Figures

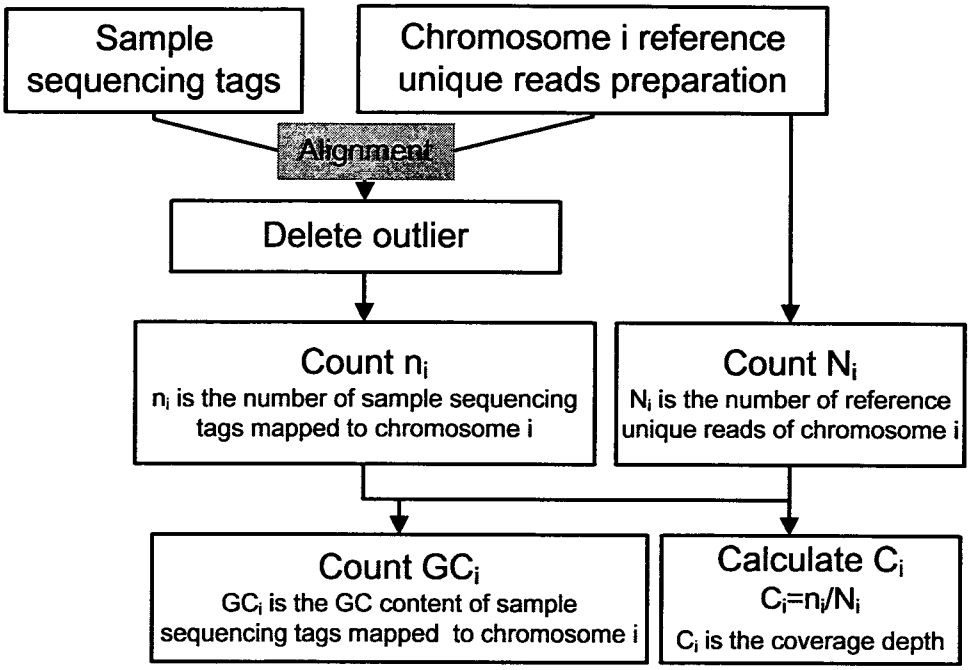

Figure 1

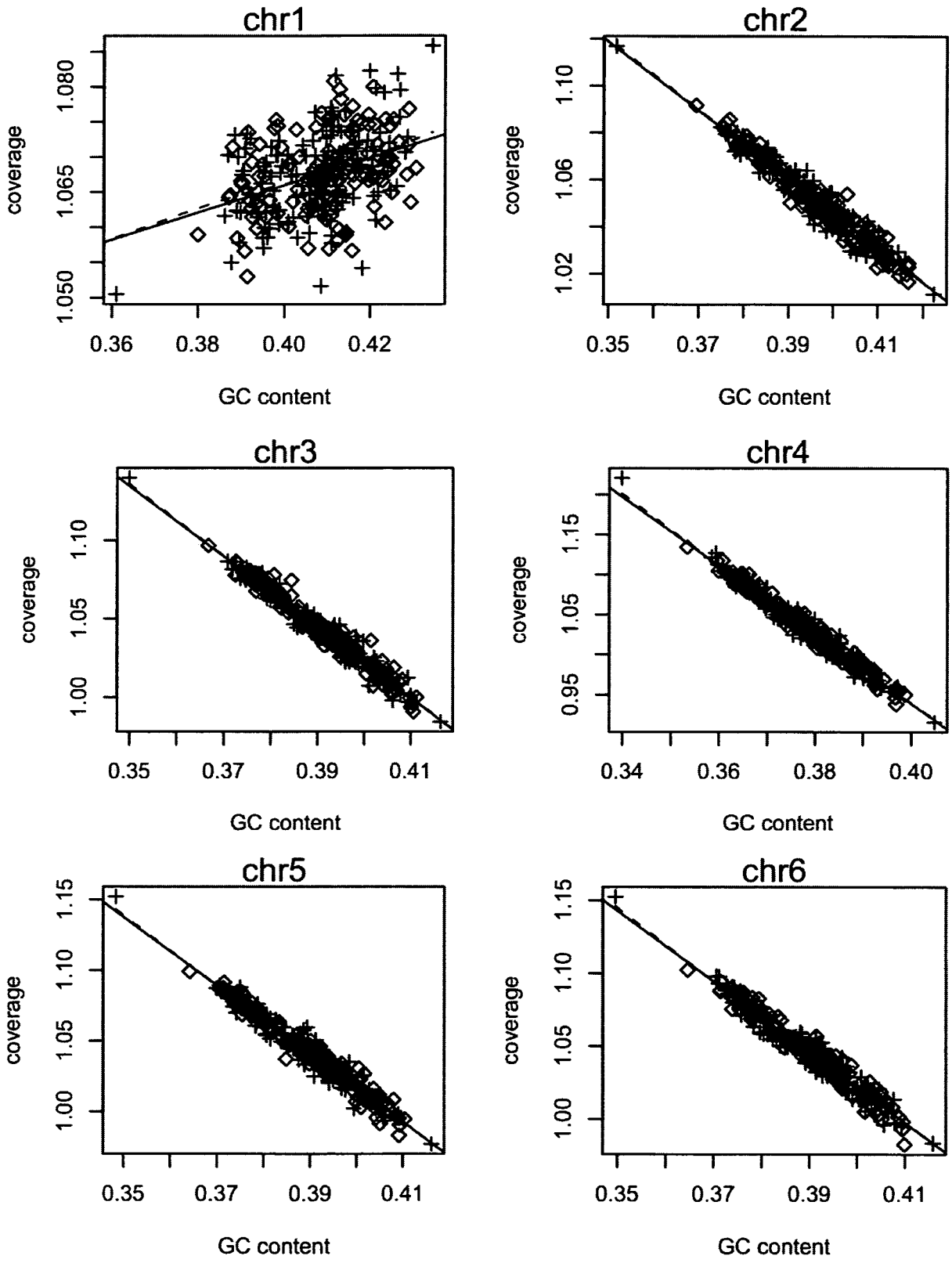

Figure 2
(page 1 of 4)

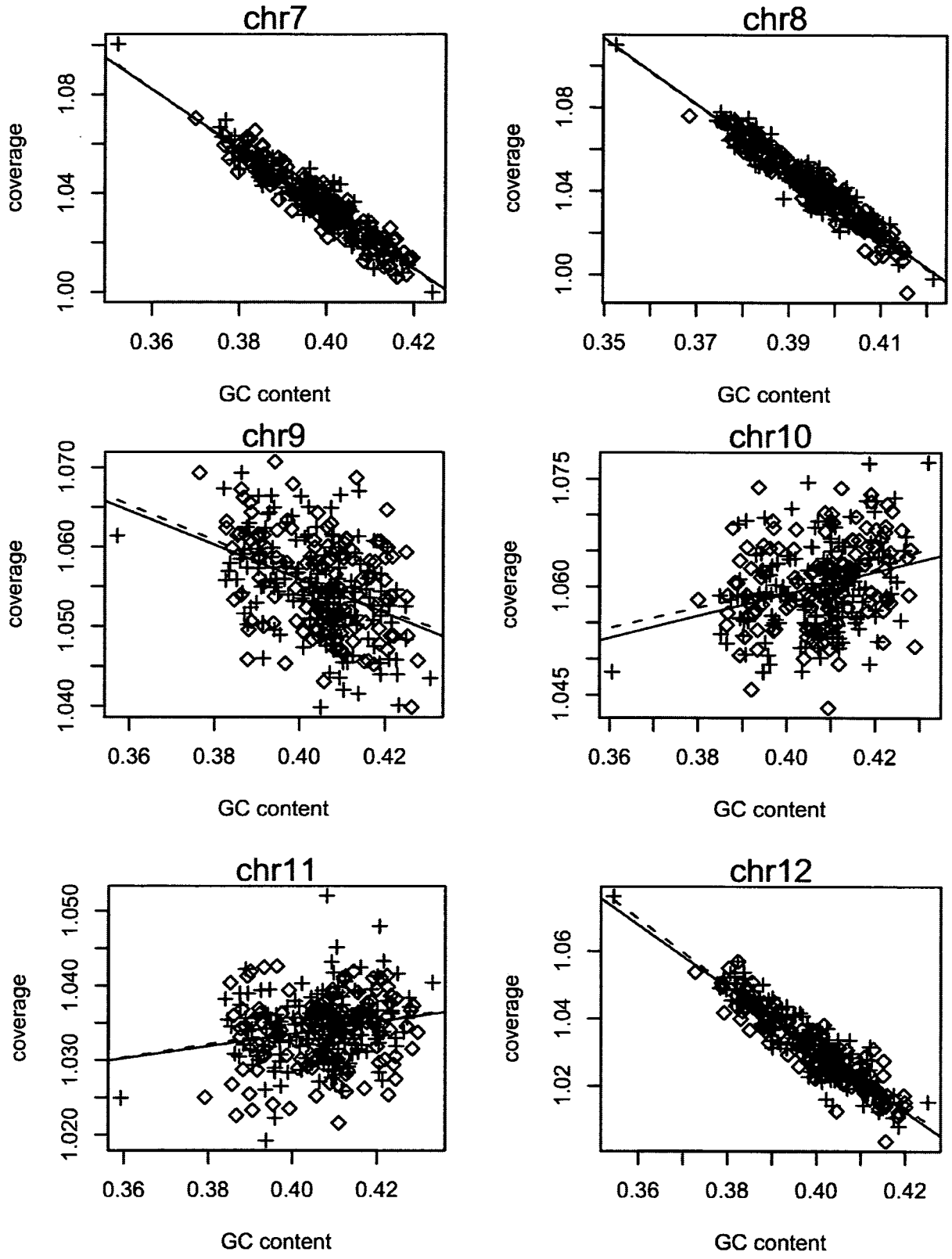

Figure 2
(page 2 of 4)

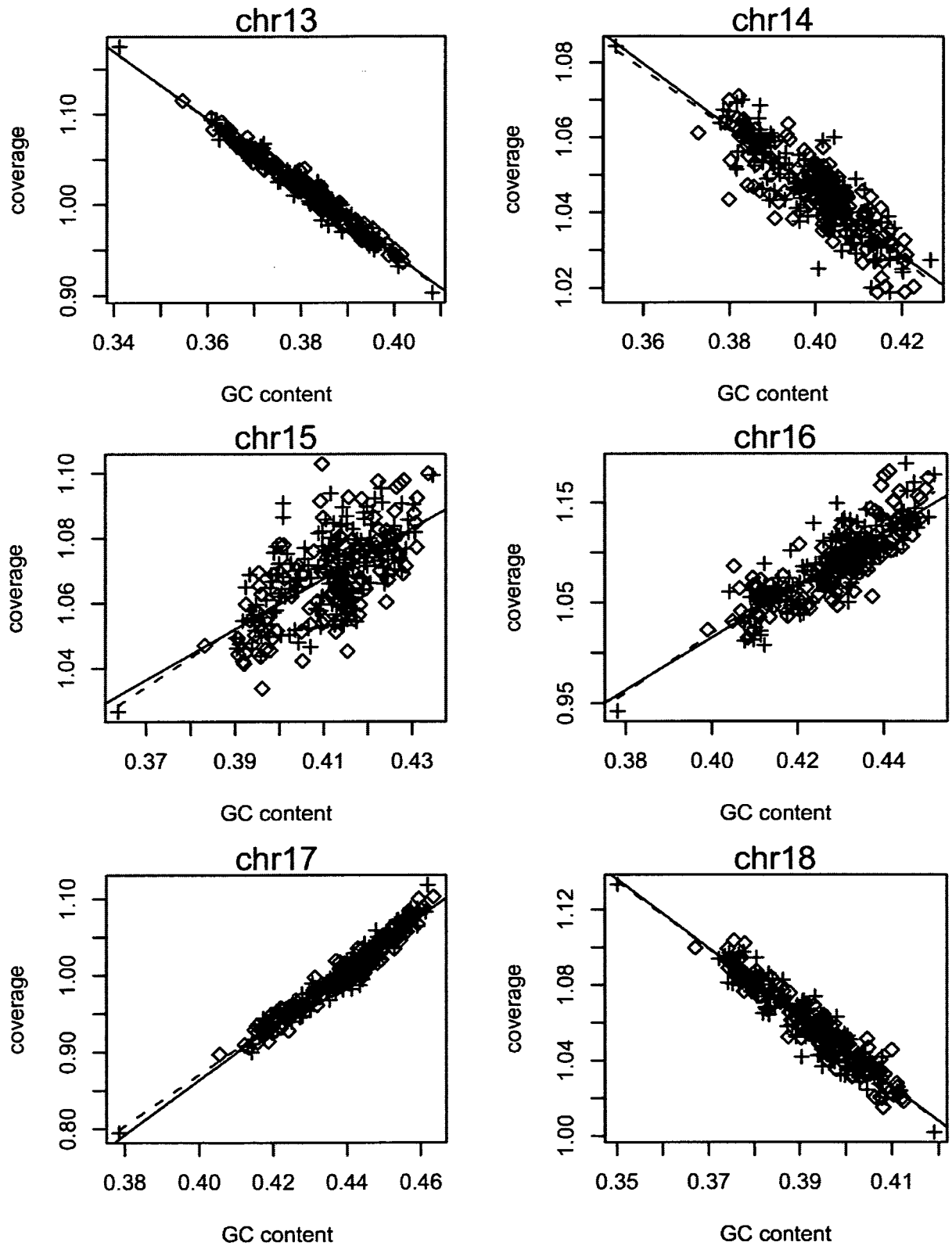

Figure 2
(page 3 of 4)

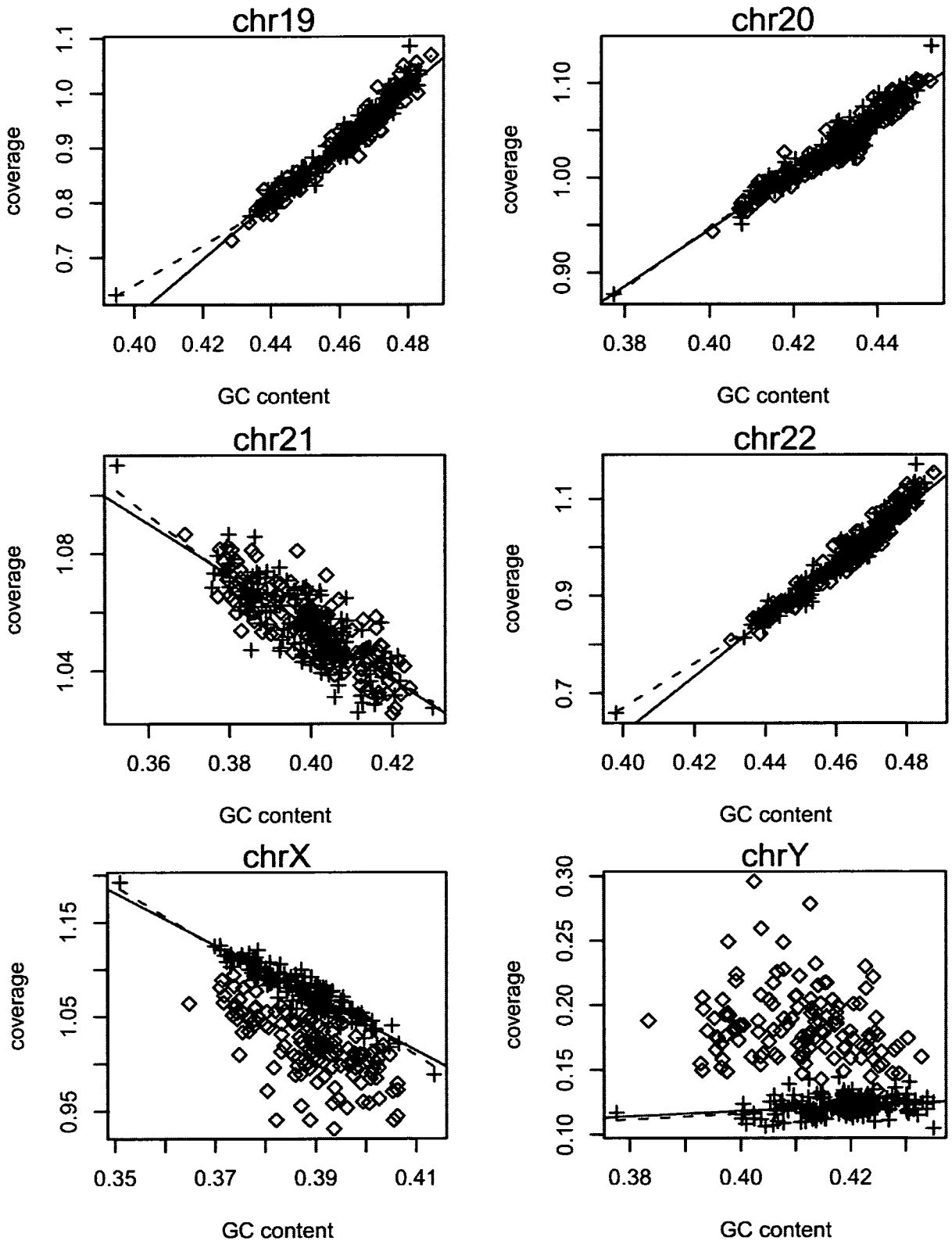

Figure 2
(page 4 of 4)

