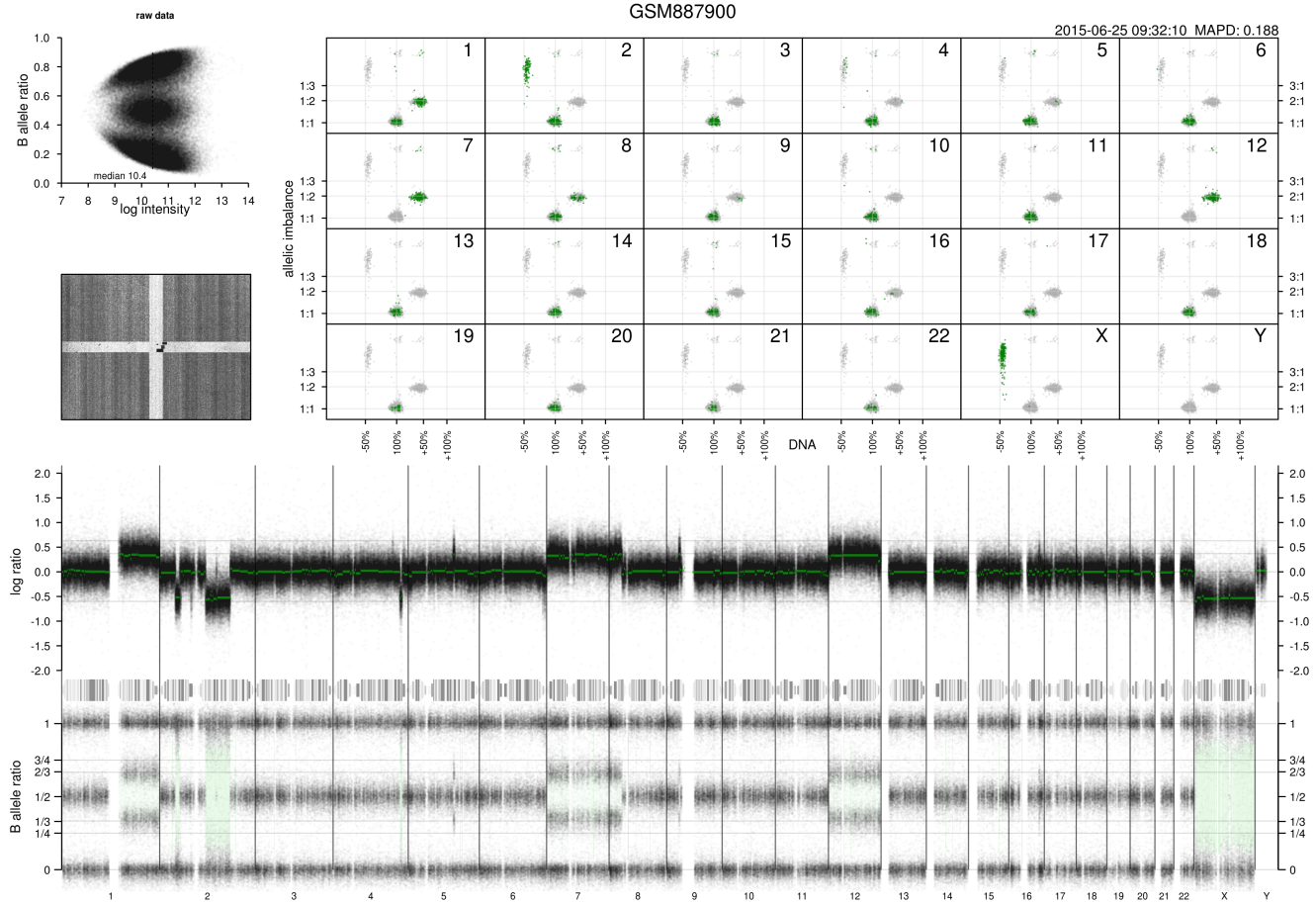
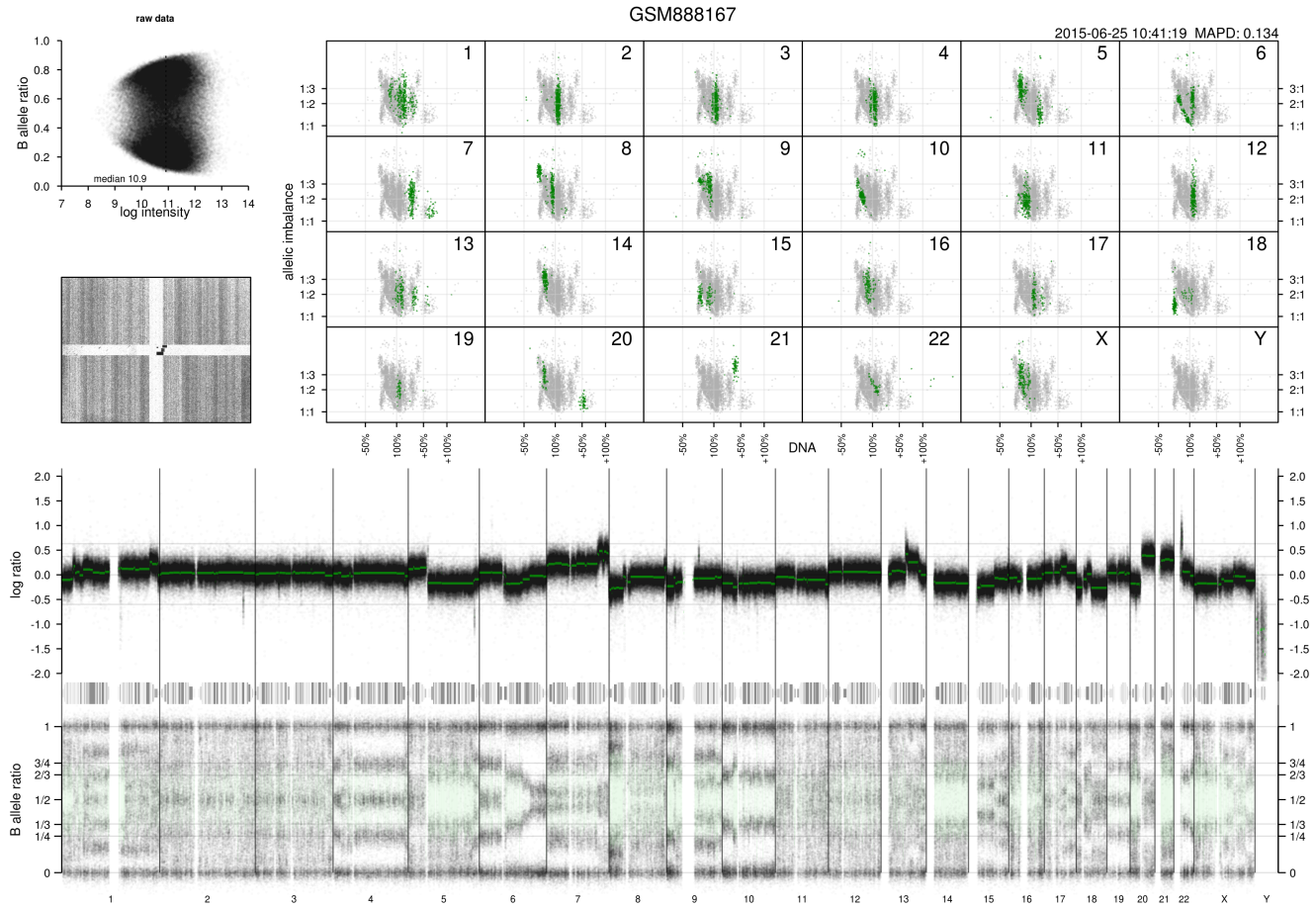
Cancer genome examples

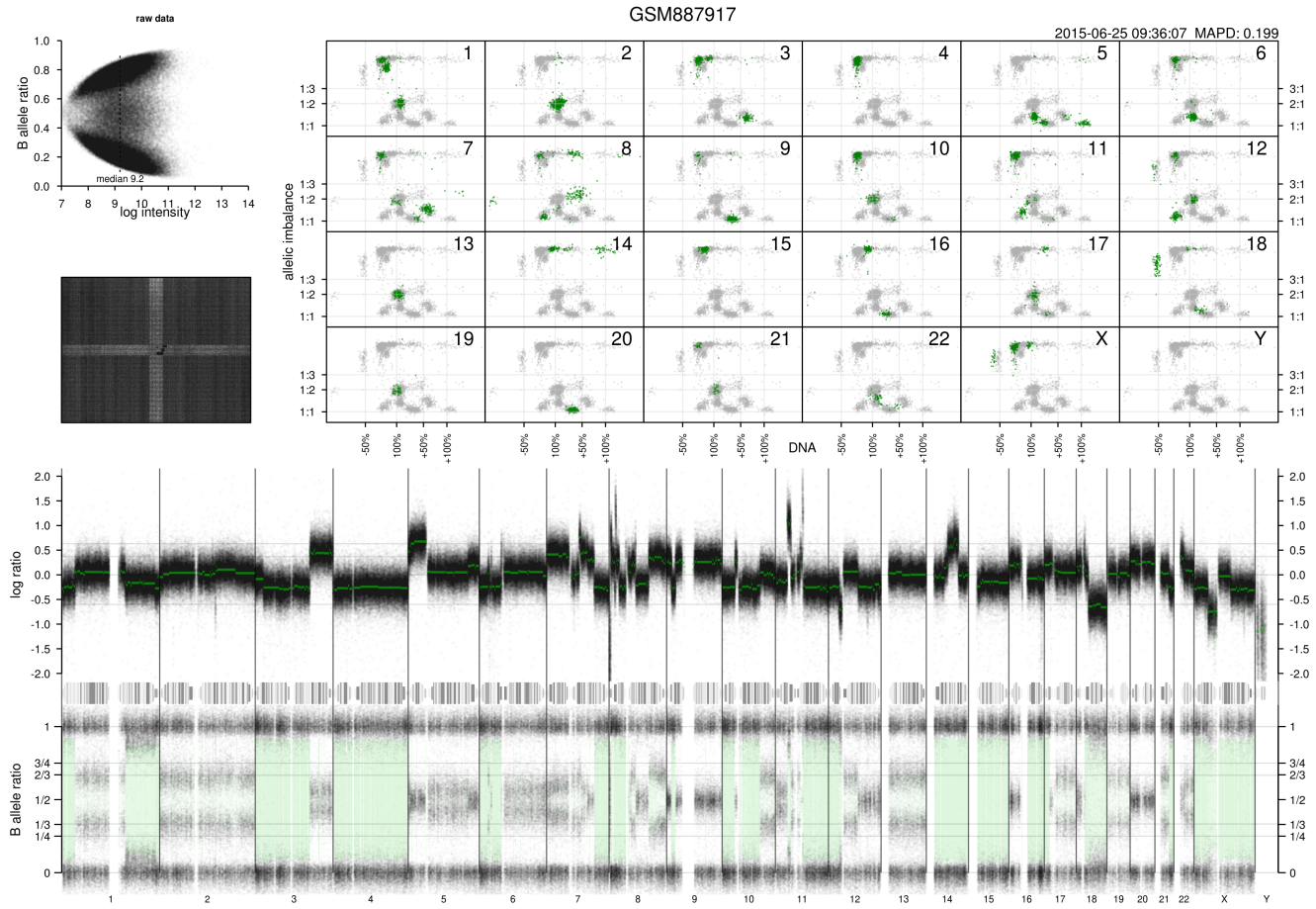
Whole-genome plots of samples analyzed with Rawcopy



