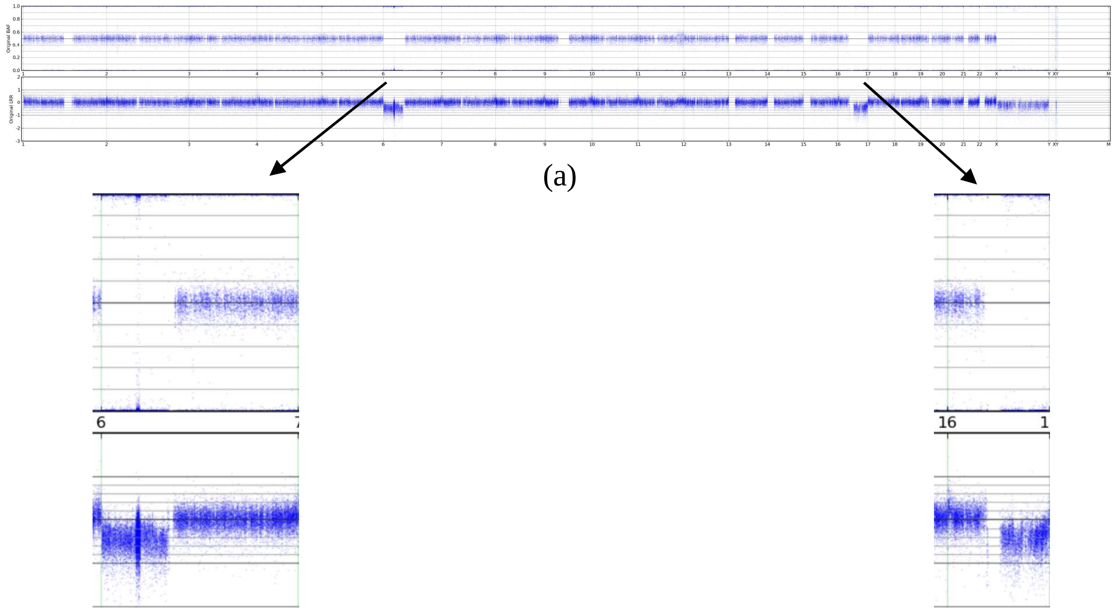
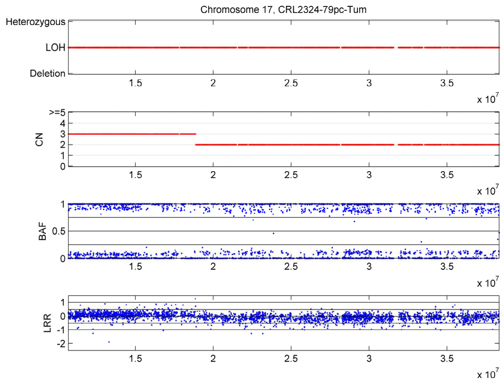
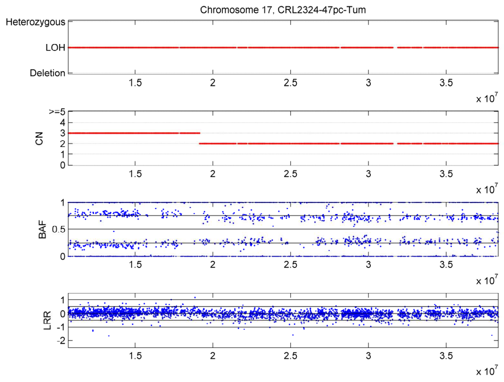


(a) (b) (c)

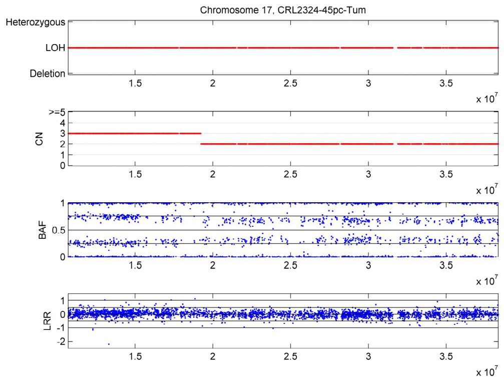
Figure S1. Illustration of the genotyping signals of normal cellline CRL-2325D. (a) Plot of the whole genomic data (b) Deletion region identified on chromosome 6 (c) Deletion region identified on chromosome 16.