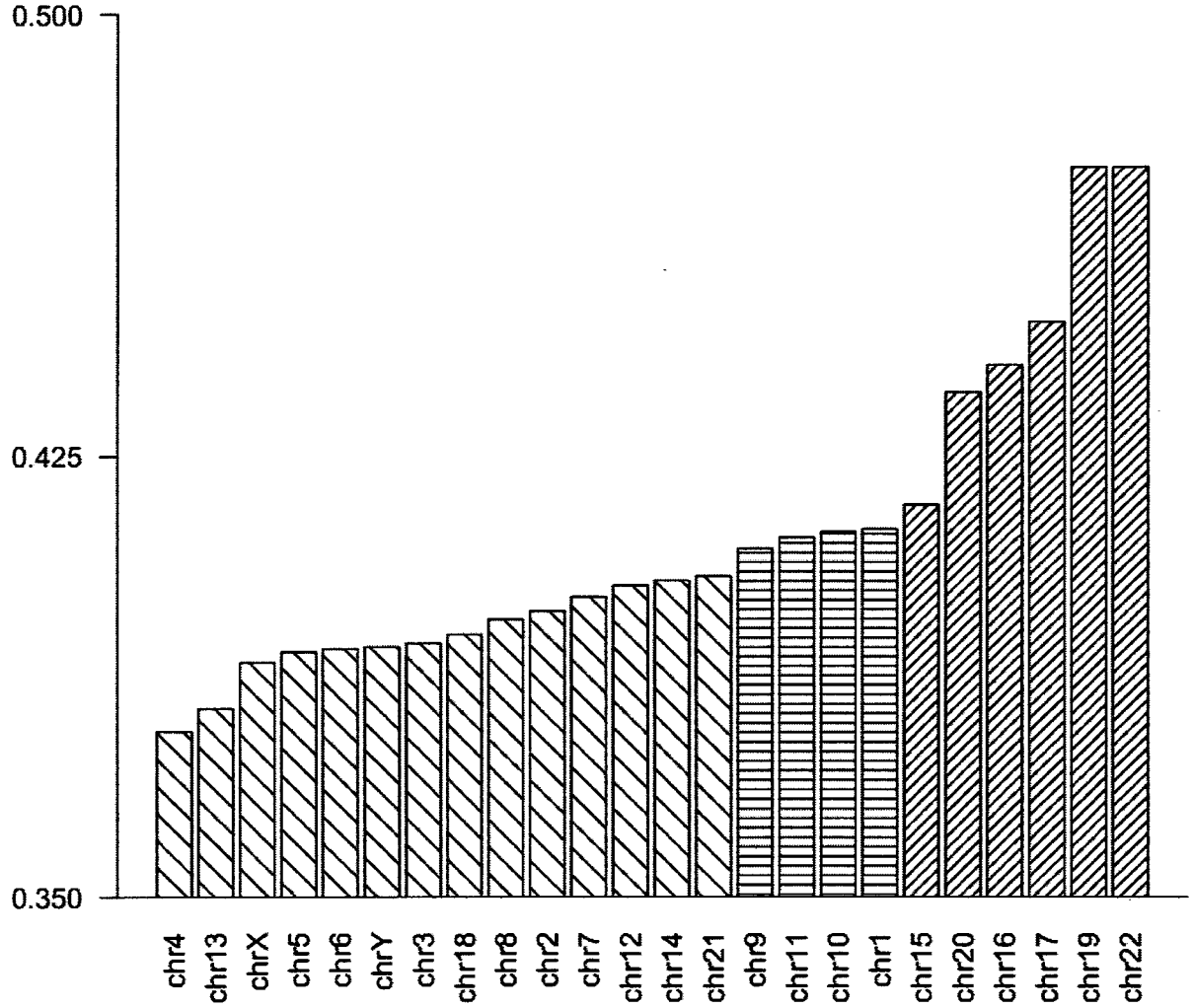
0.500
0.425
0.350
chr4
chr13
chrX
chr5
chr6
chrY
chr3
chr18
chr8
chr2
chr7
chr12
chr14
chr21
chr9
chr11
chr10
chr1
chr15
chr20
chr16
chr17
chr19
chr22

Figure 3

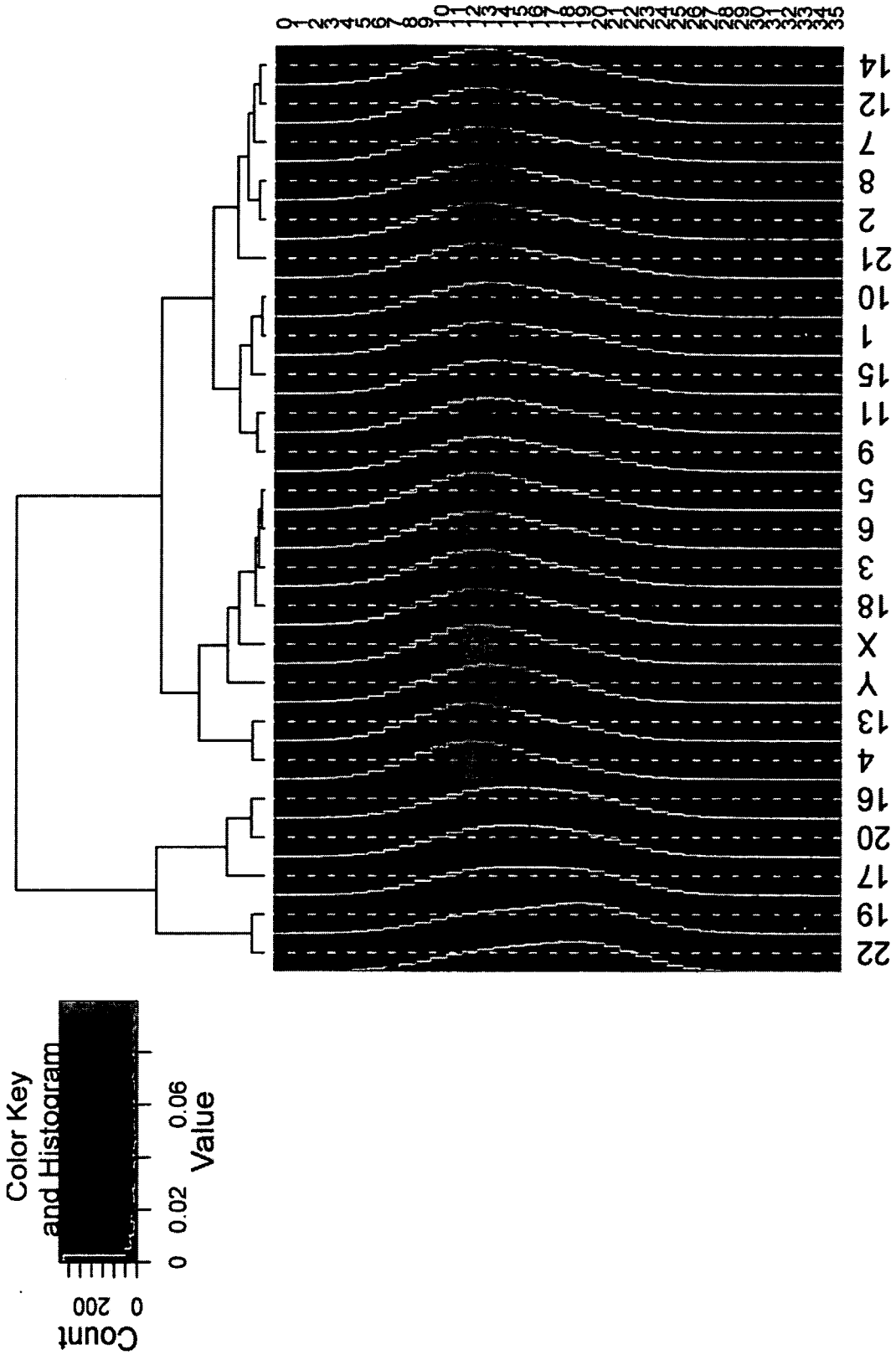
14
12
7
8
2
21
10
1
15
11
9
5
6
3
18
X
Y
13
4
16
20
17
19
22
Color Key
and Histogram
Count
0
200
0
0.02
0.06
Value

Figure 4

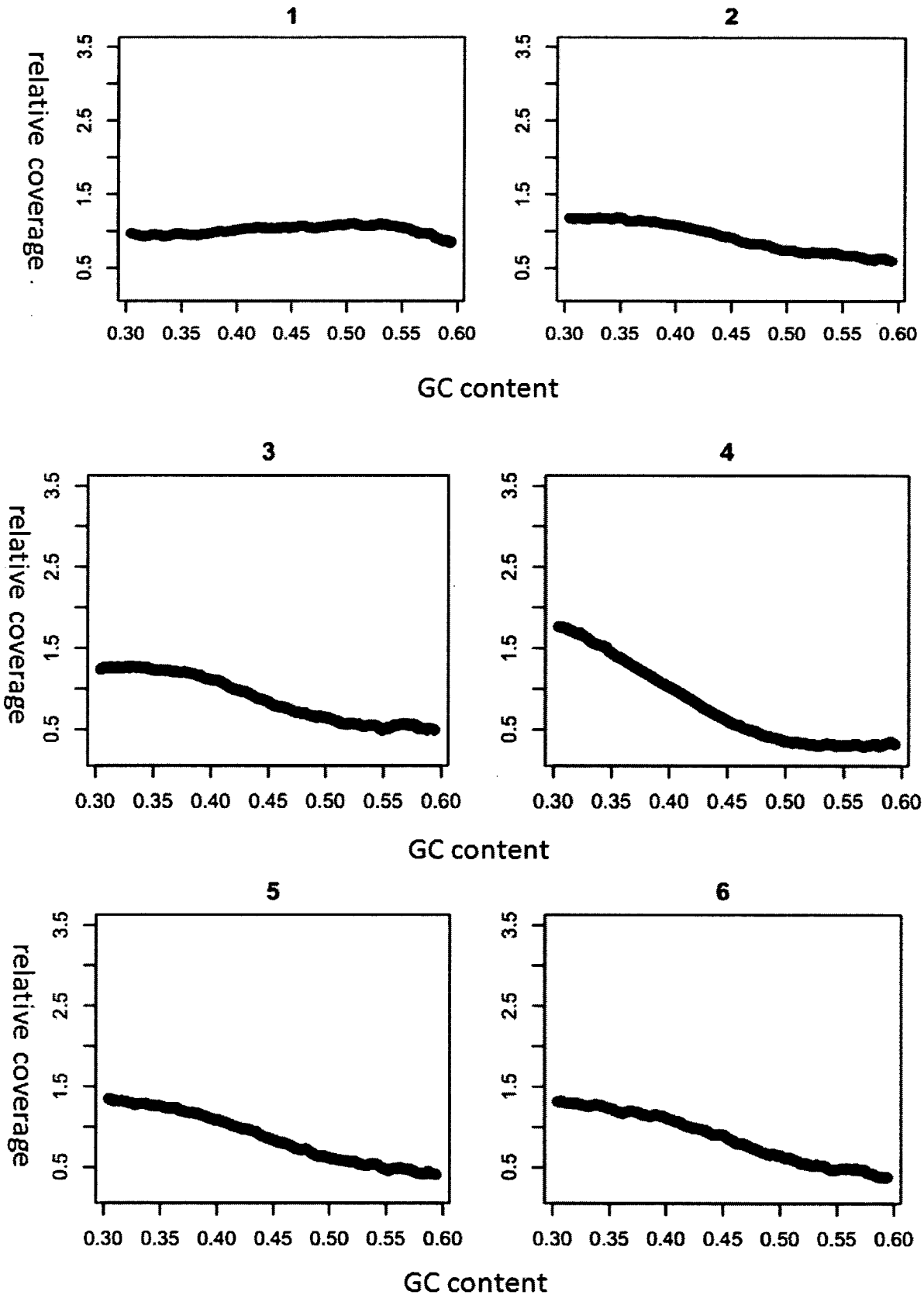

Figure 5
(page 1 of 4)

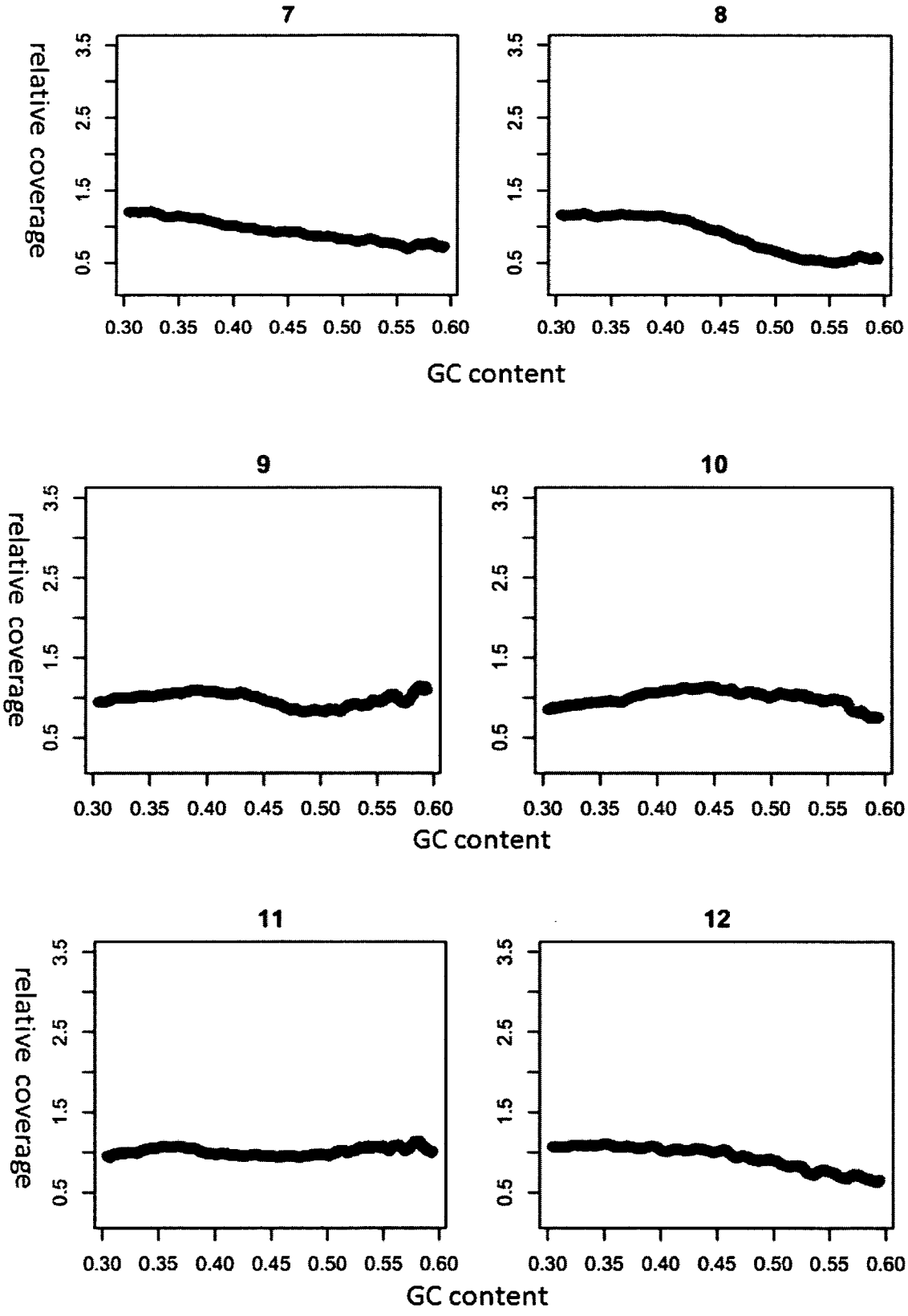

Figure 5
(page 2 of 4)

