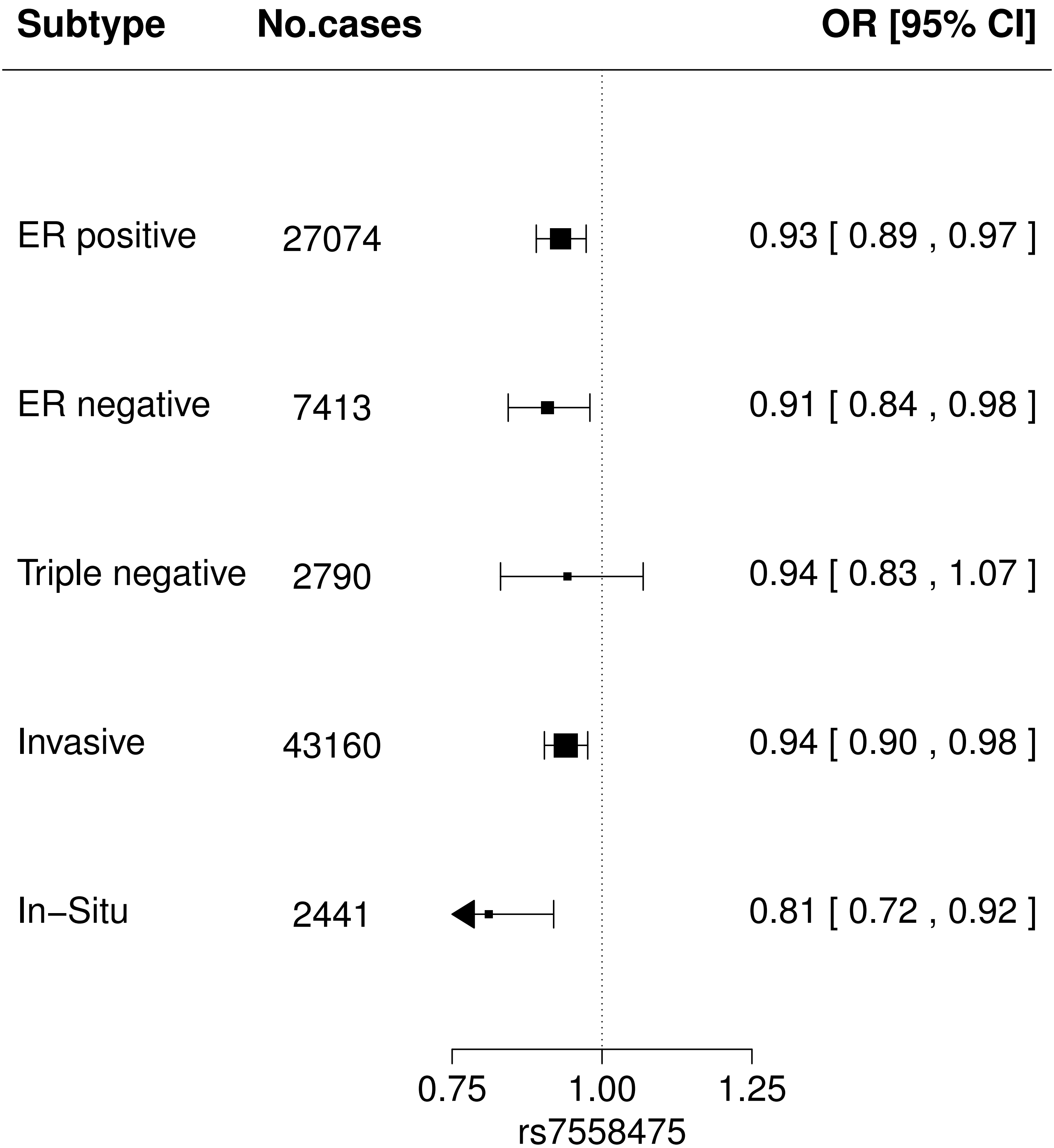

Subtype
No.cases
OR [95% CI]
Family history
famHisPos 8246 0.98 [0.93 , 1.04]
famHisNeg 21525 0.95 [0.92 , 0.99]
Tumour stage
stage234 12785 0.97 [0.93 , 1.01]
stage01 13811 0.95 [0.91 , 1.00]
Tumour grade
grade3 9406 0.97 [0.93 , 1.02]
grade12 20775 0.97 [0.94 , 1.01]
Tumour Size
sizeCat23 10600 0.95 [0.91 , 1.00]
sizeCat1 19696 0.97 [0.93 , 1.01]
Lymph node status
NodePos 11412 0.97 [0.93 , 1.01]
NodeNeg 19301 0.96 [0.93 , 1.00]
0.70
1.00
1.25
rs1045485