Chromosome 17, CRL2324-79pc-Tum
Heterozygous
LOH
Deletion
CN
>=5
BAF
LRR
1.5
2
2.5
3
3.5
x 10^7

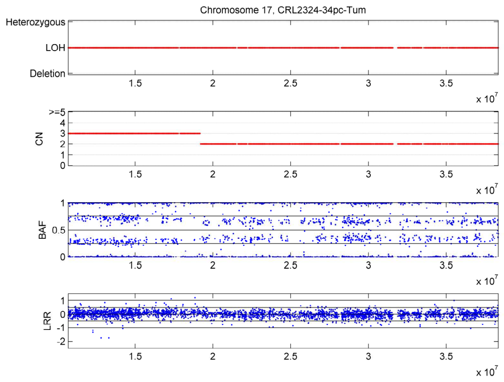
(a)


Chromosome 17, CRL2324-47pc-Tum
Heterozygous
LOH
Deletion
CN
>=5
BAF
LRR
1.5
2
2.5
3
3.5
x 10^7

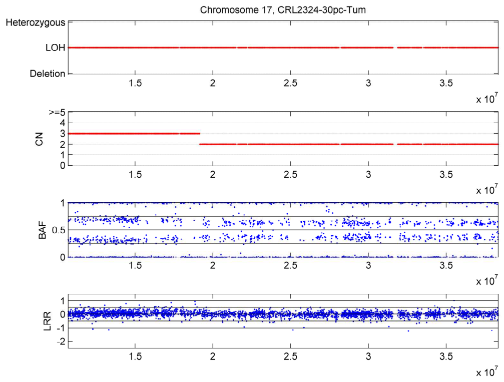
(b)


Chromosome 17, CRL2324-45pc-Tum
Heterozygous
LOH
Deletion
CN
>=5
BAF
LRR
1.5
2
2.5
3
3.5
x 10^7

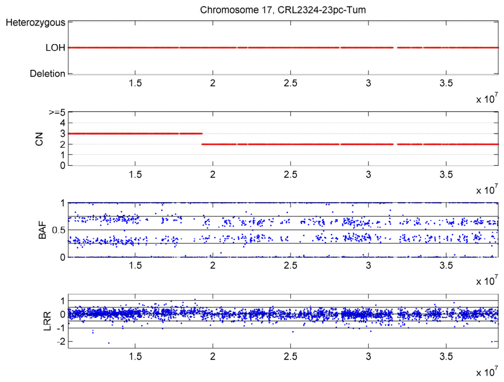
(c)


Chromosome 17, CRL2324-34pc-Tum
Heterozygous
LOH
Deletion
CN
>=5
BAF
LRR
1.5
2
2.5
3
3.5
x 10^7

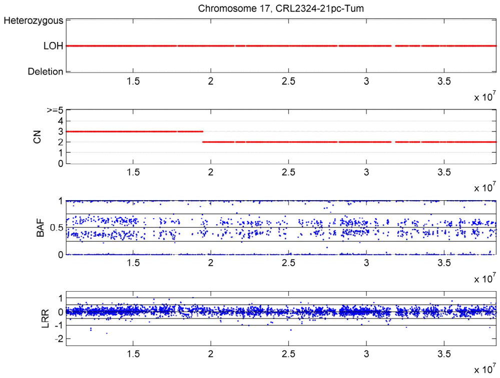
(d)


Chromosome 17, CRL2324-30pc-Tum
Heterozygous
LOH
Deletion
CN
>=5
BAF
LRR
1.5
2
2.5
3
3.5
x 10^7

(e)

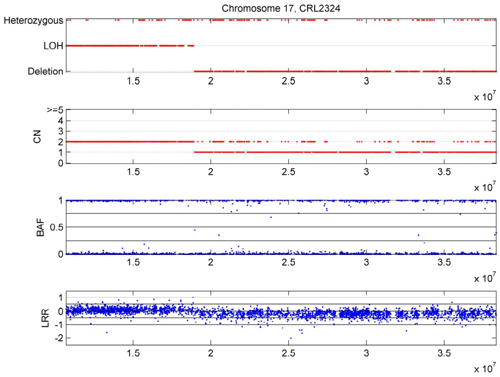

Chromosome 17, CRL2324-23pc-Tum
Heterozygous
LOH
Deletion
CN
>=5
BAF
LRR
1.5
2
2.5
3
3.5
x 10^7

(f)

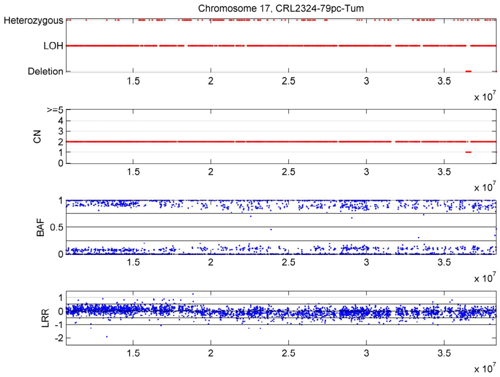


(g)