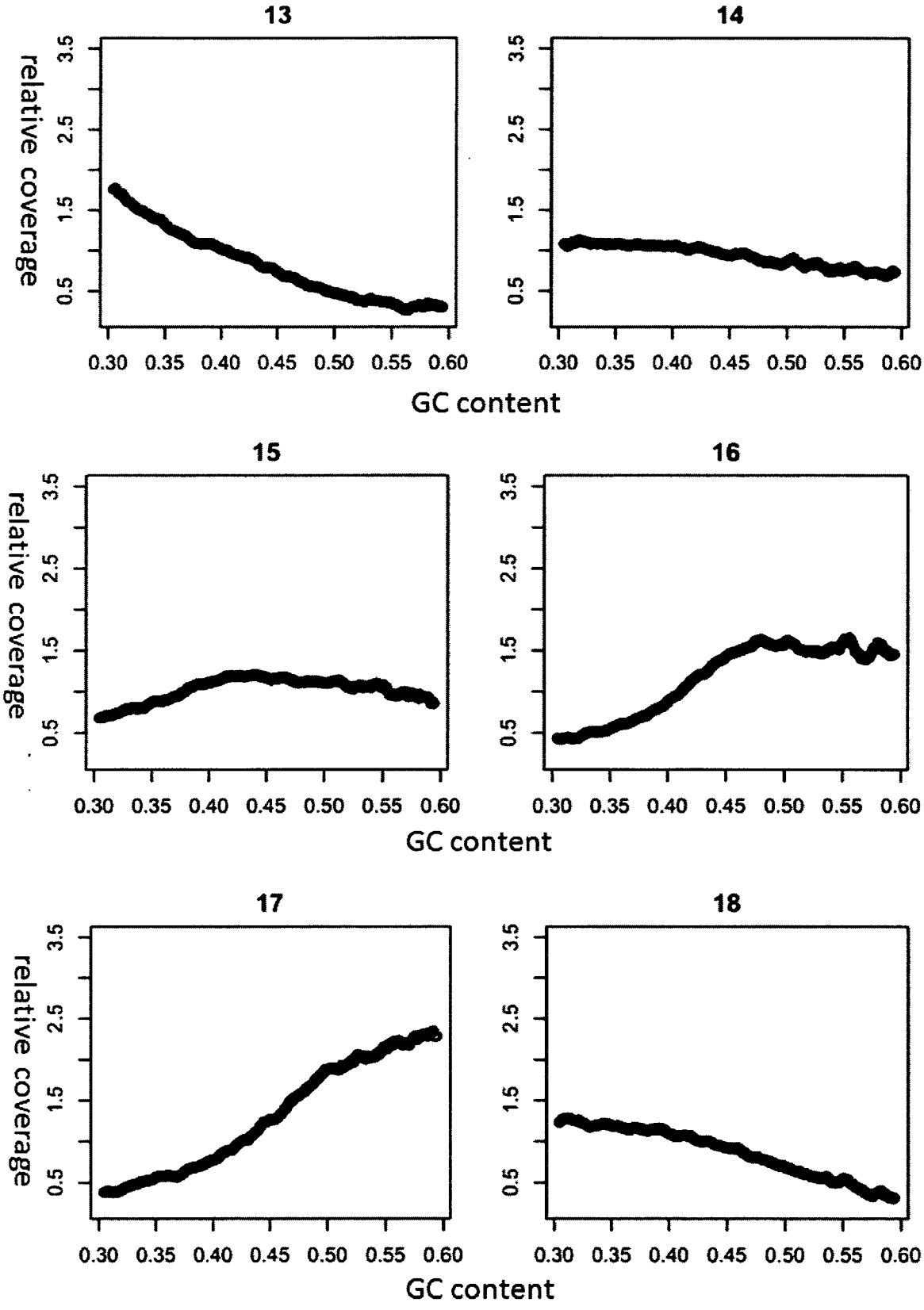

Figure 5
(page 3 of 4)

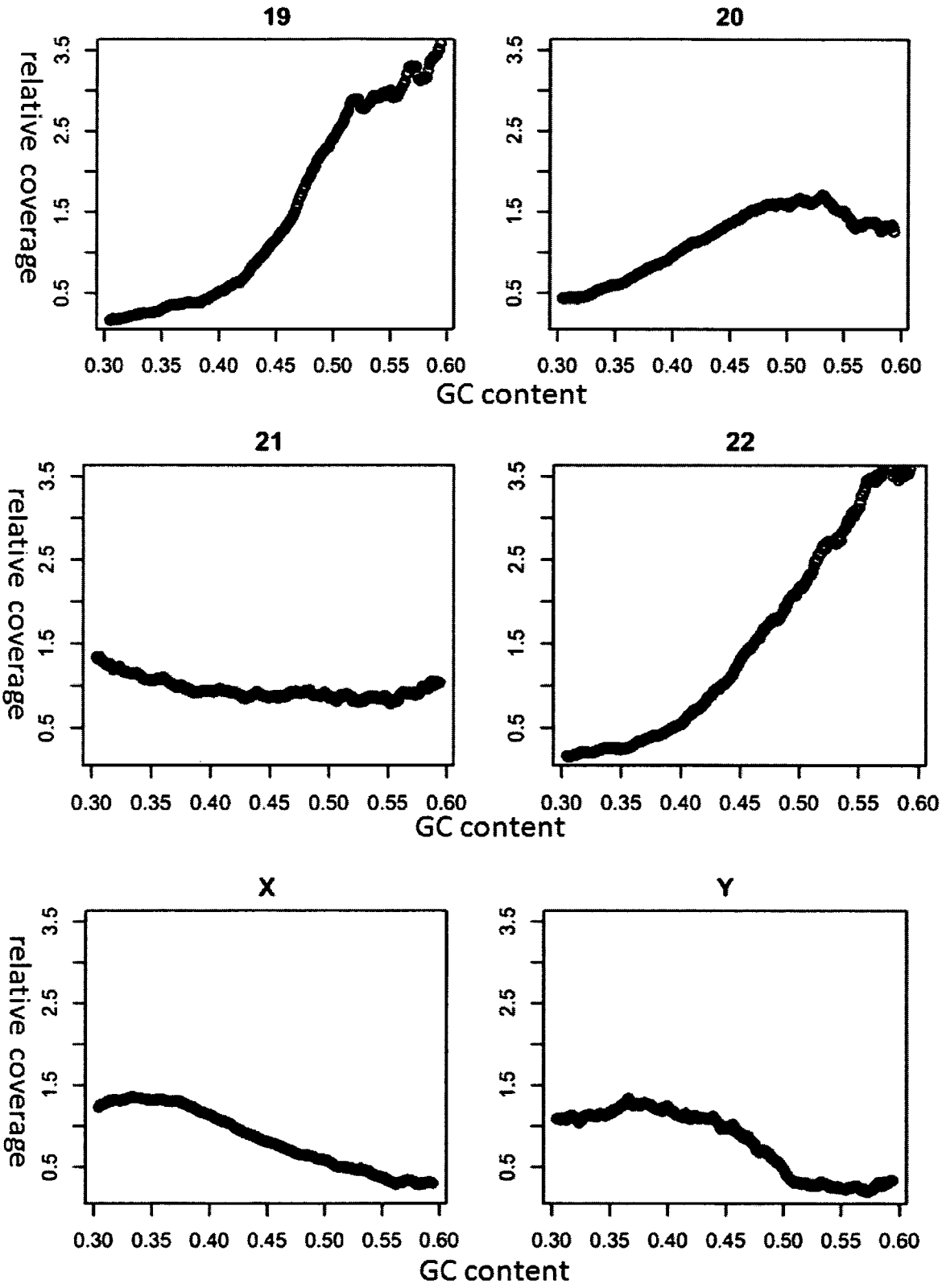

Figure 5
(page 4 of 4)

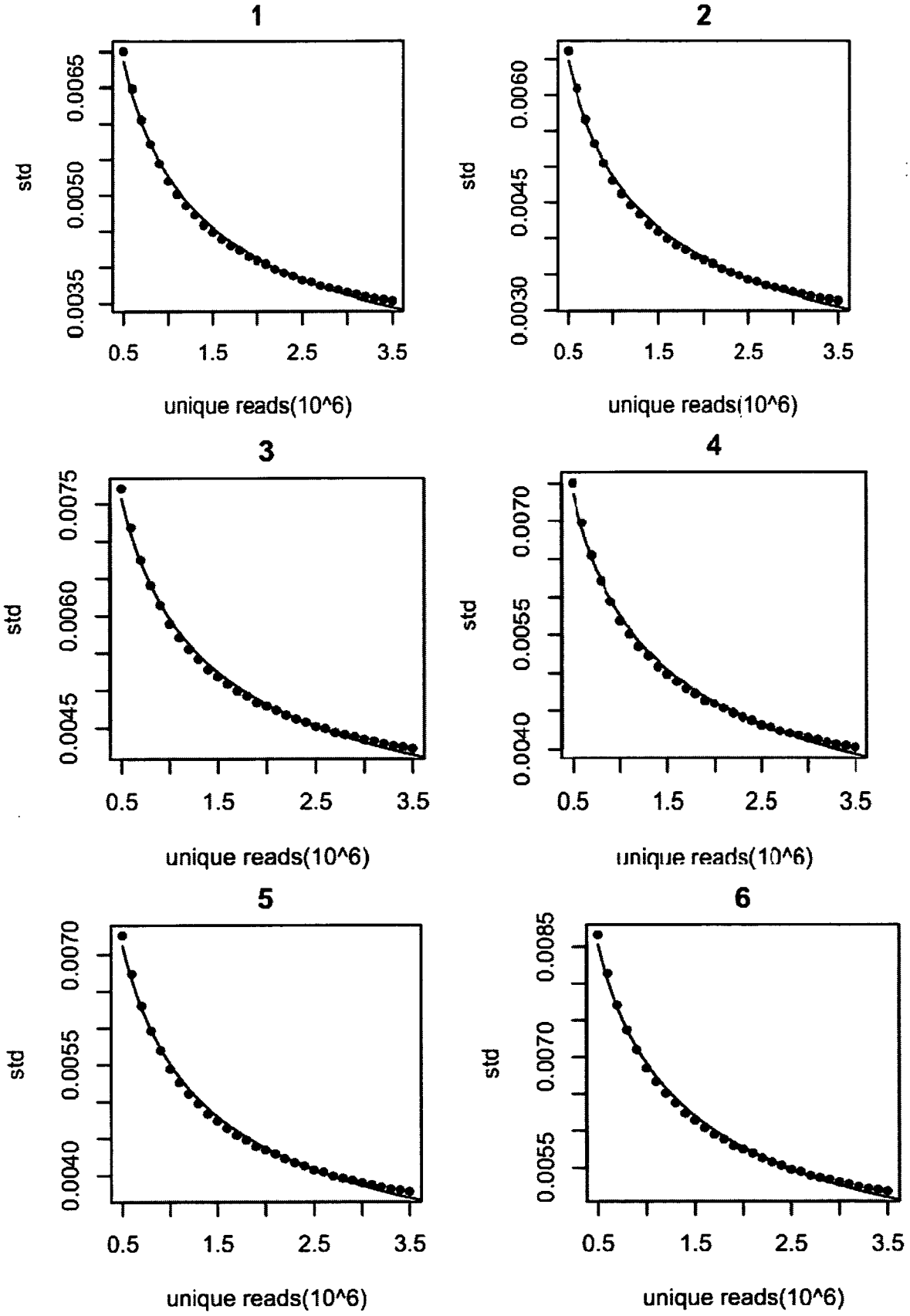

Figure 6
(page 1 of 4)

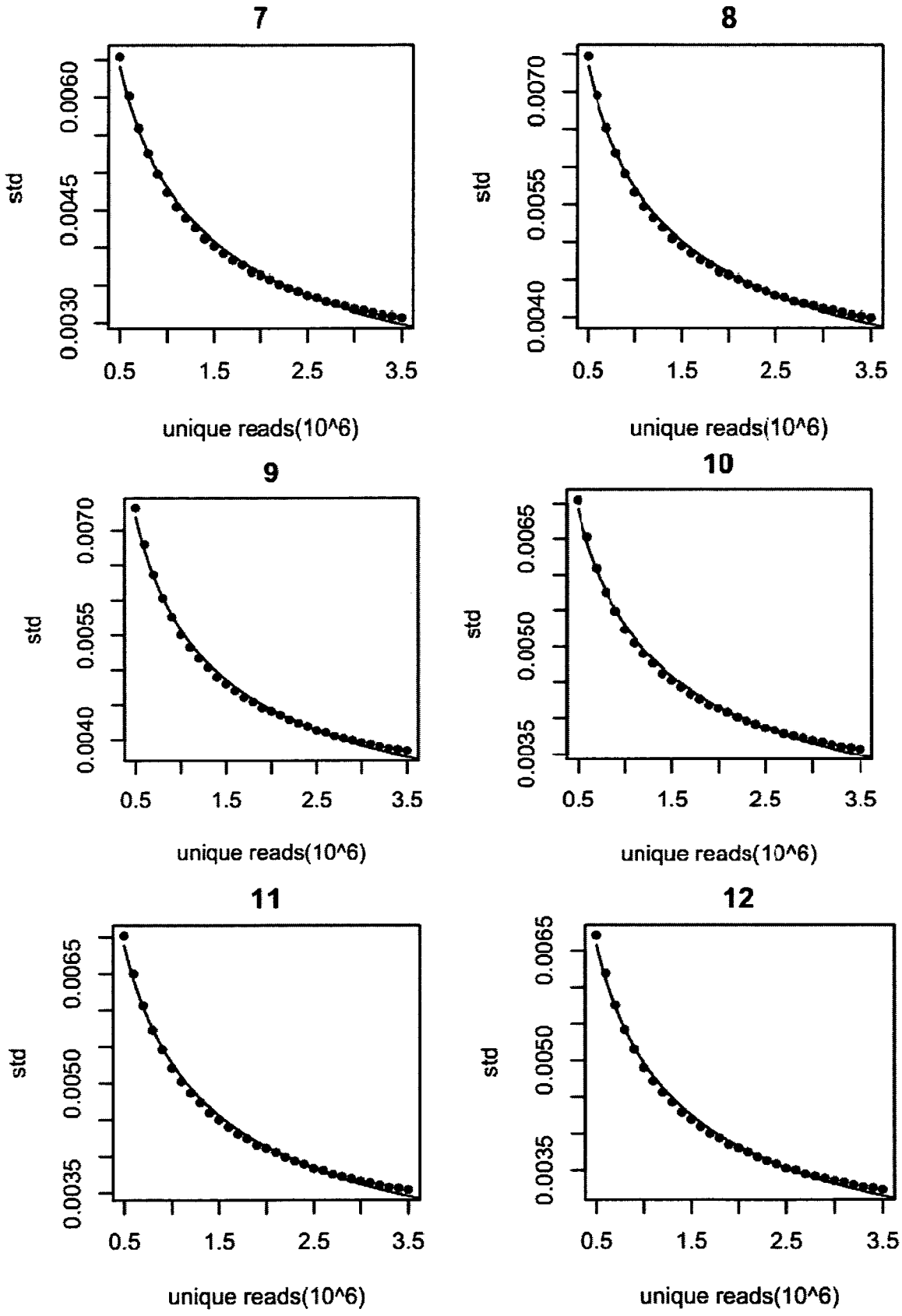

Figure 6
(page 2 of 4)

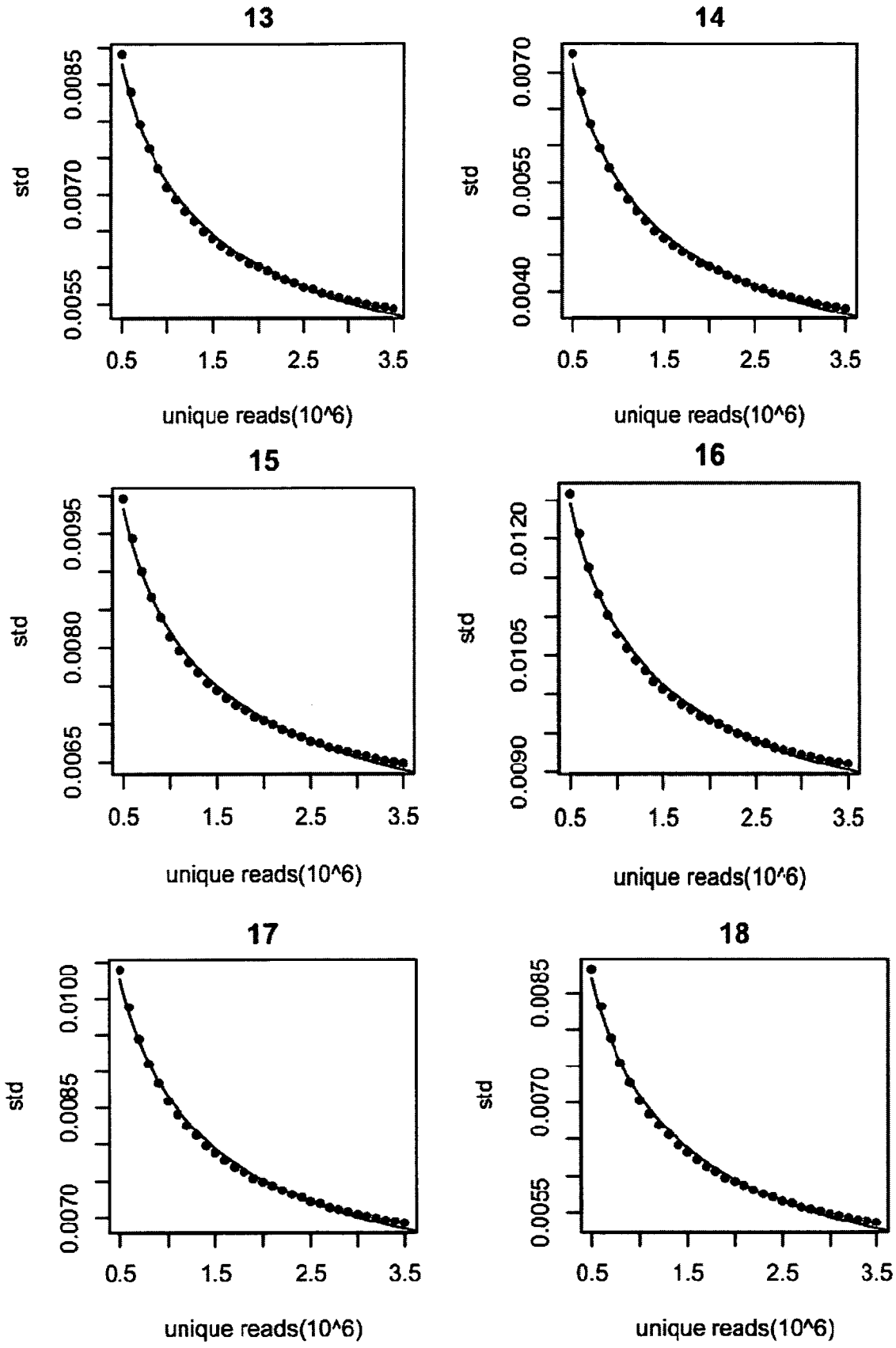

Figure 6
(page 3 of 4)