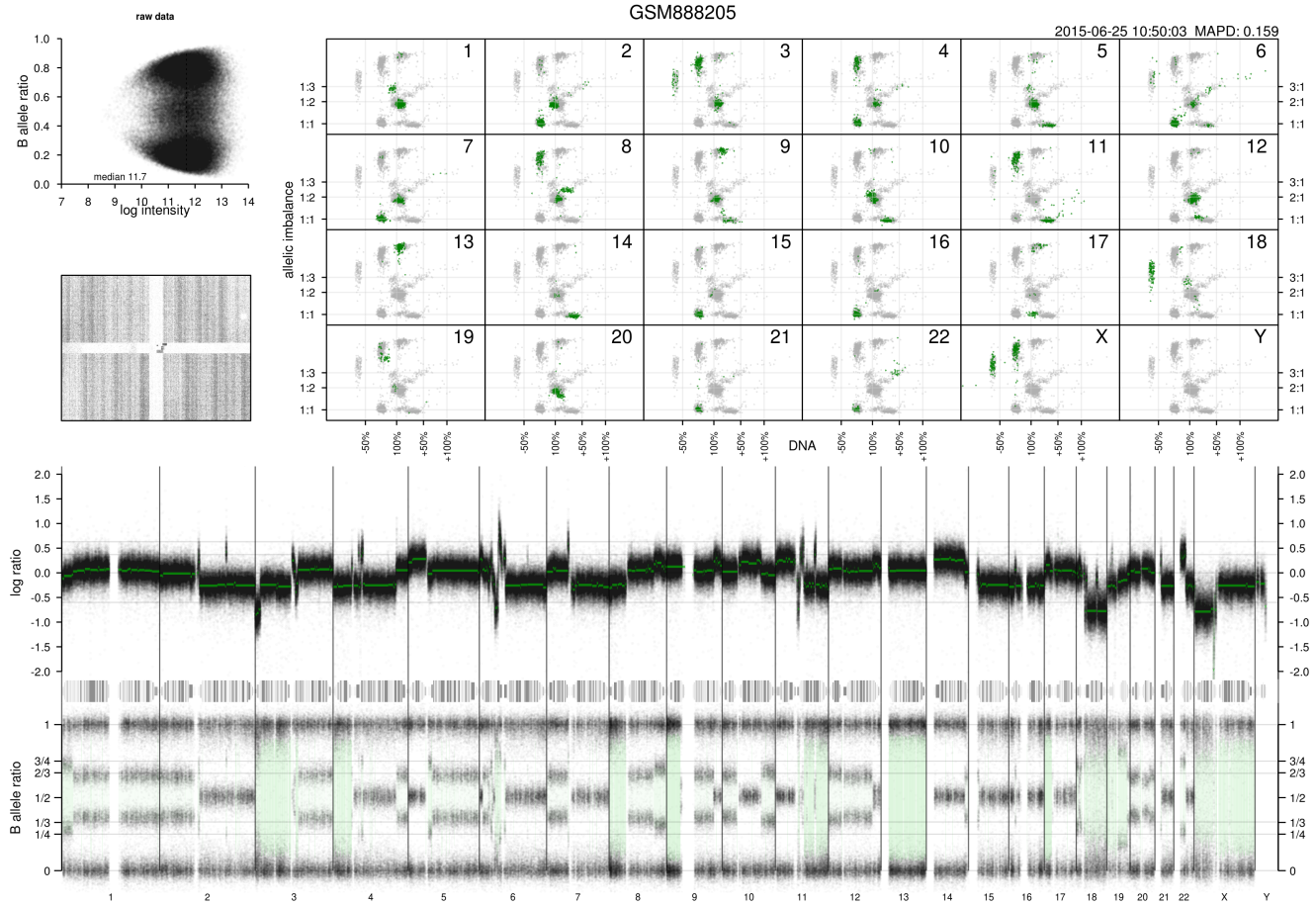
Example 1. Cancer cell line, near diploid on average, good technical quality. In the chromosome-wise scatter plots, the X chromosome and the deletion on chromosome 2 appear at -50% DNA abundance and high allelic imbalance, consistent with a single remaining copy in a sample where the average amount of DNA abundance along the genome corresponds to two copies per cell. The duplications on 1q, 7 and 8 appear at near 50% increased DNA abundance and a 1:2 allelic imbalance, consistent with 3 copies (1 and 2 of the parental homologs) in each cell.