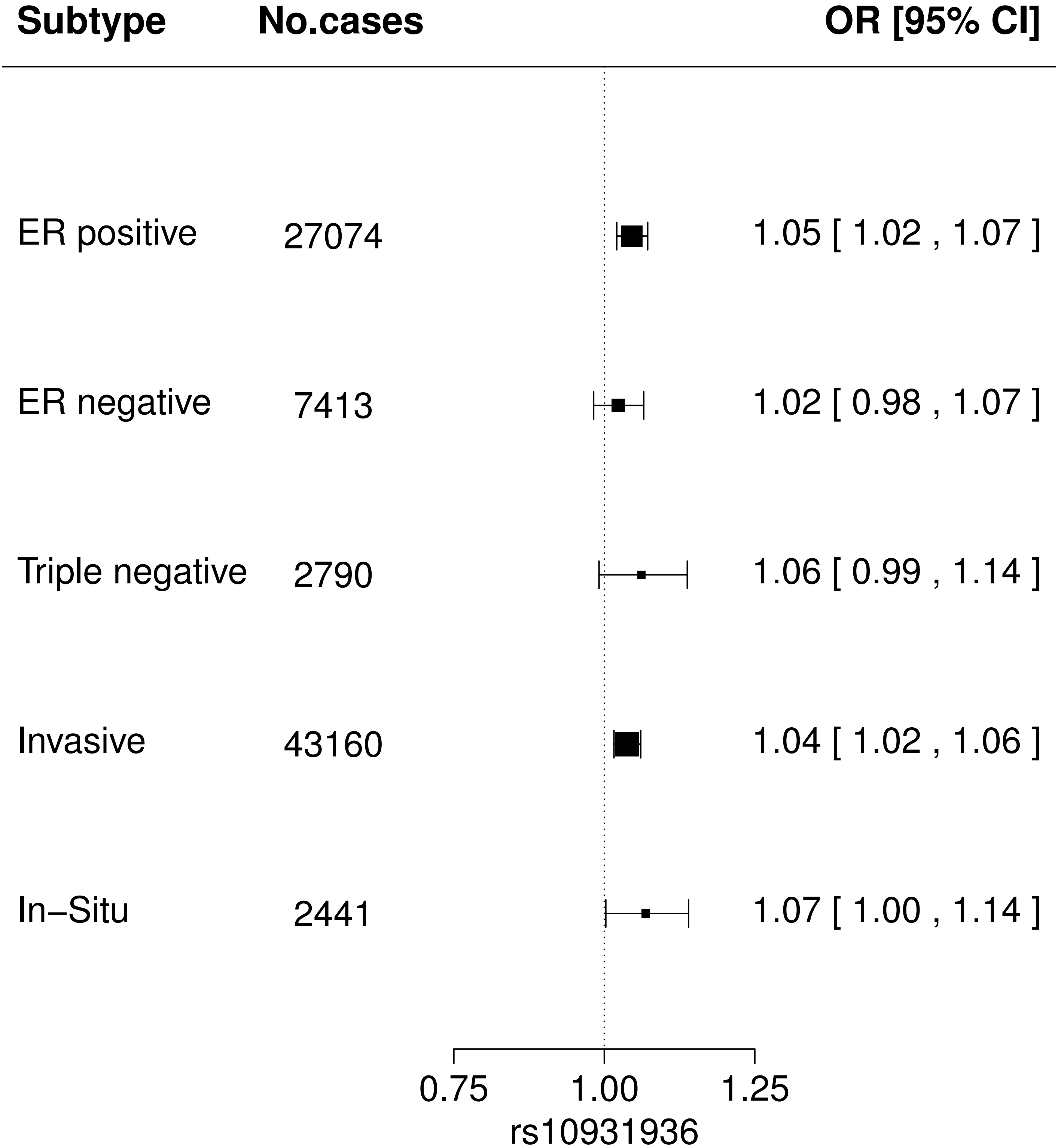


Subtype	No.cases	OR [95% CI]
ER positive	27074	0.92 [0.88 , 0.96]
ER negative	7413	0.89 [0.82 , 0.96]
Triple negative	2790	0.86 [0.75 , 0.98]
Invasive	43160	0.93 [0.89 , 0.97]
In−Situ	2441	0.91 [0.80 , 1.04]