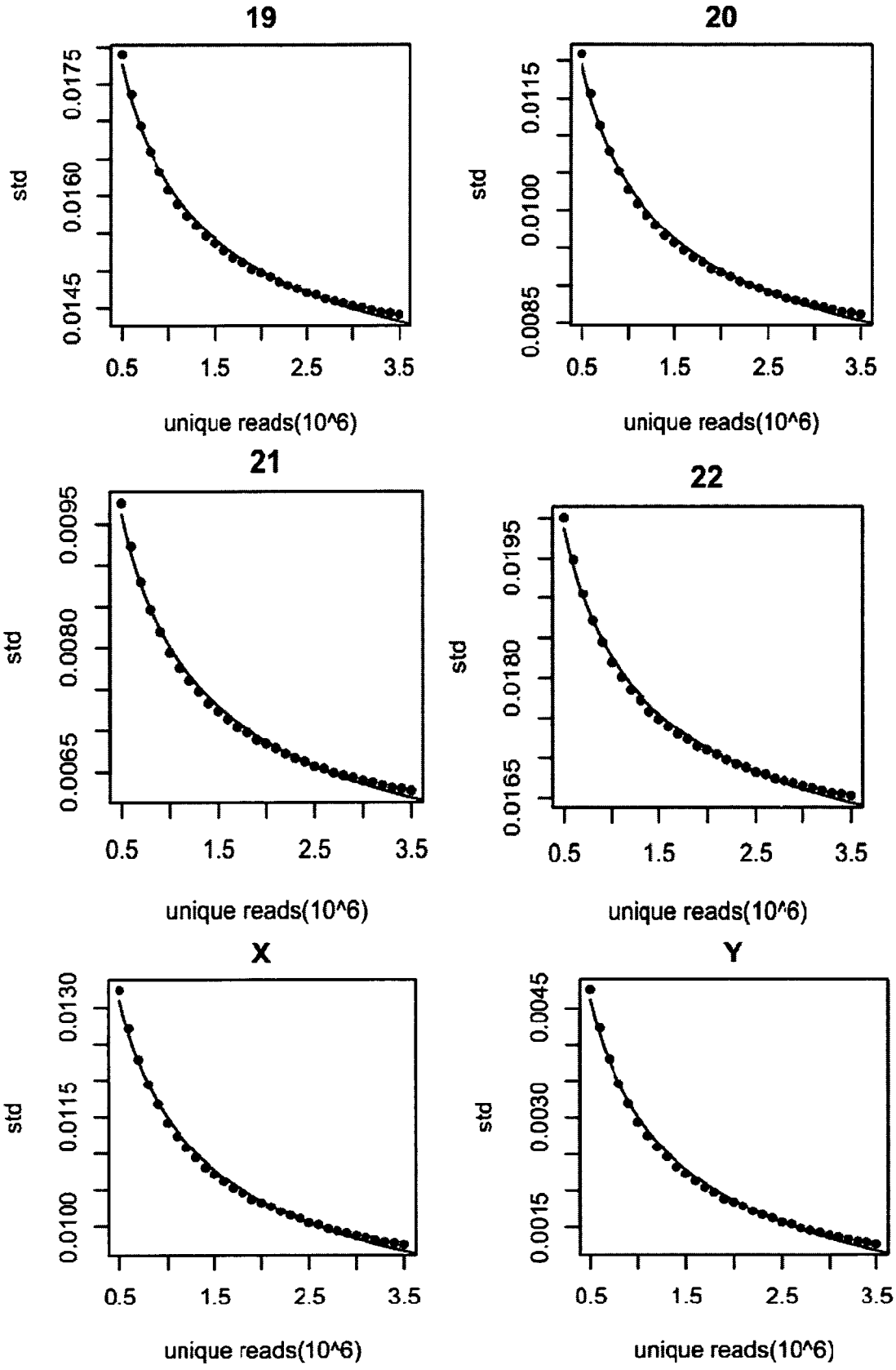

Figure 6
(page 4 of 4)

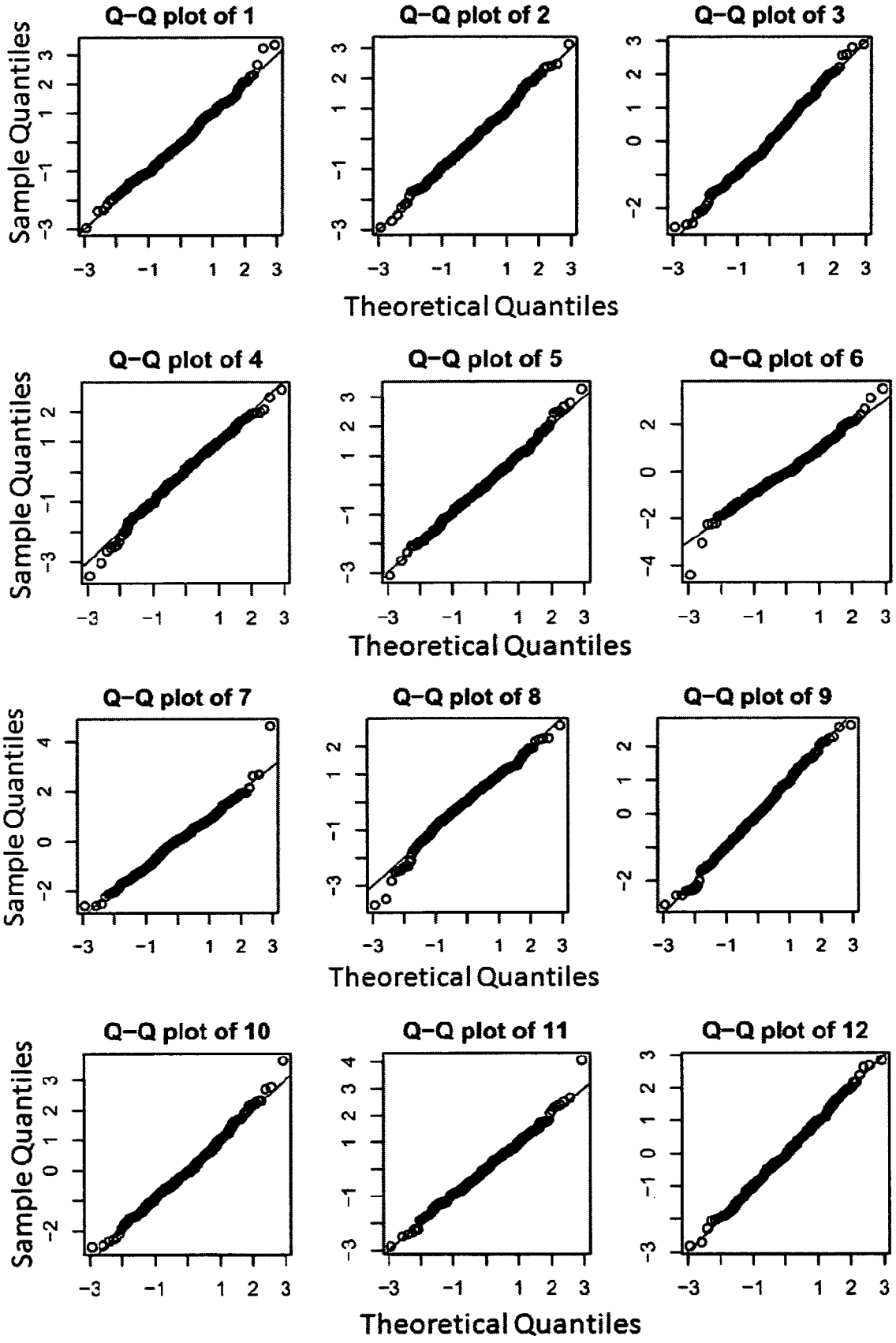

Figure 7
(page 1 of 2)

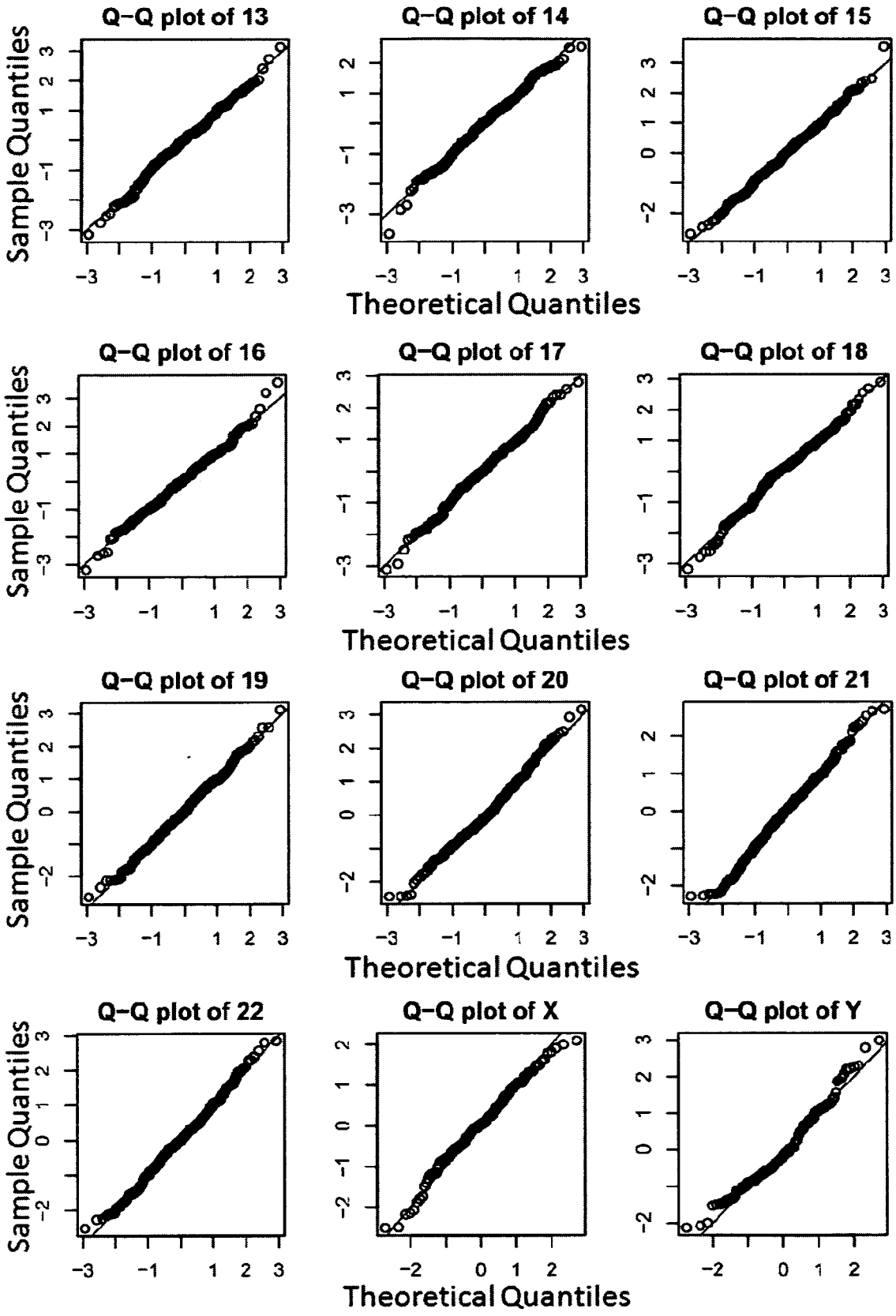

Figure 7
(page 2 of 2)

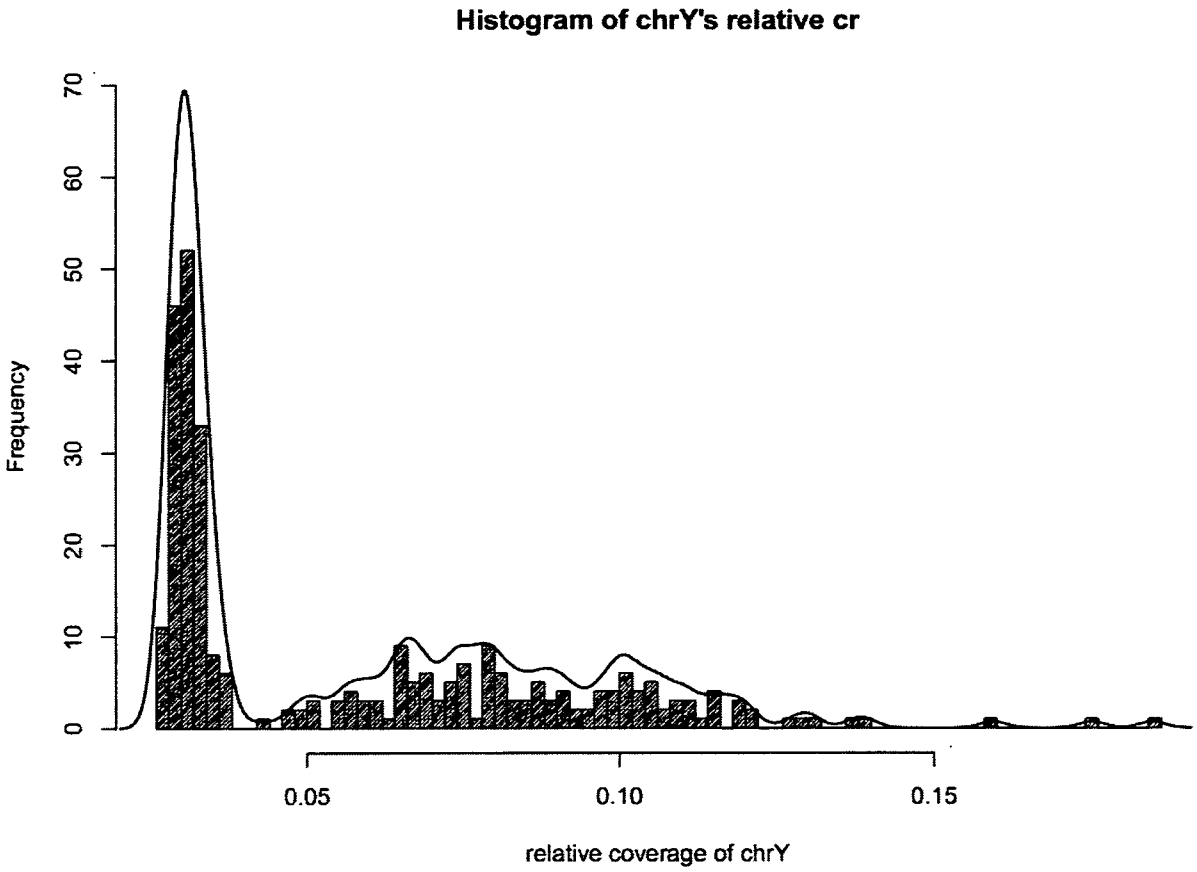
Histogram of chrY's relative cr
Frequency
0
10
20
30
40
50
60
70
0.05
0.10
0.15
relative coverage of chrY