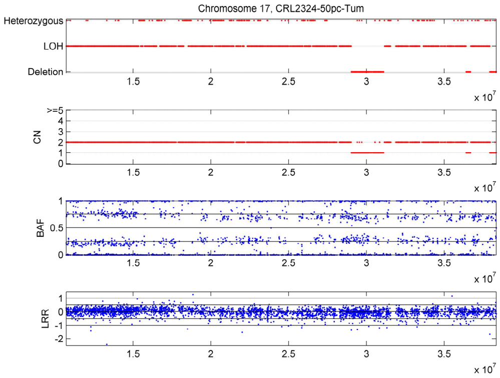
Figure S2. Plots of two adjacent LOH regions on chromosome 17 and GPHMM results for dilution series data. (a) Plot of sample “CRL2324-79pc-Tum” (b) Plot of sample “CRL2324-47pc-Tum” (c) Plot of sample “CRL2324-45pc-Tum” (d) Plot of sample “CRL2324-34pc-Tum” (e) Plot of sample “CRL2324-30pc-Tum” (f) Plot of sample “CRL2324-23pc-Tum” (g) Plot of sample “CRL2324-21pc-Tum”


Chromosome 17, CRL2324
Heterozygous
LOH
Deletion
CN
>=5
BAF
LRR
1.5
2
2.5
3
3.5
x 10^7

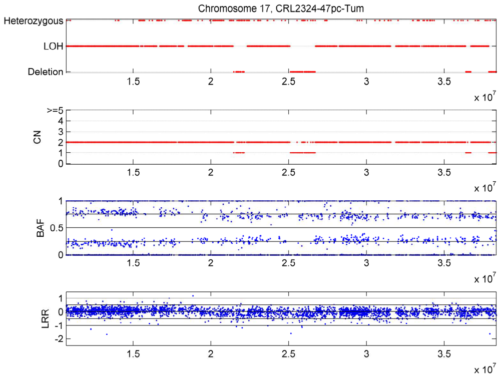
(a)


Chromosome 17, CRL2324-79pc-Tum
Heterozygous
LOH
Deletion
CN
>=5
BAF
LRR
1.5
2
2.5
3
3.5
x 10^7

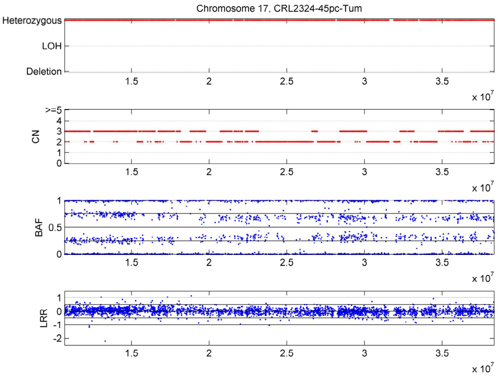
(b)


Chromosome 17, CRL2324-50pc-Tum
Heterozygous
LOH
Deletion
CN
>=5
BAF
LRR
1.5
2
2.5
3
3.5
x 10^7

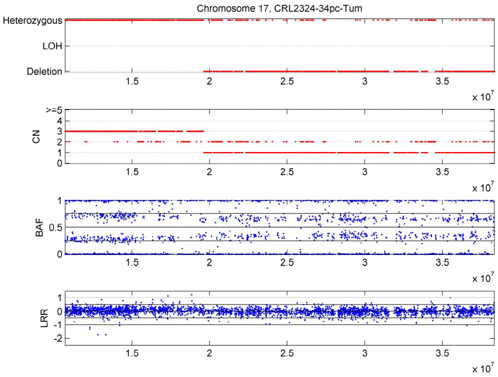
(c)


Chromosome 17, CRL2324-47pc-Tum
Heterozygous
LOH
Deletion
CN
>=5
BAF
LRR
1.5
2
2.5
3
3.5
x 10^7

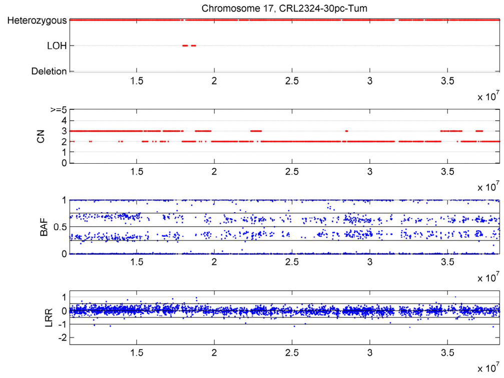
(d)


Chromosome 17, CRL2324-45pc-Tum
Heterozygous
LOH
Deletion
CN
>=5
BAF
LRR
1.5
2
2.5
3
3.5
x 10^7

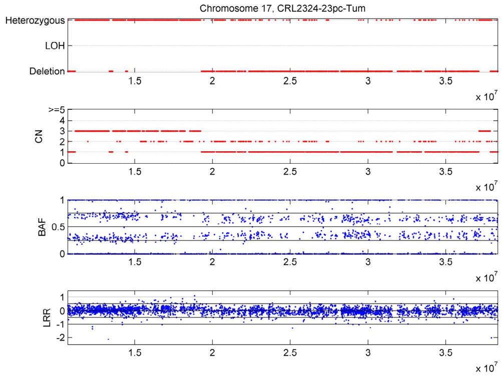
(e)