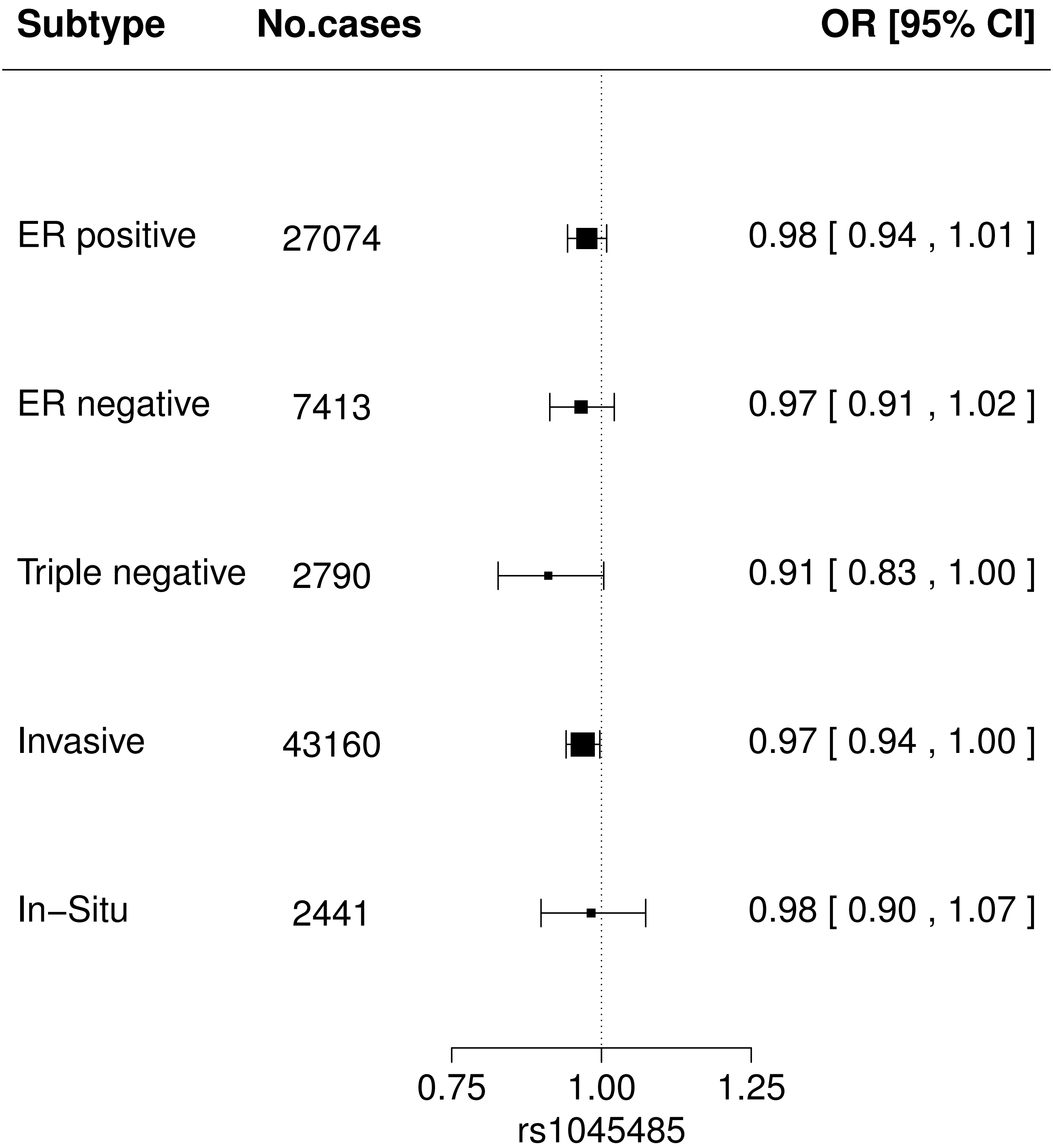
Subtype	No.cases	OR [95% CI]
ER positive	27074	0.91 [0.87 , 0.95]
ER negative	7413	0.93 [0.87 , 1.00]
Triple negative	2790	0.90 [0.80 , 1.02]
Invasive	43160	0.93 [0.89 , 0.96]
In−Situ	2441	0.98 [0.87 , 1.09]