Figure 8

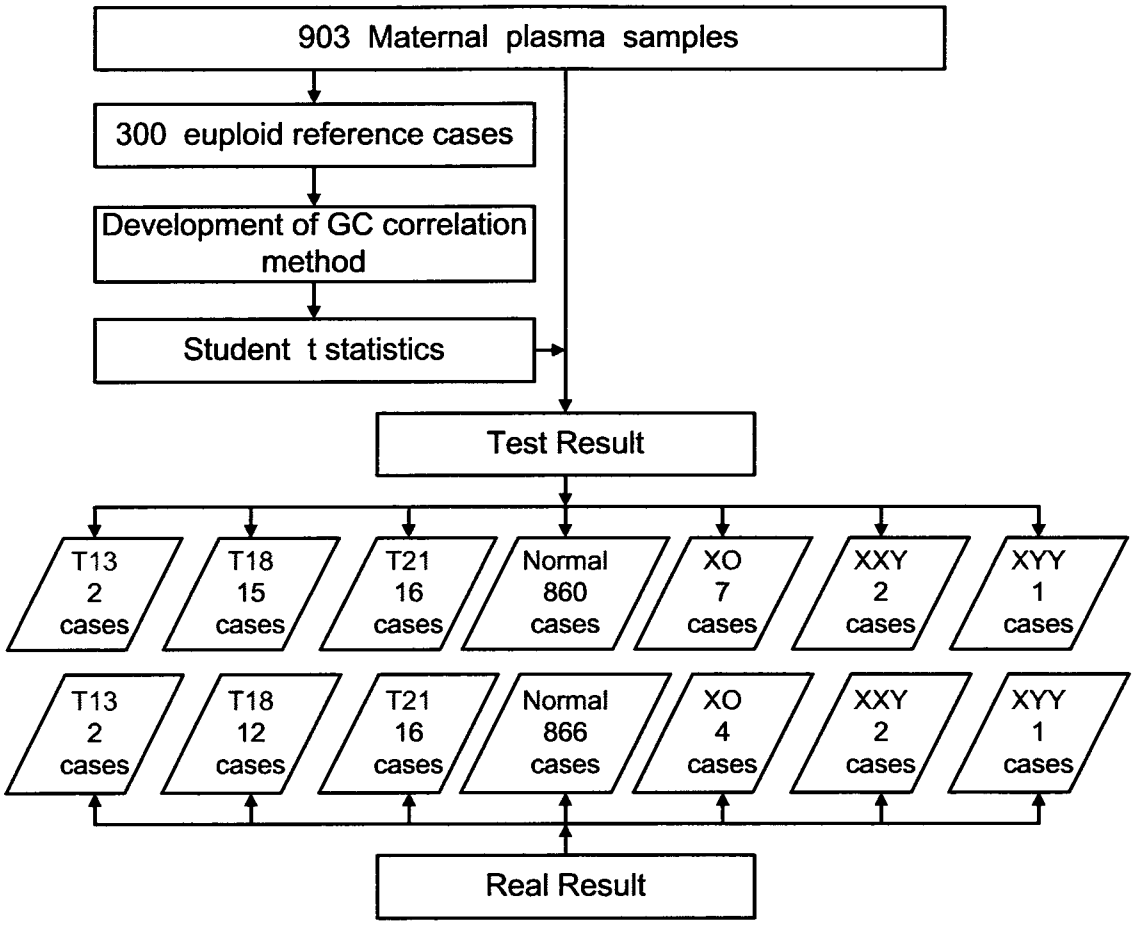
903 Maternal plasma samples
300 euploid reference cases
Development of GC correlation method
Student t statistics
Test Result
T13 2 cases
T18 15 cases
T21 16 cases
Normal 860 cases
XO 7 cases
XXY 2 cases
XYY 1 cases
T13 2 cases
T18 12 cases
T21 16 cases
Normal 866 cases
XO 4 cases
XXY 2 cases
XYY 1 cases
Real Result

Figure 9

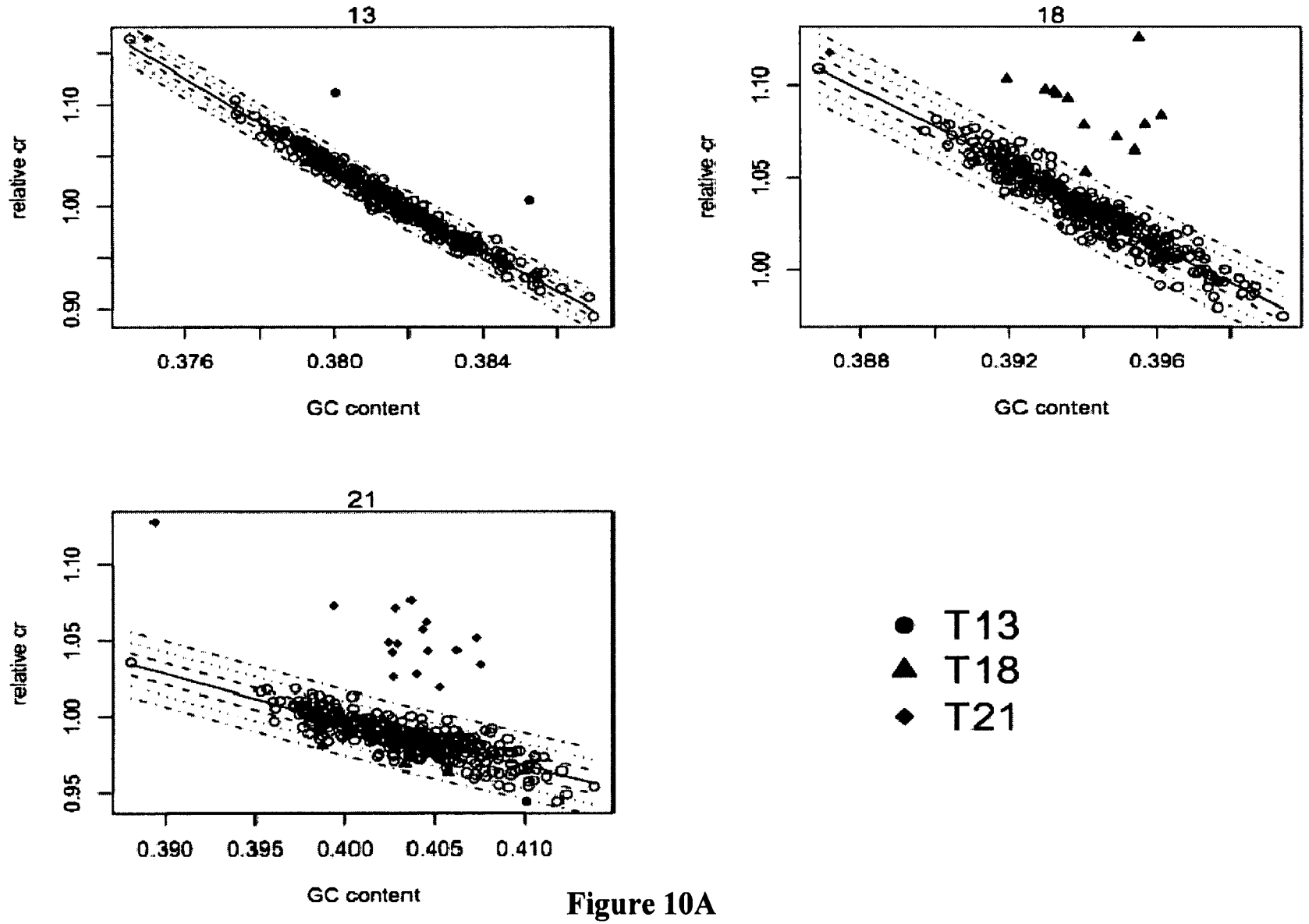
13
relative cr
1.10
1.00
0.90
0.376
0.380
0.384
GC content
18
relative cr
1.10
1.05
1.00
0.388
0.392
0.396
GC content
21
relative cr
1.10
1.05
1.00
0.95
0.390
0.395
0.400
0.405
0.410
GC content
T13
T18
T21

Figure 10A

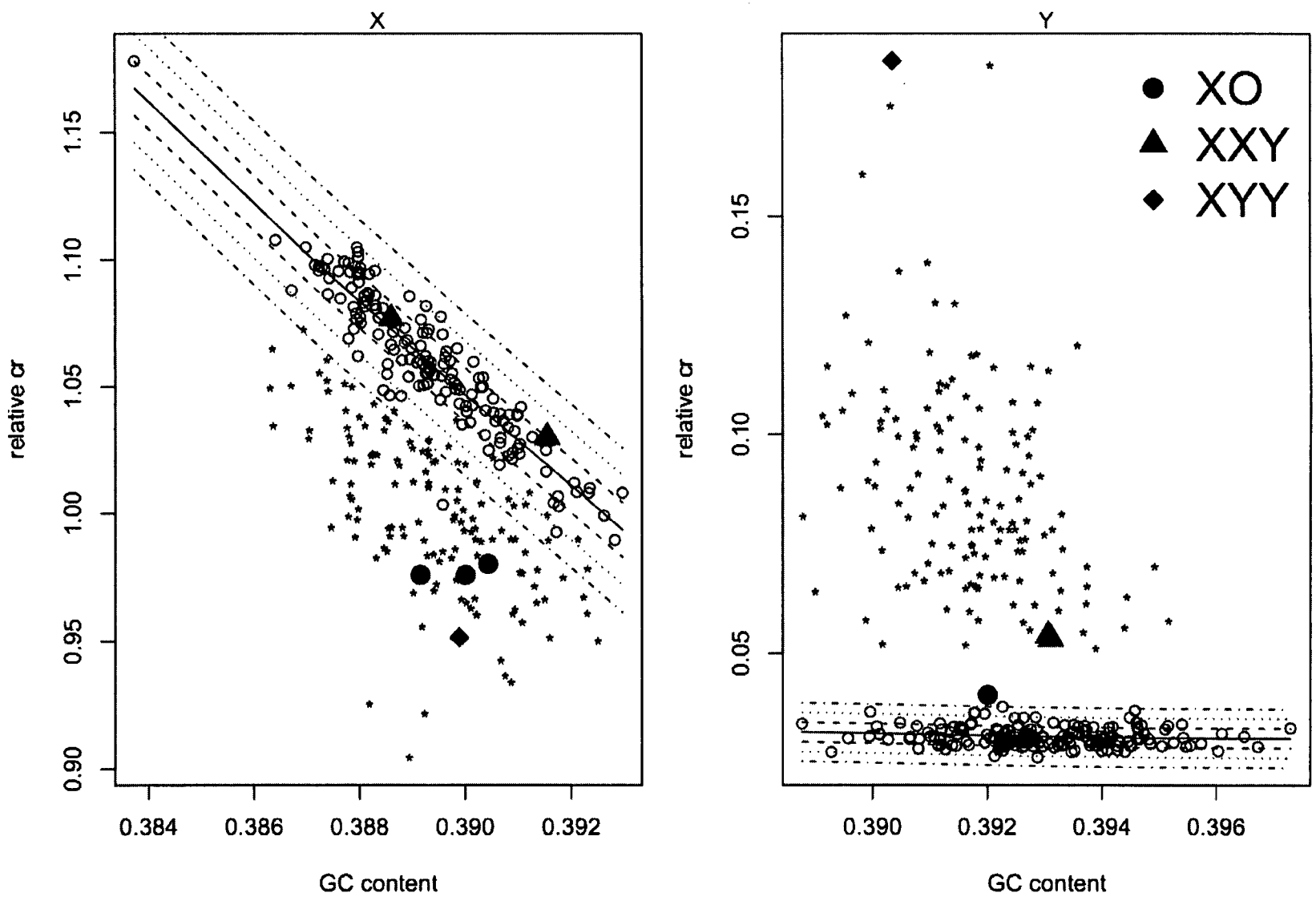
X
Y
relative cr
relative cr
0.90
0.95
1.00
1.05
1.10
1.15
0.05
0.10
0.15
0.384
0.386
0.388
0.390
0.392
0.390
0.392
0.394
0.396
GC content
GC content
XO
XXY
XYY

Figure 10B

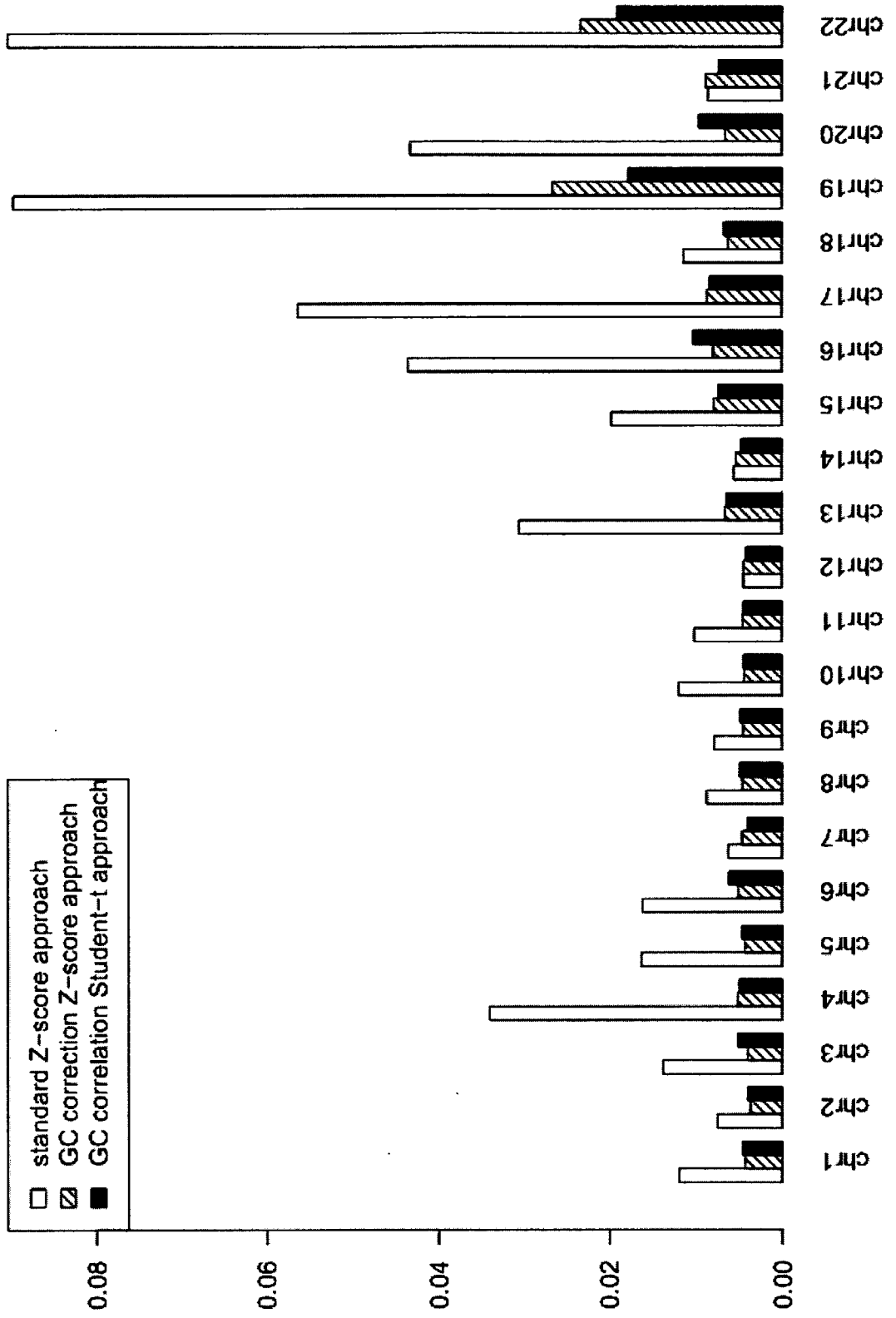
standard Z-score approach
GC correction Z-score approach
GC correlation Student-t approach
0.00
0.02
0.04
0.06
0.08
chr1
chr2
chr3
chr4
chr5
chr6
chr7
chr8
chr9
chr10
chr11
chr12
chr13
chr14
chr15
chr16
chr17
chr18
chr19
chr20
chr21
chr22