Chromosome 17, CRL2324-34pc-Tum
Heterozygous
LOH
Deletion
CN
>=5
BAF
LRR
1.5
2
2.5
3
3.5
x 10^7

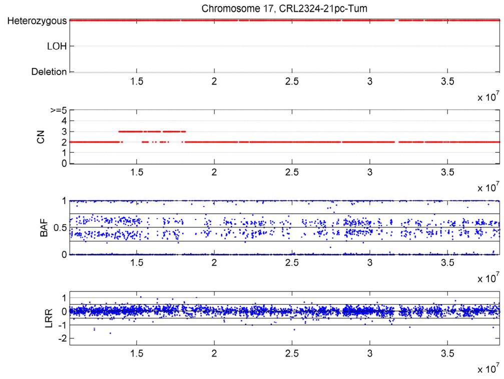
(f)



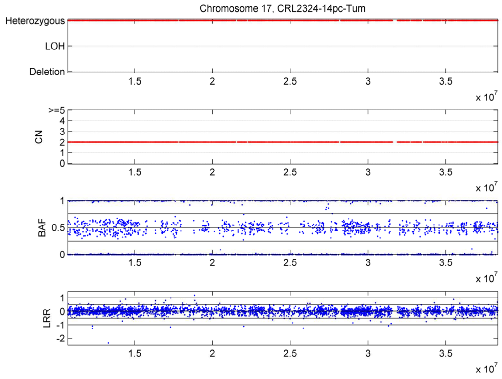
(g)



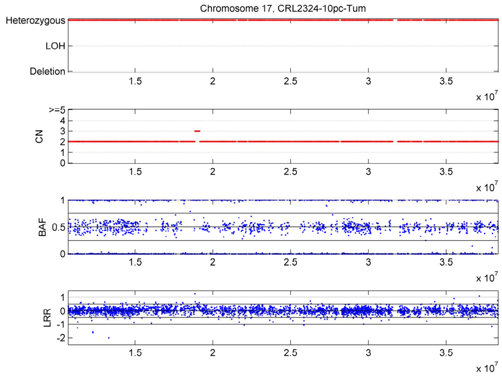
(h)



(i)

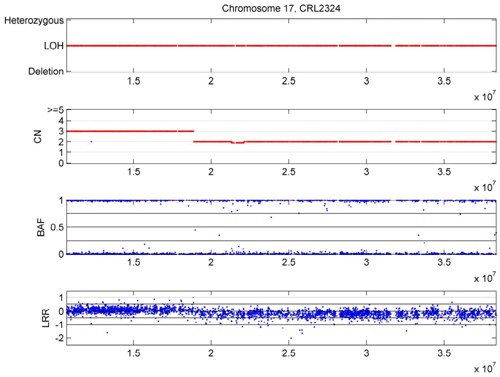


(j)

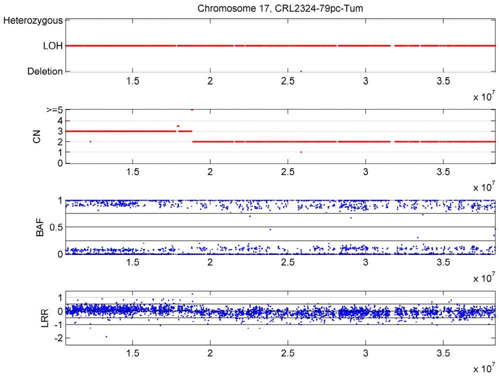


(k)

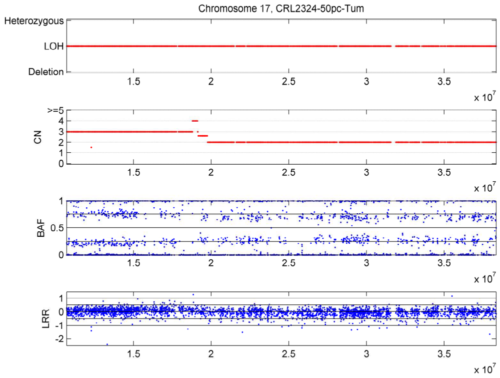
Figure S3. Plots of two adjacent LOH regions on chromosome 17 and genoCNA results for dilution series data. (a) Plot of sample "CRL2324" (b) Plot of sample "CRL2324-79pc-Tum" (c) Plot of sample "CRL2324-50pc-Tum" (d) Plot of sample "CRL2324-47pc-Tum" (e) Plot of sample "CRL2324-45pc-Tum" (f) Plot of sample "CRL2324-34pc-Tum" (g) Plot of sample "CRL2324-30pc-Tum" (h) Plot of sample "CRL2324-23pc-Tum" (i) Plot of sample "CRL2324-21pc-Tum" (j) Plot of sample "CRL2324-14pc-Tum" (k) Plot of sample "CRL2324-10pc-Tum"