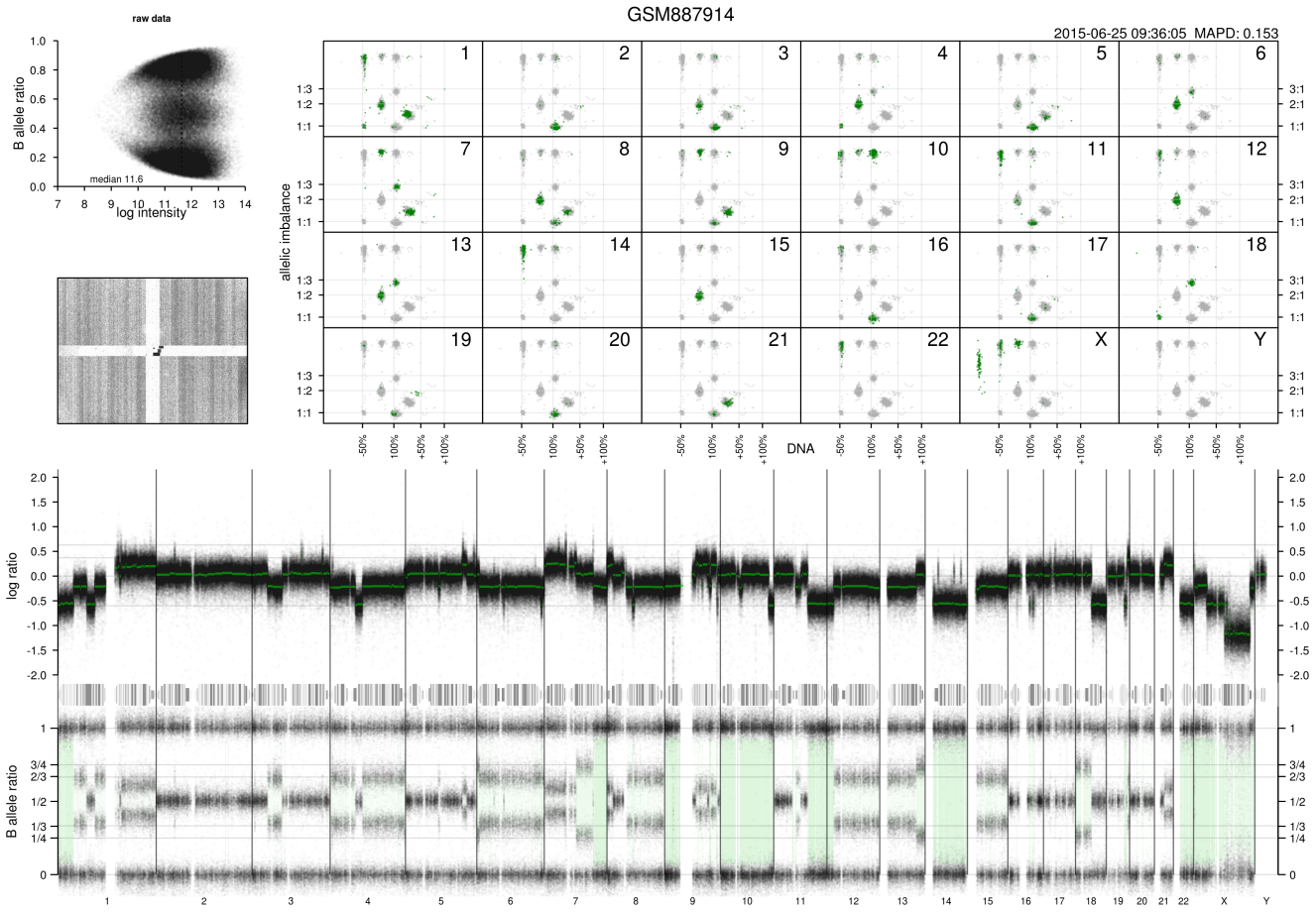
Example 2. Cancer cell line with substantial DNA contamination. This is indicated by multiple bands in the bottom track due to non-matching genotypes in the two involved patients. For example, the five-band pattern on chromosome 4 is the likely outcome of equal DNA mixing and normal diploid copy number of chromosome 4 for both cell populations.