Figure 11

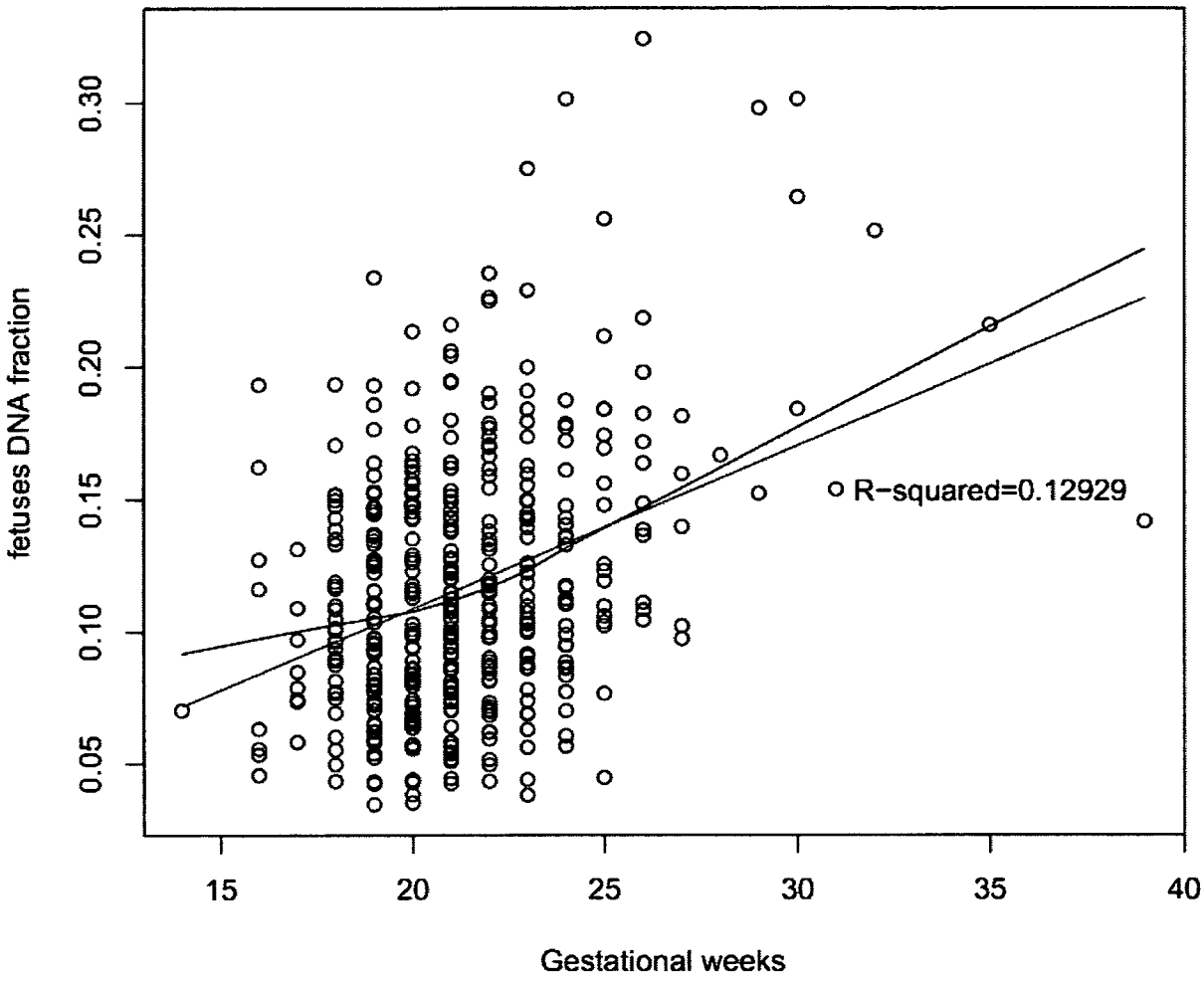
fetuses DNA fraction
0.05
0.10
0.15
0.20
0.25
0.30
15
20
25
30
35
40
Gestational weeks
R-squared=0.12929

Figure 12

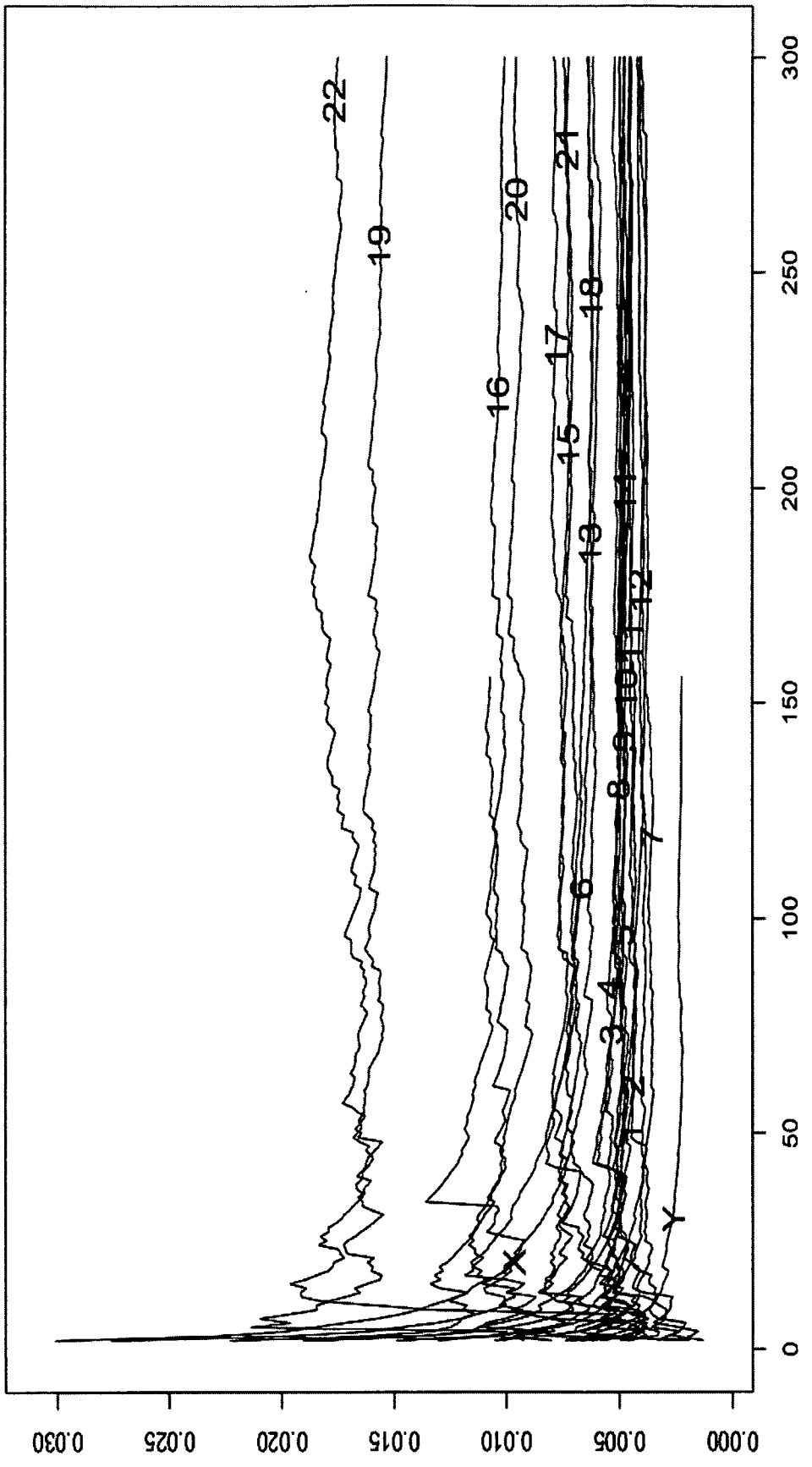
standard variance
0.000
0.005
0.010
0.015
0.020
0.025
0.030
sample number
0
50
100
150
200
250
300
22
19
20
21
16
17
18
15
13
14
12
6
7
8
9
10
11
1
2
3
4
5
X
Y

Figure 13

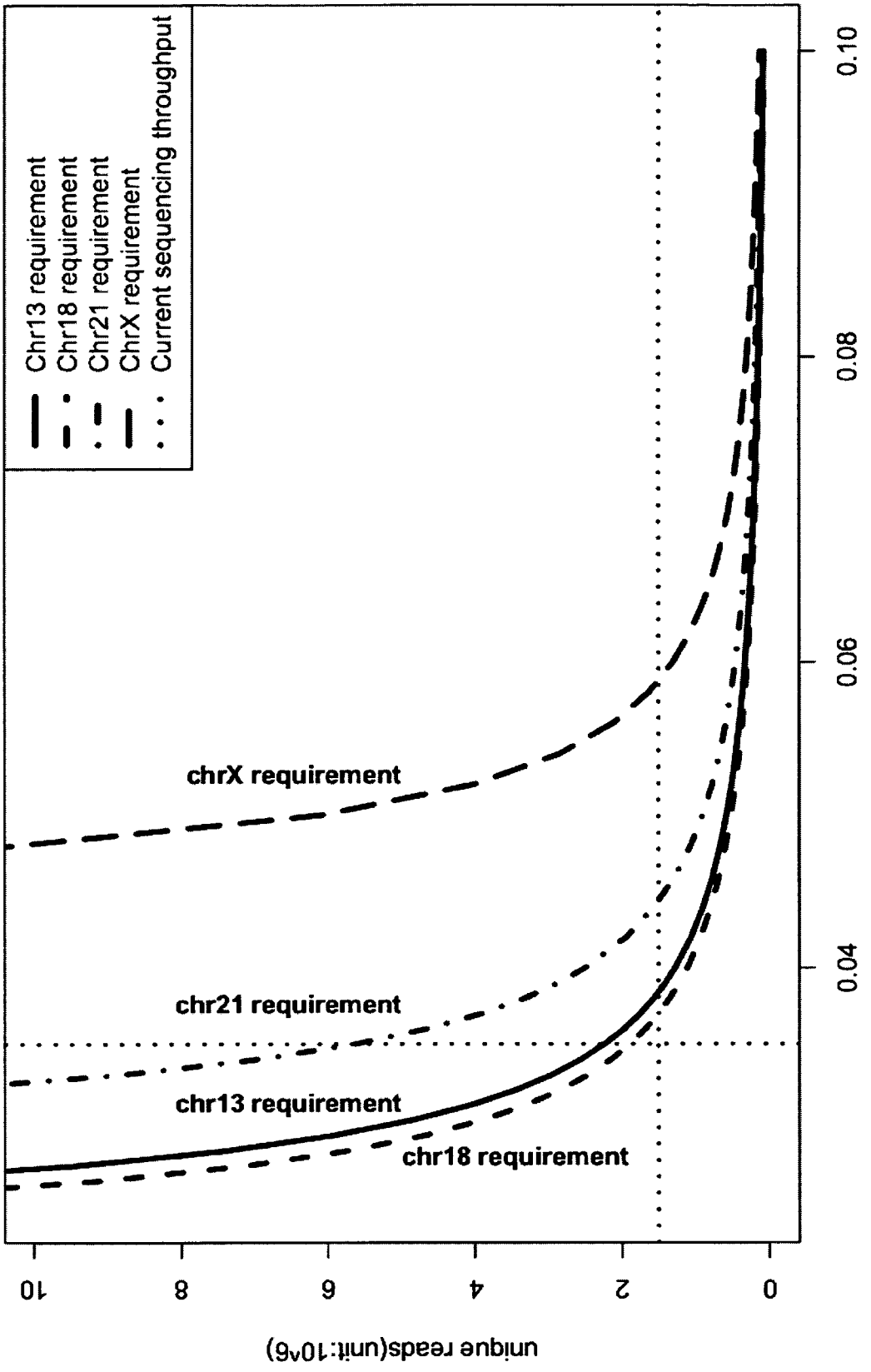
Chr13 requirement
Chr18 requirement
Chr21 requirement
ChrX requirement
Current sequencing throughput
chrX requirement
chr21 requirement
chr13 requirement
chr18 requirement
0
2
4
6
8
10
unique reads(unit:10^6)
0.04
0.06
0.08
0.10
fetal DNA fraction

Figure 14

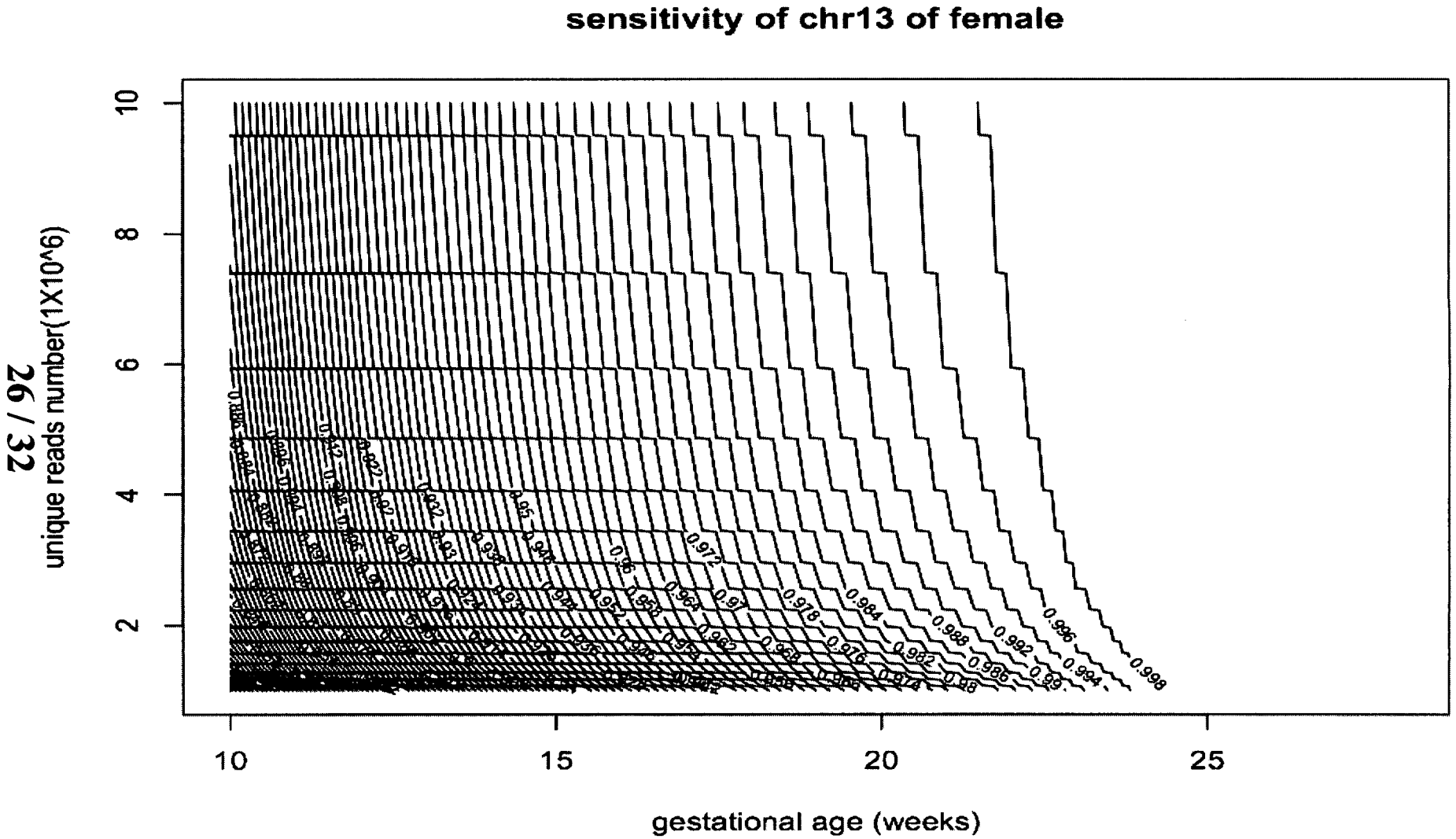
sensitivity of chr13 of female
unique reads number(1X10^6)
2
4
6
8
10
10
15
20
25
gestational age (weeks)
0.998
0.996
0.994
0.992
0.99
0.988
0.986
0.984
0.982
0.98
0.978
0.976
0.974
0.972
0.97
0.968
0.966
0.964
0.962
0.96
0.958
0.956
0.954
0.952
0.95
0.948
0.946
0.944
0.942
0.94
0.938
0.936
0.934
0.932
0.93
0.928
0.926
0.924
0.922
0.92
0.918
0.916
0.914
0.912
0.91
0.908
0.906
0.904
0.902
0.9
0.898
0.896
0.894
0.892
0.89
0.888
0.886
0.884
0.882
0.88
0.878
0.876
0.874
0.872
0.87
0.868
0.866
0.864
0.862
0.86
0.858
0.856
0.854
0.852
0.85
0.848
0.846
0.844
0.842
0.84
0.838
0.836
0.834
0.832
0.83
0.828
0.826
0.824
0.822
0.82
0.818
0.816
0.814
0.812
0.81
0.808
0.806
0.804
0.802
0.8
0.798
0.796
0.794
0.792
0.79
0.788
0.786
0.784
0.782
0.78
0.778
0.776
0.774
0.772
0.77
0.768
0.766
0.764
0.762
0.76
0.758
0.756
0.754
0.752
0.75
0.748
0.746
0.744
0.742
0.74
0.738
0.736
0.734
0.732
0.73
0.728
0.726
0.724
0.722
0.72
0.718
0.716
0.714
0.712
0.71
0.708
0.706
0.704
0.702
0.7
0.698
0.696
0.694
0.692
0.69
0.688
0.686
0.684
0.682
0.68
0.678
0.676
0.674
0.672
0.67
0.668
0.666
0.664
0.662
0.66
0.658
0.656
0.654
0.652
0.65
0.648
0.646
0.644
0.642
0.64
0.638
0.636
0.634
0.632
0.63
0.628
0.626
0.624
0.622
0.62
0.618
0.616
0.614
0.612
0.61
0.608
0.606
0.604
0.602
0.6
0.598
0.596
0.594
0.592
0.59
0.588
0.586
0.584
0.582
0.58
0.578
0.576
0.574
0.572
0.57
0.568
0.566
0.564
0.562
0.56
0.558
0.556
0.554
0.552
0.55
0.548
0.546
0.544
0.542
0.54
0.538
0.536
0.534
0.532
0.53
0.528
0.526
0.524
0.522
0.52
0.518
0.516
0.514
0.512
0.51
0.508
0.506
0.504
0.502
0.5
0.498
0.496
0.494
0.492
0.49
0.488
0.486
0.484
0.482
0.48
0.478
0.476
0.474
0.472
0.47
0.468
0.466
0.464
0.462
0.46
0.458
0.456
0.454
0.452
0.45
0.448
0.446
0.444
0.442
0.44
0.438
0.436
0.434
0.432
0.43
0.428
0.426
0.424
0.422
0.42
0.418
0.416
0.414
0.412
0.41
0.408
0.406
0.404
0.402
0.4
0.398
0.396
0.394
0.392
0.39
0.388
0.386
0.384
0.382
0.38
0.378
0.376
0.374
0.372
0.37
0.368
0.366
0.364
0.362
0.36
0.358
0.356
0.354
0.352
0.35
0.348
0.346
0.344
0.342
0.34
0.338
0.336
0.334
0.332
0.33
0.328
0.326
0.324
0.322
0.32
0.318
0.316
0.314
0.312
0.31
0.308
0.306
0.304
0.302
0.3
0.298
0.296
0.294
0.292
0.29
0.288
0.286
0.284
0.282
0.28
0.278
0.276
0.274
0.272
0.27
0.268
0.266
0.264
0.262
0.26
0.258
0.256
0.254
0.252
0.25
0.248
0.246
0.244
0.242
0.24
0.238
0.236
0.234
0.232
0.23
0.228
0.226
0.224
0.222
0.22
0.218
0.216
0.214
0.212
0.21
0.208
0.206
0.204
0.202
0.2
0.198
0.196
0.194
0.192
0.19
0.188
0.186
0.184
0.182
0.18
0.178
0.176
0.174
0.172
0.17
0.168
0.166
0.164
0.162
0.16
0.158
0.156
0.154
0.152
0.15
0.148
0.146
0.144
0.142
0.14
0.138
0.136
0.134
0.132
0.13
0.128
0.126
0.124
0.122
0.12
0.118
0.116
0.114
0.112
0.11
0.108
0.106
0.104
0.102
0.1
0.098
0.096
0.094
0.092
0.09
0.088
0.086
0.084
0.082
0.08
0.078
0.076
0.074
0.072
0.07
0.068
0.066
0.064
0.062
0.06
0.058
0.056
0.054
0.052
0.05
0.048
0.046
0.044
0.042
0.04
0.038
0.036
0.034
0.032
0.03
0.028
0.026
0.024
0.022
0.02
0.018
0.016
0.014
0.012
0.01
0.008
0.006
0.004
0.002
0