Chromosome 17, CRL2324
Heterozygous
LOH
Deletion
CN
BAF
LRR
1.5
2
2.5
3
3.5
x 10^7

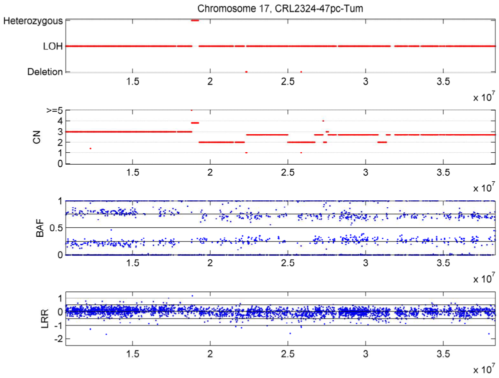
(a)


Chromosome 17, CRL2324-79pc-Tum
Heterozygous
LOH
Deletion
CN
BAF
LRR
x 10^7

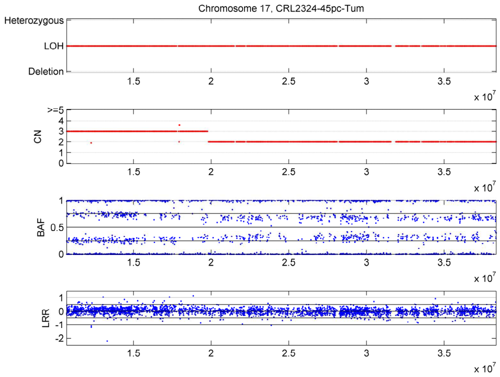
(b)


Chromosome 17, CRL2324-50pc-Tum
Heterozygous
LOH
Deletion
CN
BAF
LRR
x 10^7

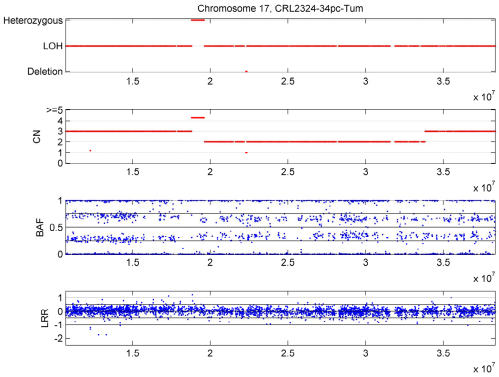
(c)


Chromosome 17, CRL2324-47pc-Tum
Heterozygous
LOH
Deletion
CN
BAF
LRR
x 10^7

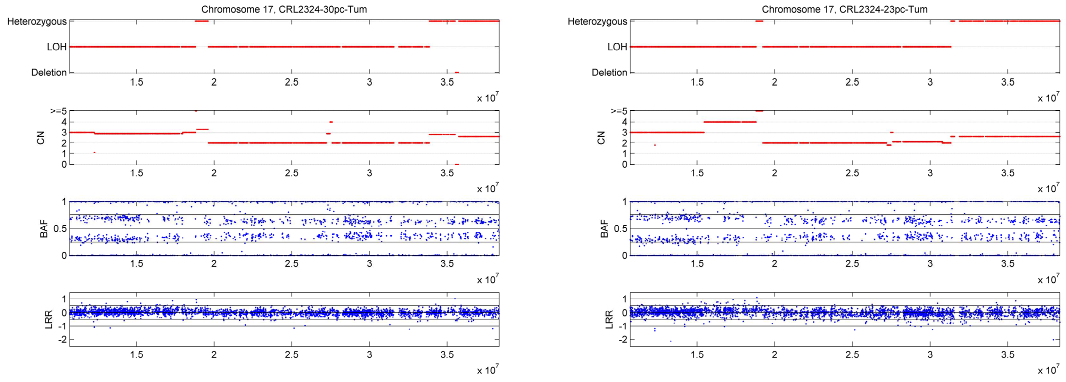
(d)


Chromosome 17, CRL2324-45pc-Tum
Heterozygous
LOH
Deletion
CN
BAF
LRR
x 10^7

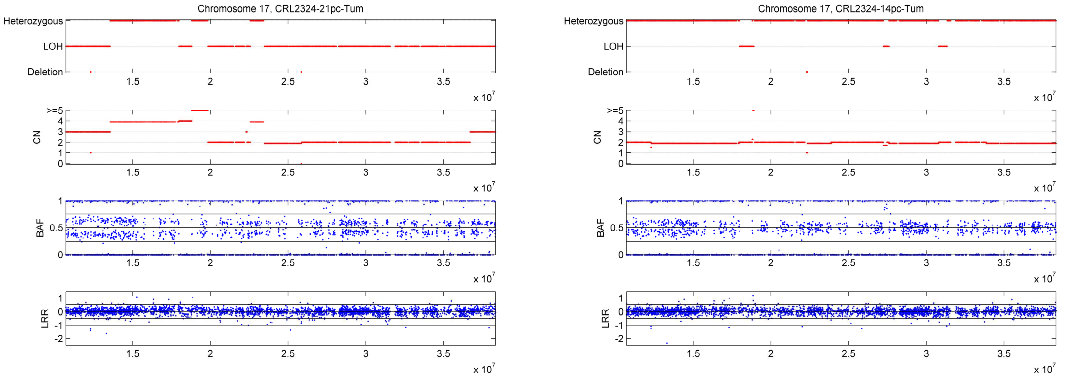
(e)