Subtype
No.cases
OR [95% CI]
ER positive
27074
0.93 [0.89 , 0.97]
ER negative
7413
0.91 [0.84 , 0.98]
Triple negative
2790
0.94 [0.83 , 1.07]
Invasive
43160
0.94 [0.90 , 0.98]
In−Situ
2441
0.81 [0.72 , 0.92]
0.75
1.00
1.25
rs7558475



Subtype
No.cases
OR [95% CI]
ER positive
27074
1.05 [1.02 , 1.07]
ER negative
7413
1.02 [0.98 , 1.07]
Triple negative
2790
1.06 [0.99 , 1.14]
Invasive
43160
1.04 [1.02 , 1.06]
In−Situ
2441
1.07 [1.00 , 1.14]
0.75
1.00
1.25
rs10931936


Subtype
No.cases
OR [95% CI]
ER positive
27074
0.98 [0.94 , 1.01]
ER negative
7413
0.97 [0.91 , 1.02]
Triple negative
2790
0.91 [0.83 , 1.00]
Invasive
43160
0.97 [0.94 , 1.00]
In−Situ
2441
0.98 [0.90 , 1.07]
0.75
1.00
1.25
rs1045485

Supplemental Figure Legends

Figure S1 Associations of 1,732 SNPs conditional on rs1830298. The most significantly associated SNP after conditioning on rs1830298 is indicated. The upper panel plots SNPs based on their chromosomal coordinates on the x axis and their p values on the $-\log_{10}$ scale on the y axis. Circle and diamond symbols represent typed and imputed SNPs respectively. The colours indicate the pairwise r^2 with the best hit (highlighted in purple); r^2 are calculated based on the European panel in the 1000 genomes project. Genes within the region are indicated in the lower panel, with arrows indicating transcript direction, dense blocks for exons and lines for introns. The highlighted region contains the SNPs with p values $\leq 1\text{x}10^{-4}$. The plot was generated using LocusZoom [17].

Figure S2. Associations of family history, tumour stage, tumour grade, tumour size and lymph node status by SNP.

Squares denote the individual study per-allele OR with the size of the symbol indicating sample size. Notation: famHisPos indicates at least one first degree relative with breast cancer, famHisNeg indicates no first degree relatives with breast cancer. Tumours were grouped into stages 2,3,4 or stages 0,1. Tumours were grouped by grade into grade 3 or grades 2 and 1). sizeCat 23 indicates tumour size ≥2cm, sizeCat1 indicates <2cm. NodePos indicates the presence of positive lymph nodes and NodeNeg indicates no positive lymph nodes. Cases in each of the above groups were compared to all controls.

Figure S3. Associations of ER positive, ER negative, triple negative, invasive and non-invasive breast cancer by SNP.

Squares denote the individual study per-allele OR with the size of the symbol indicating sample size. Triple negative indicates the absence of tumour receptors for ER, PR and HER2. Cases in each subtype group were compared to all controls.