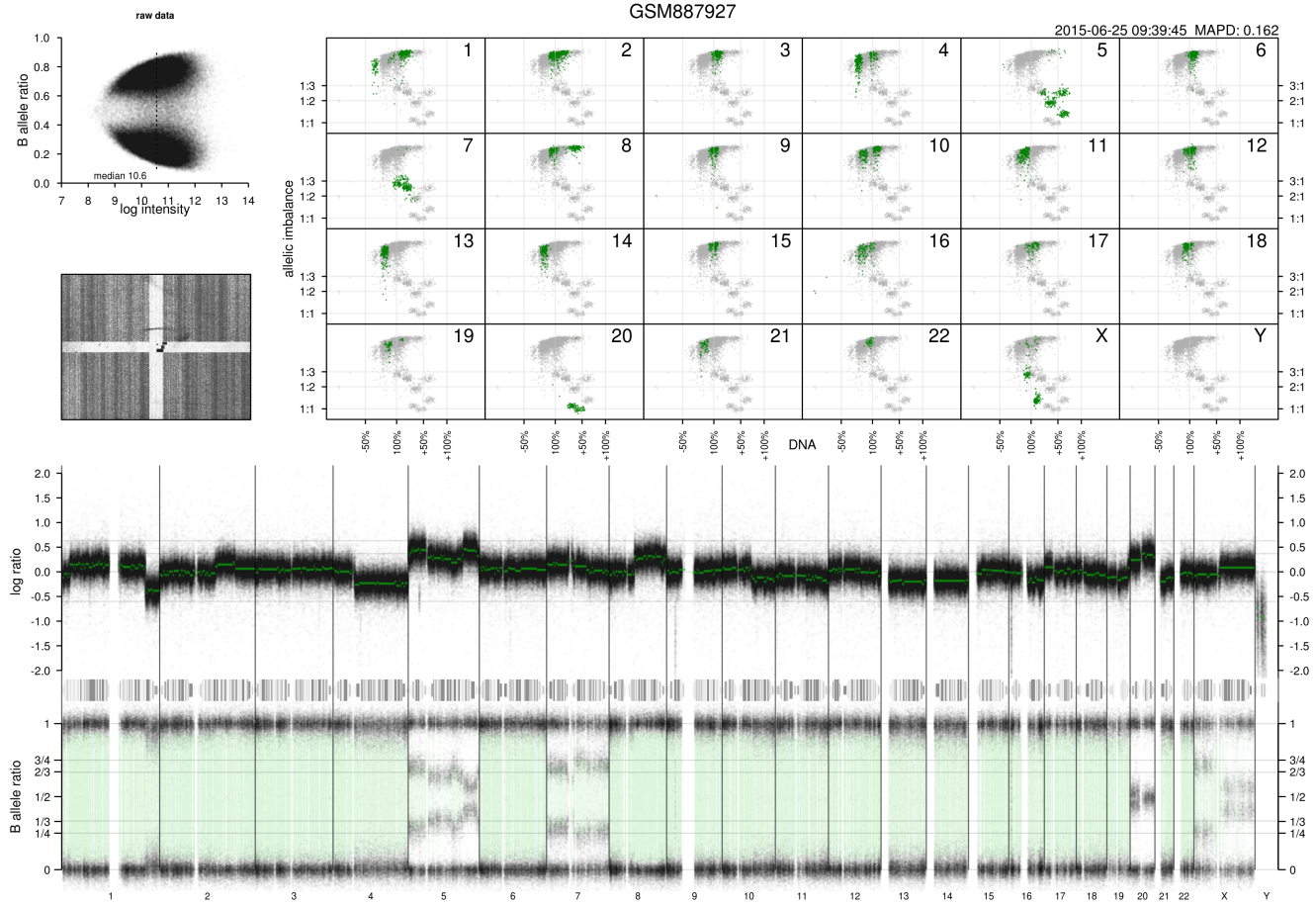
Example 3. Cancer cell line with unusually low hybridization to the array. Overall quality remains good but SNP allele frequency quality (bottom track) is affected. The scatter plots of DNA abundance versus allelic imbalance indicate an average copy number per cell near 3. There are 3 copies (2+1 of the parental homologs) of chromosome 2, 4 copies (2+2) och chromosome 20 and 2 copies (2+0, loss of heterozygosity) of chromosome 4, per cell.