Chromosome 17, CRL2324-34pc-Tum
Heterozygous
LOH
Deletion
CN
BAF
LRR
x 10^7

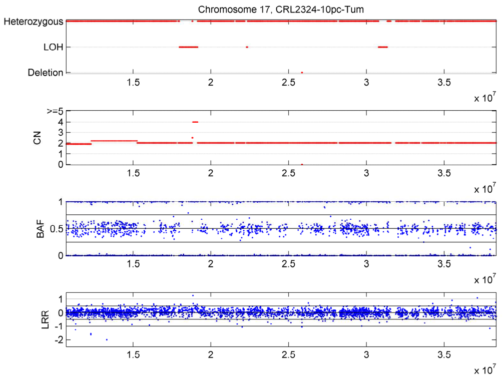
(f)



(g) (h)

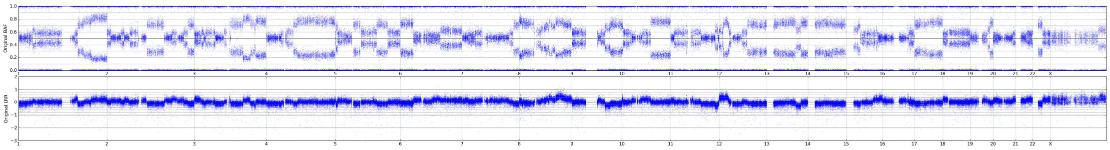


(i) (j)

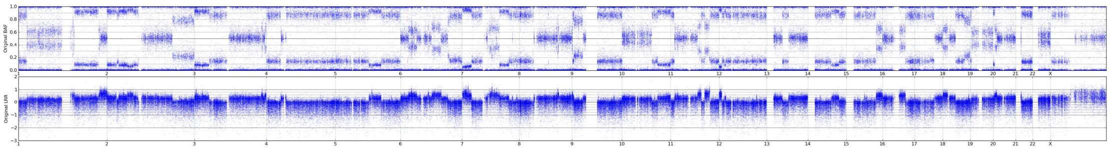


(k)

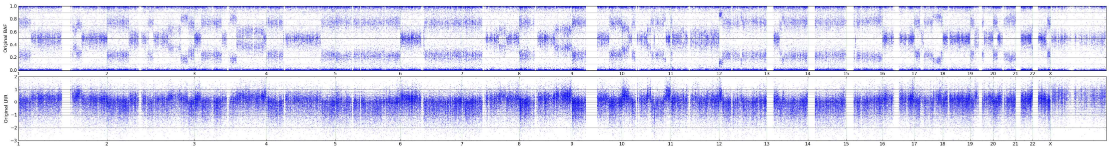
Figure S4. Plots of two adjacent LOH regions on chromosome 17 and GAP results for dilution series data. (a) Plot of sample “CRL2324” (b) Plot of sample “CRL2324-79pc-Tum” (c) Plot of sample “CRL2324-50pc-Tum” (d) Plot of sample “CRL2324-47pc-Tum” (e) Plot of sample “CRL2324-45pc-Tum” (f) Plot of sample “CRL2324-34pc-Tum” (g) Plot of sample “CRL2324-30pc-Tum” (h) Plot of sample “CRL2324-23pc-Tum” (i) Plot of sample “CRL2324-21pc-Tum” (j) Plot of sample “CRL2324-14pc-Tum” (k) Plot of sample “CRL2324-10pc-Tum”



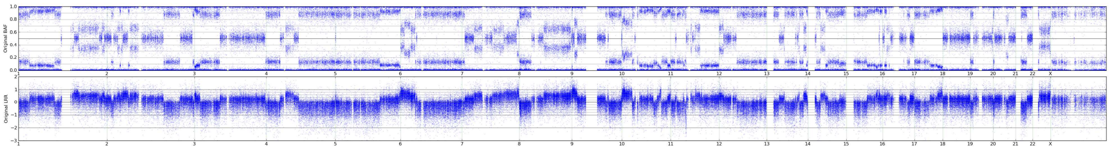
BLC_B1_T19



BLC_B1_T14



BLC_B1_T17



BLC_B1_T22

Figure S5. Plots of whole genome genotyping signals of different samples in GAP data