Figure 15

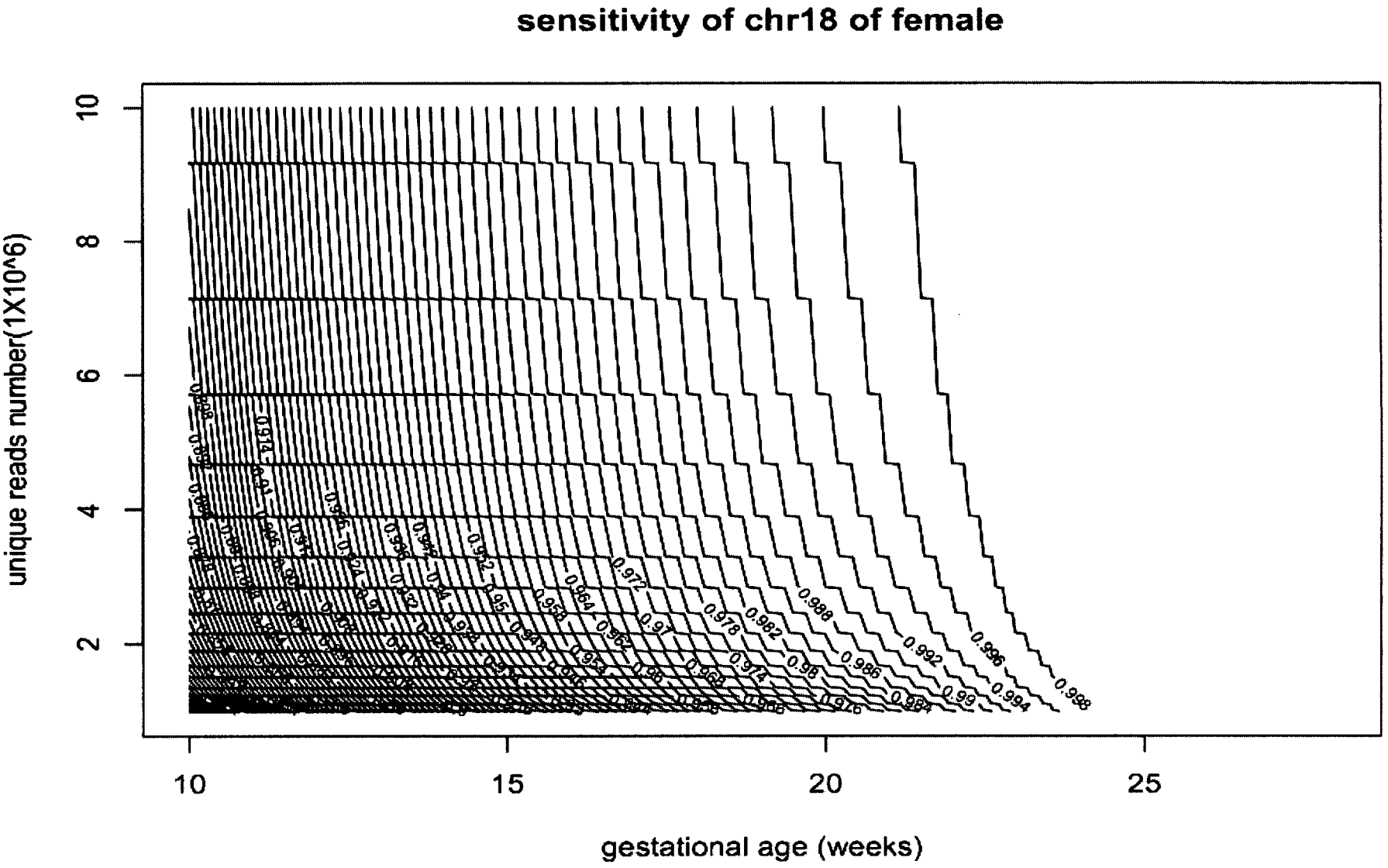
sensitivity of chr18 of female
unique reads number(1X10^6)
gestational age (weeks)
10
15
20
25
2
4
6
8
10
0.998
0.996
0.994
0.992
0.99
0.988
0.986
0.984
0.982
0.98
0.978
0.976
0.974
0.972
0.97
0.968
0.966
0.964
0.962
0.96
0.958
0.956
0.954
0.952
0.95
0.948
0.946
0.944
0.942
0.94
0.938
0.936
0.934
0.932
0.93
0.928
0.926
0.924
0.922
0.92
0.918
0.916
0.914
0.912
0.91
0.908
0.906
0.904
0.902
0.9
0.898
0.896
0.894
0.892
0.89
0.888
0.886
0.884
0.882
0.88
0.878
0.876
0.874
0.872
0.87
0.868
0.866
0.864
0.862
0.86
0.858
0.856
0.854
0.852
0.85
0.848
0.846
0.844
0.842
0.84
0.838
0.836
0.834
0.832
0.83
0.828
0.826
0.824
0.822
0.82
0.818
0.816
0.814
0.812
0.81
0.808
0.806
0.804
0.802
0.8

Figure 16

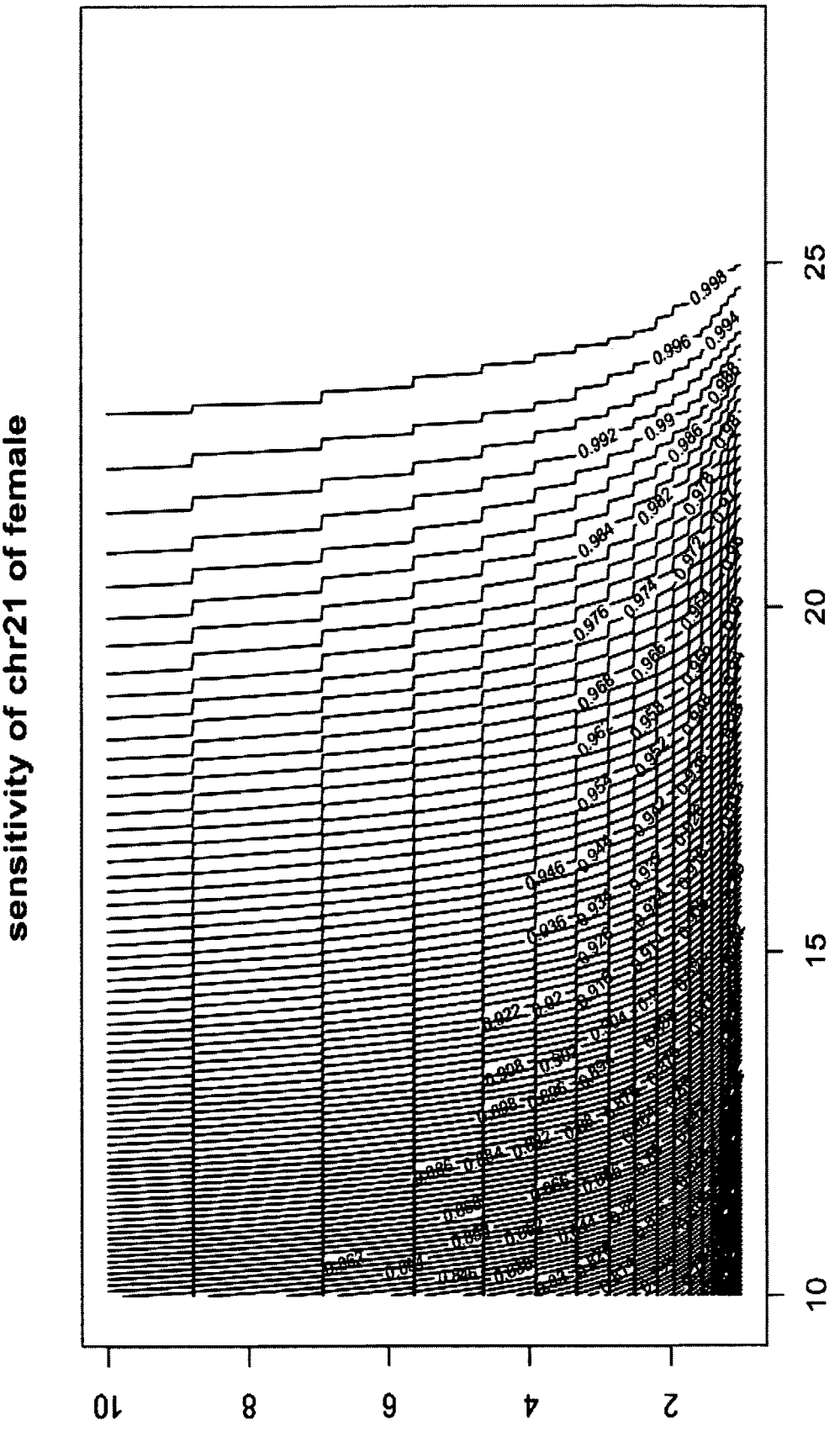
sensitivity of chr21 of female
unique reads number(unit:10^6)
gestational age (weeks)
2
4
6
8
10
15
20
25
0.998
0.996
0.994
0.992
0.99
0.988
0.986
0.984
0.982
0.98
0.978
0.976
0.974
0.972
0.97
0.968
0.966
0.964
0.962
0.96
0.958
0.956
0.954
0.952
0.95
0.948
0.946
0.944
0.942
0.94
0.938
0.936
0.934
0.932
0.93
0.928
0.926
0.924
0.922
0.92
0.918
0.916
0.914
0.912
0.91
0.908
0.906
0.904
0.902
0.9
0.898
0.896
0.894
0.892
0.89
0.888
0.886
0.884
0.882
0.88
0.878
0.876
0.874
0.872
0.87
0.868
0.866
0.864
0.862
0.86
0.858
0.856
0.854
0.852
0.85
0.848
0.846
0.844
0.842
0.84
0.838
0.836
0.834
0.832
0.83
0.828
0.826
0.824
0.822
0.82
0.818
0.816
0.814
0.812
0.81
0.808
0.806
0.804
0.802
0.8
0.798
0.796
0.794
0.792
0.79
0.788
0.786
0.784
0.782
0.78
0.778
0.776
0.774
0.772
0.77
0.768
0.766
0.764
0.762
0.76
0.758
0.756
0.754
0.752
0.75
0.748
0.746
0.744
0.742
0.74
0.738
0.736
0.734
0.732
0.73
0.728
0.726
0.724
0.722
0.72
0.718
0.716
0.714
0.712
0.71
0.708
0.706
0.704
0.702
0.7
0.698
0.696
0.694
0.692
0.69
0.688
0.686
0.684
0.682
0.68
0.678
0.676
0.674
0.672
0.67
0.668
0.666
0.664
0.662
0.66
0.658
0.656
0.654
0.652
0.65
0.648
0.646
0.644
0.642
0.64
0.638
0.636
0.634
0.632
0.63
0.628
0.626
0.624
0.622
0.62
0.618
0.616
0.614
0.612
0.61
0.608
0.606
0.604
0.602
0.6
0.598
0.596
0.594
0.592
0.59
0.588
0.586
0.584
0.582
0.58
0.578
0.576
0.574
0.572
0.57
0.568
0.566
0.564
0.562
0.56
0.558
0.556
0.554
0.552
0.55
0.548
0.546
0.544
0.542
0.54
0.538
0.536
0.534
0.532
0.53
0.528
0.526
0.524
0.522
0.52
0.518
0.516
0.514
0.512
0.51
0.508
0.506
0.504
0.502
0.5
0.498
0.496
0.494
0.492
0.49
0.488
0.486
0.484
0.482
0.48
0.478
0.476
0.474
0.472
0.47
0.468
0.466
0.464
0.462
0.46
0.458
0.456
0.454
0.452
0.45
0.448
0.446
0.444
0.442
0.44
0.438
0.436
0.434
0.432
0.43
0.428
0.426
0.424
0.422
0.42
0.418
0.416
0.414
0.412
0.41
0.408
0.406
0.404
0.402
0.4
0.398
0.396
0.394
0.392
0.39
0.388
0.386
0.384
0.382
0.38
0.378
0.376
0.374
0.372
0.37
0.368
0.366
0.364
0.362
0.36
0.358
0.356
0.354
0.352
0.35
0.348
0.346
0.344
0.342
0.34
0.338
0.336
0.334
0.332
0.33
0.328
0.326
0.324
0.322
0.32
0.318
0.316
0.314
0.312
0.31
0.308
0.306
0.304
0.302
0.3
0.298
0.296
0.294
0.292
0.29
0.288
0.286
0.284
0.282
0.28
0.278
0.276
0.274
0.272
0.27
0.268
0.266
0.264
0.262
0.26
0.258
0.256
0.254
0.252
0.25
0.248
0.246
0.244
0.242
0.24
0.238
0.236
0.234
0.232
0.23
0.228
0.226
0.224
0.222
0.22
0.218
0.216
0.214
0.212
0.21
0.208
0.206
0.204
0.202
0.2
0.198
0.196
0.194
0.192
0.19
0.188
0.186
0.184
0.182
0.18
0.178
0.176
0.174
0.172
0.17
0.168
0.166
0.164
0.162
0.16
0.158
0.156
0.154
0.152
0.15
0.148
0.146
0.144
0.142
0.14
0.138
0.136
0.134
0.132
0.13
0.128
0.126
0.124
0.122
0.12
0.118
0.116
0.114
0.112
0.11
0.108
0.106
0.104
0.102
0.1
0.098
0.096
0.094
0.092
0.09
0.088
0.086
0.084
0.082
0.08
0.078
0.076
0.074
0.072
0.07
0.068
0.066
0.064
0.062
0.06
0.058
0.056
0.054
0.052
0.05
0.048
0.046
0.044
0.042
0.04
0.038
0.036
0.034
0.032
0.03
0.028
0.026
0.024
0.022
0.02
0.018
0.016
0.014
0.012
0.01
0.008
0.006
0.004
0.002