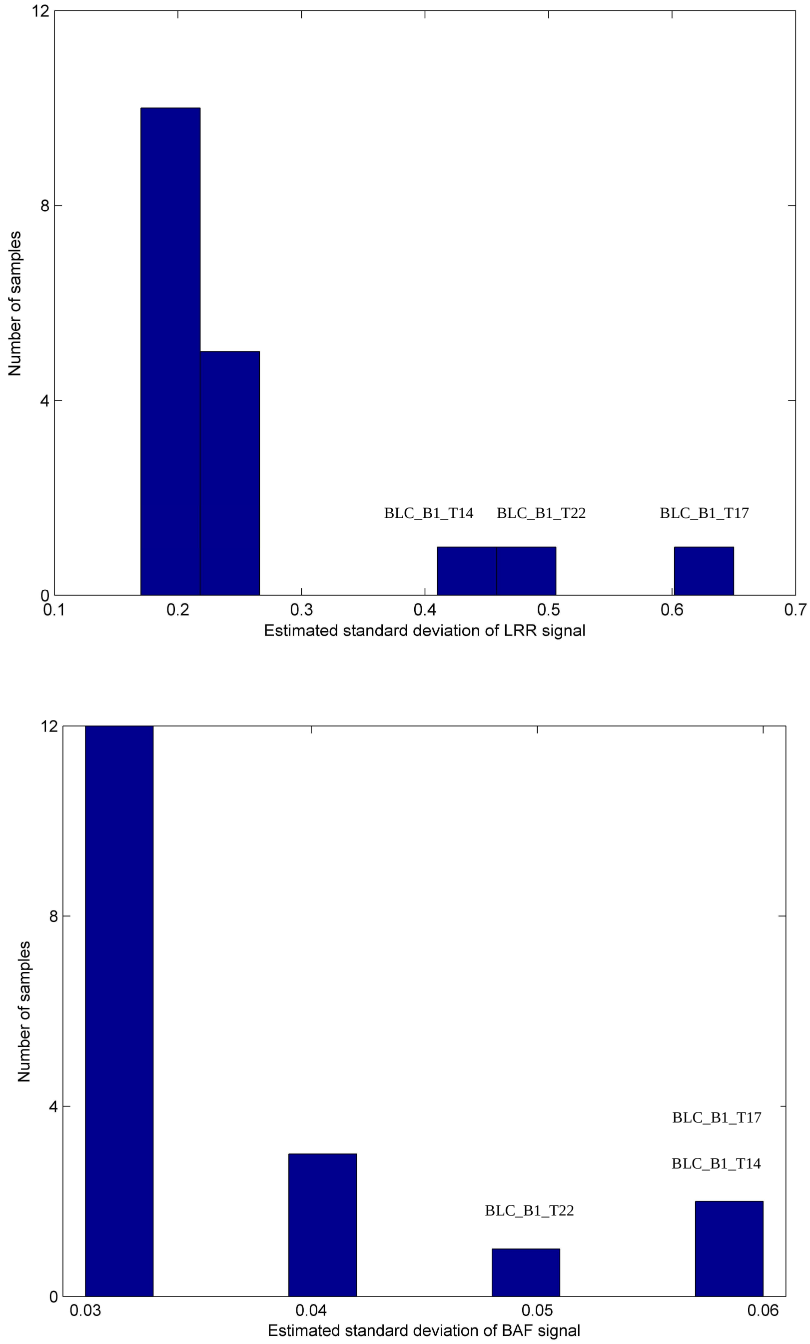
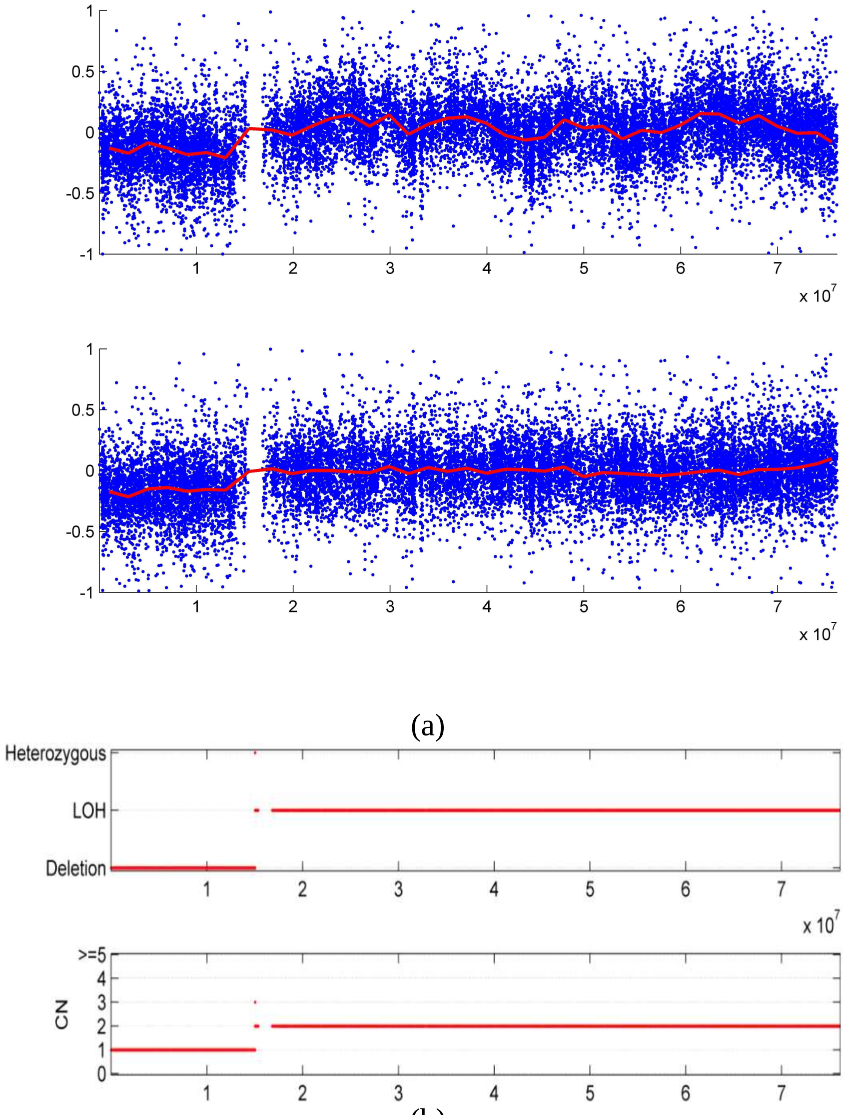


Figure S6. Histograms of global parameters σ_l and σ_b estimated from GAP data. Samples with abnormal values are illustrated.



(a)

(b)

Figure S7. Genomic waves in one tumor sample (YBCID: 120) of the HER2-positive breast cancer data. (a) Genome waves observed in raw LRR signals (top) are significantly reduced in the filtered LRR signals (bottom) by applying a linear model of GC content with coefficient estimated from GPHMM. (b) The corresponding results of GPHMM for this chromosomal region.

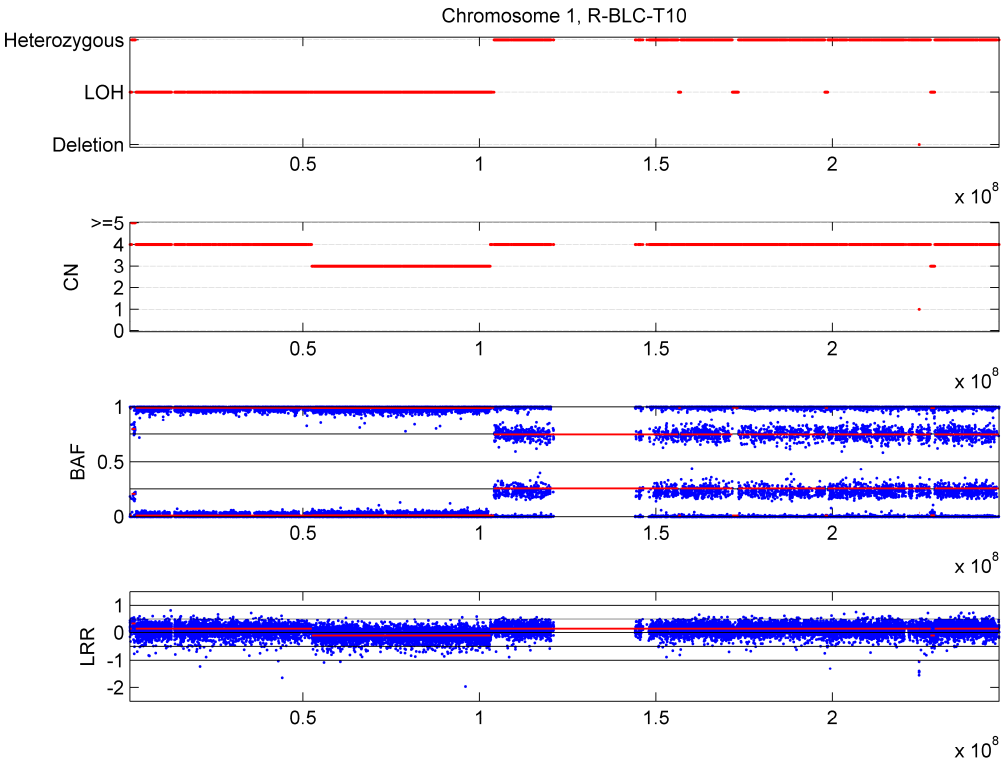


Figure S8. LRR/BAF signals and the prediction results of chromosome 1 of GAP sample "BLC_T10". The BAF signals of two LOH regions on the left have smaller variances than those of the heterozygous regions on the right and are very close to the boundaries, suggesting some of them are probably truncated to 0 or 1, respectively. But apparently the estimated LRR/BAF mean values (red lines in the two panels at the bottom) are consistent with the real signal distributions.