Figure 17

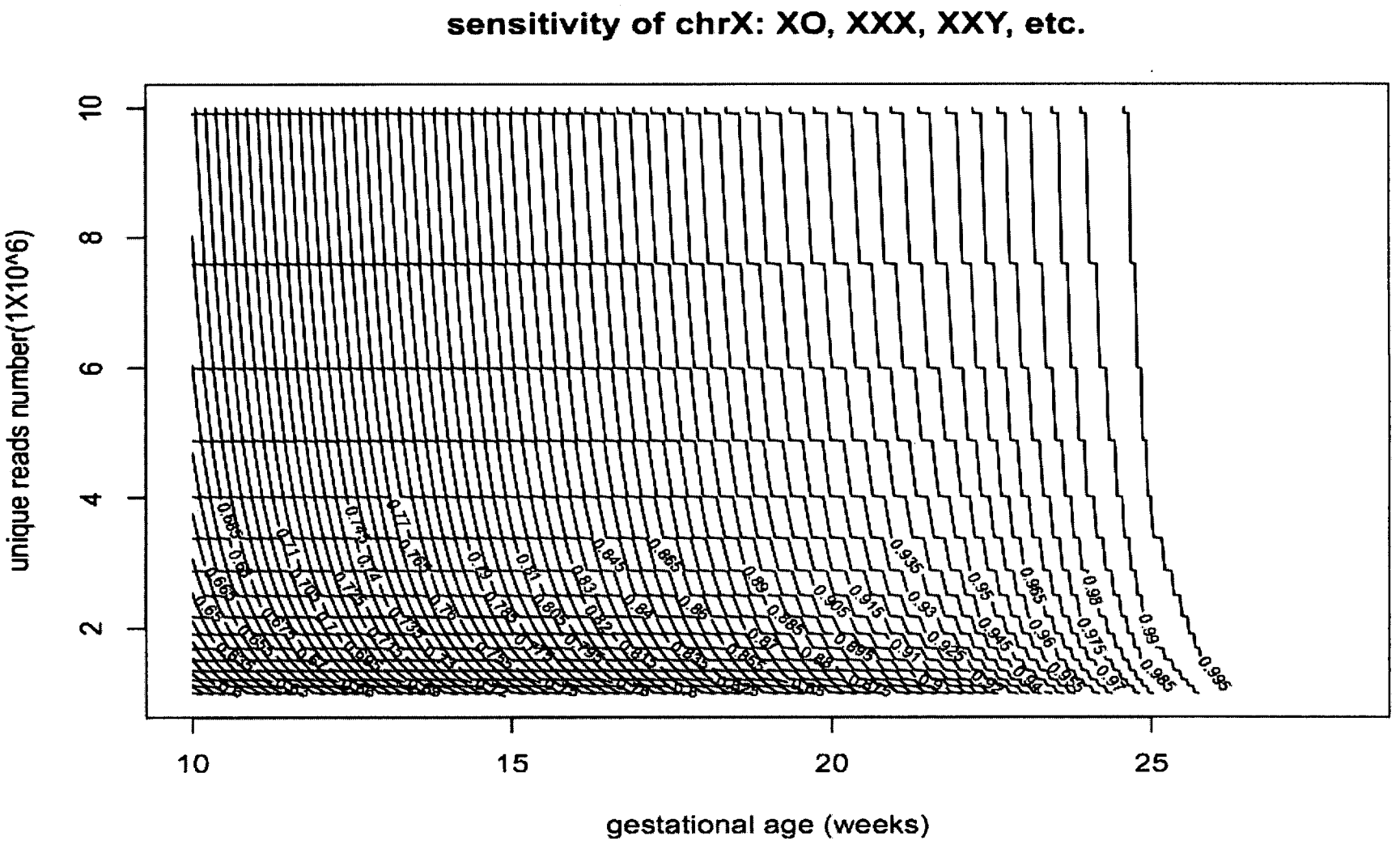
sensitivity of chrX: XO, XXX, XXY, etc.
unique reads number(1X10^6)
2
4
6
8
10
10
15
20
25
gestational age (weeks)

Figure 18

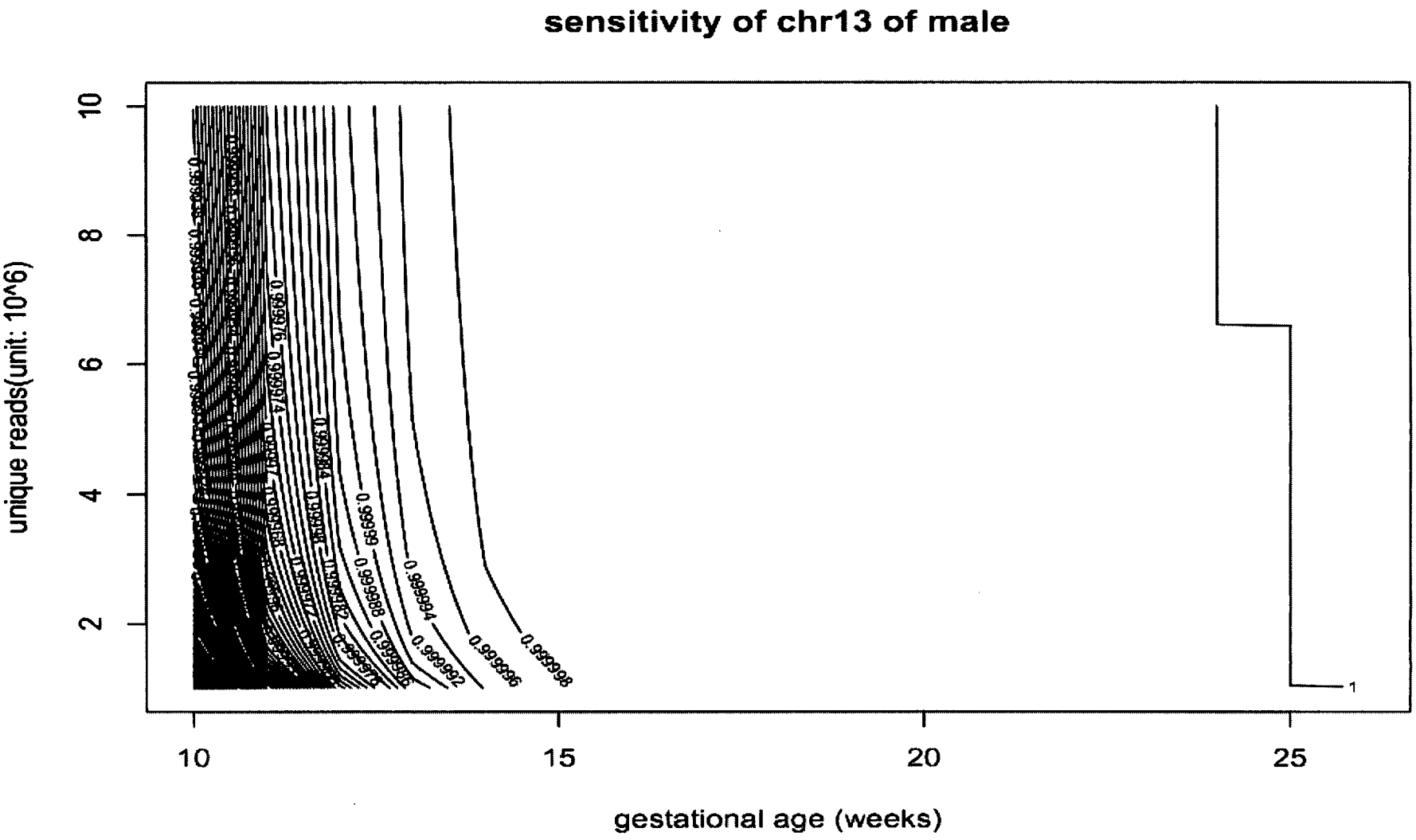
sensitivity of chr13 of male
unique reads(unit: 10^6)
2
4
6
8
10
10
15
20
25
gestational age (weeks)
0.999998
0.999996
0.999992
0.999984
0.99998
0.99996
0.99992
0.99984
0.9998
0.9996
0.9992
0.9984
0.998
0.996
0.992
0.984
0.98
0.976
0.96
0.95
0.94
0.92
0.9
0.88
0.86
0.84
0.82
0.8
0.78
0.76
0.74
0.72
0.7
0.68
0.66
0.64
0.62
0.6
0.58
0.56
0.54
0.52
0.5
0.48
0.46
0.44
0.42
0.4
0.38
0.36
0.34
0.32
0.3
0.28
0.26
0.24
0.22
0.2
0.18
0.16
0.14
0.12
0.1
0.08
0.06
0.04
0.02
1

Figure 19

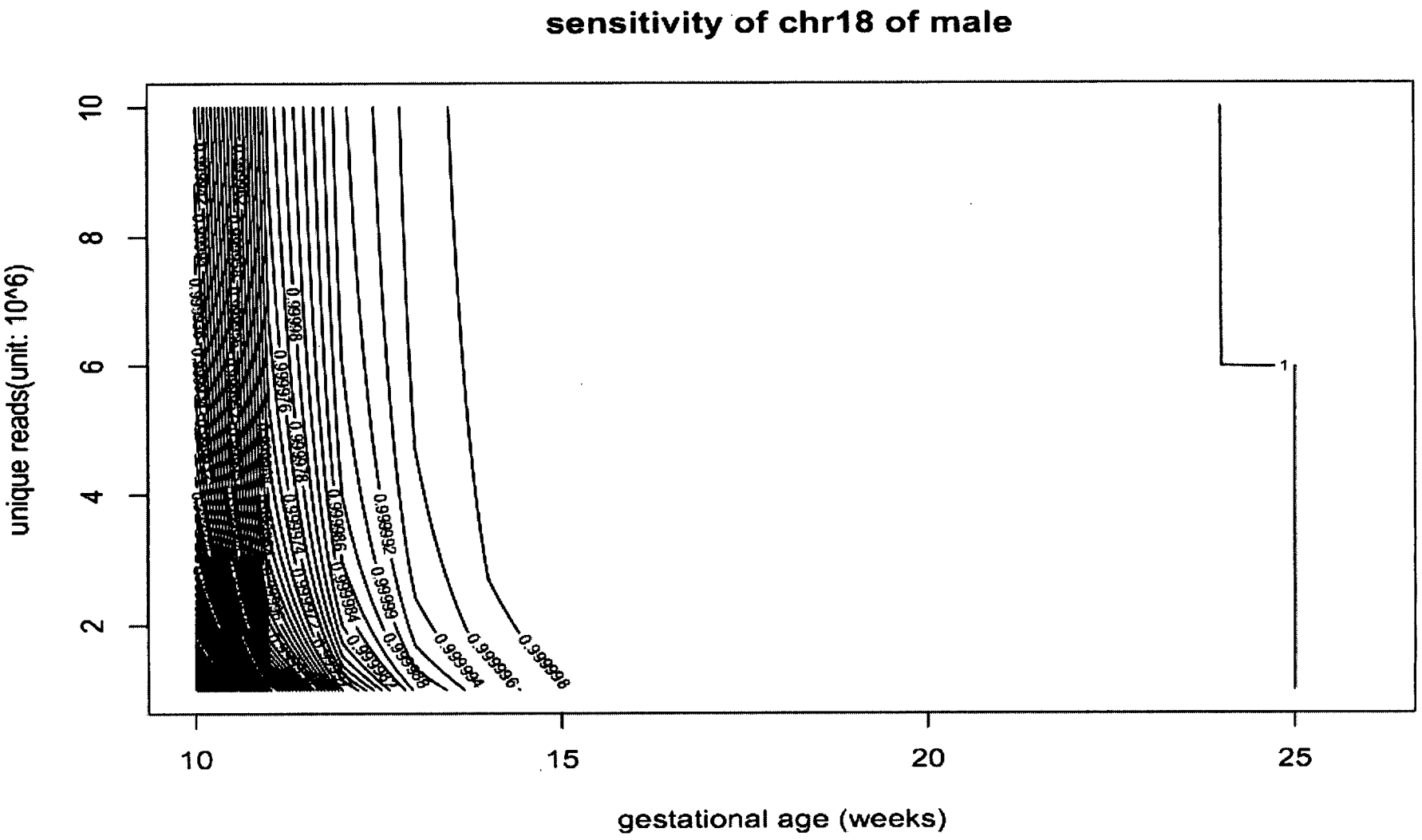
sensitivity of chr18 of male
unique reads(unit: 10^6)
2
4
6
8
10
10
15
20
25
gestational age (weeks)
0.999998
0.999996
0.999994
0.99999
0.99998
1

Figure 20

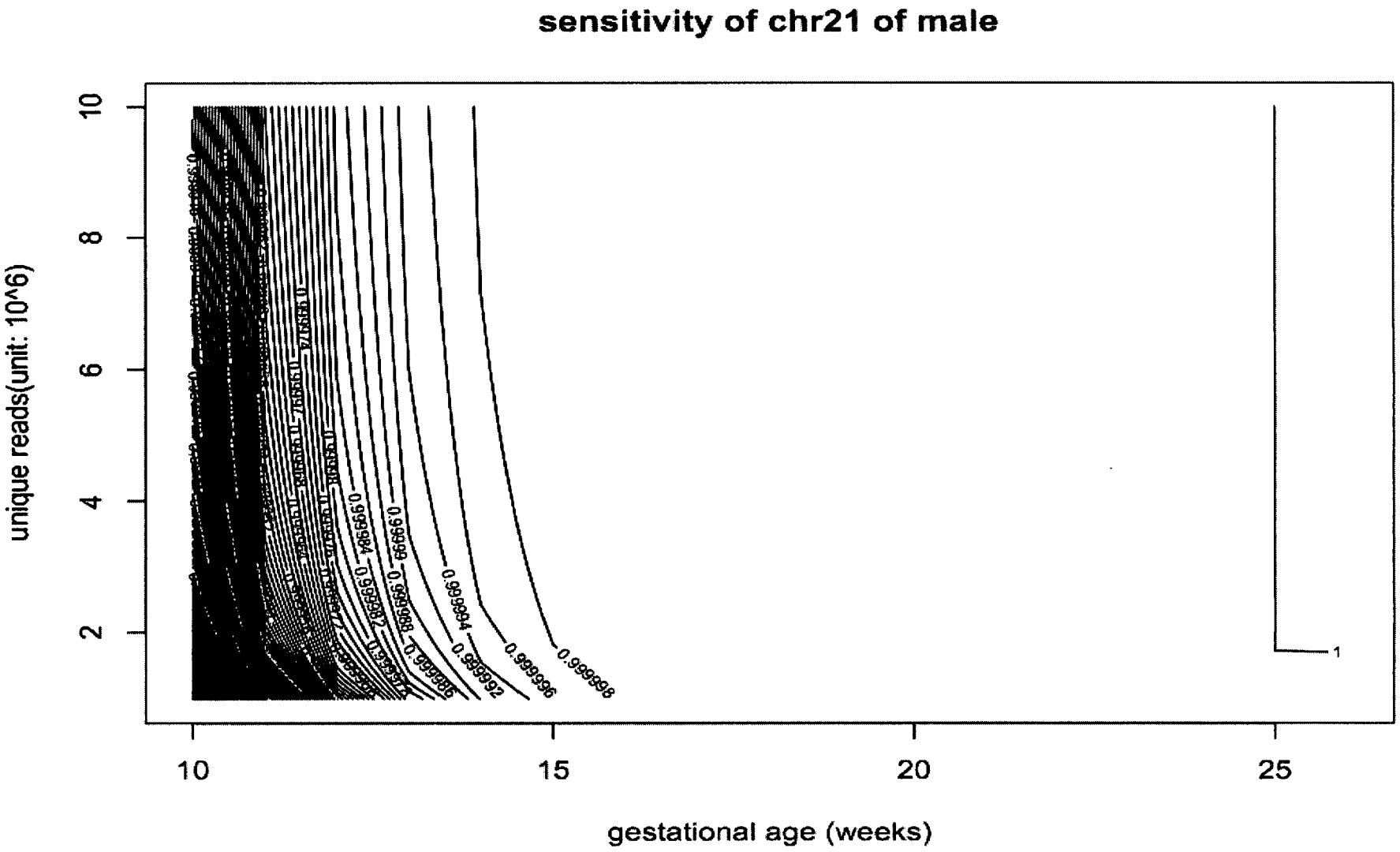
sensitivity of chr21 of male
unique reads(unit: 10^6)
2
4
6
8
10
10
15
20
25
gestational age (weeks)
0.999998
0.999996
0.999992
0.999984
0.999968
0.999936
0.999872
1

Figure 21