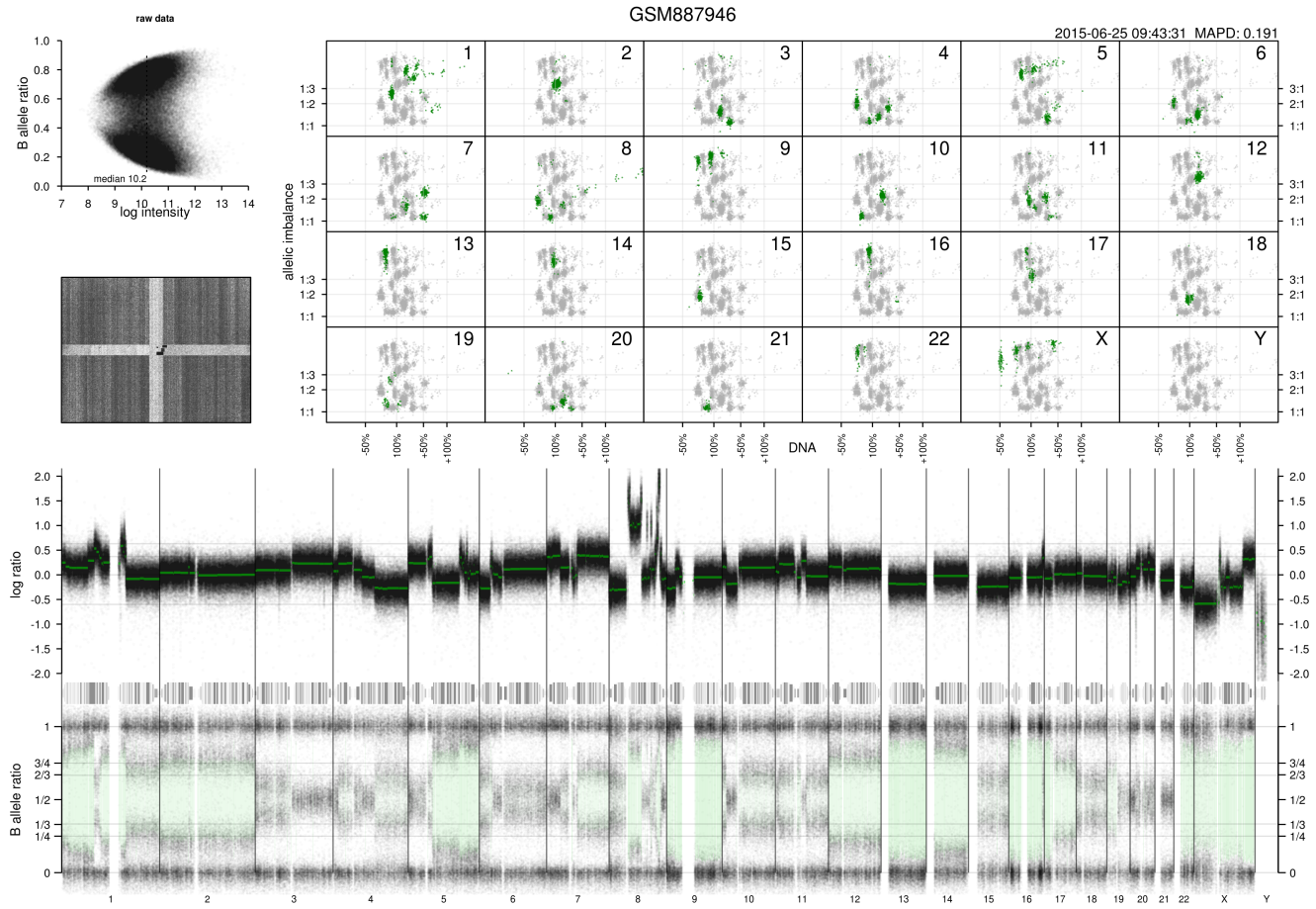
Example 4. Cancer cell line with unusually high hybridization to the array. Average copy number is near 3.



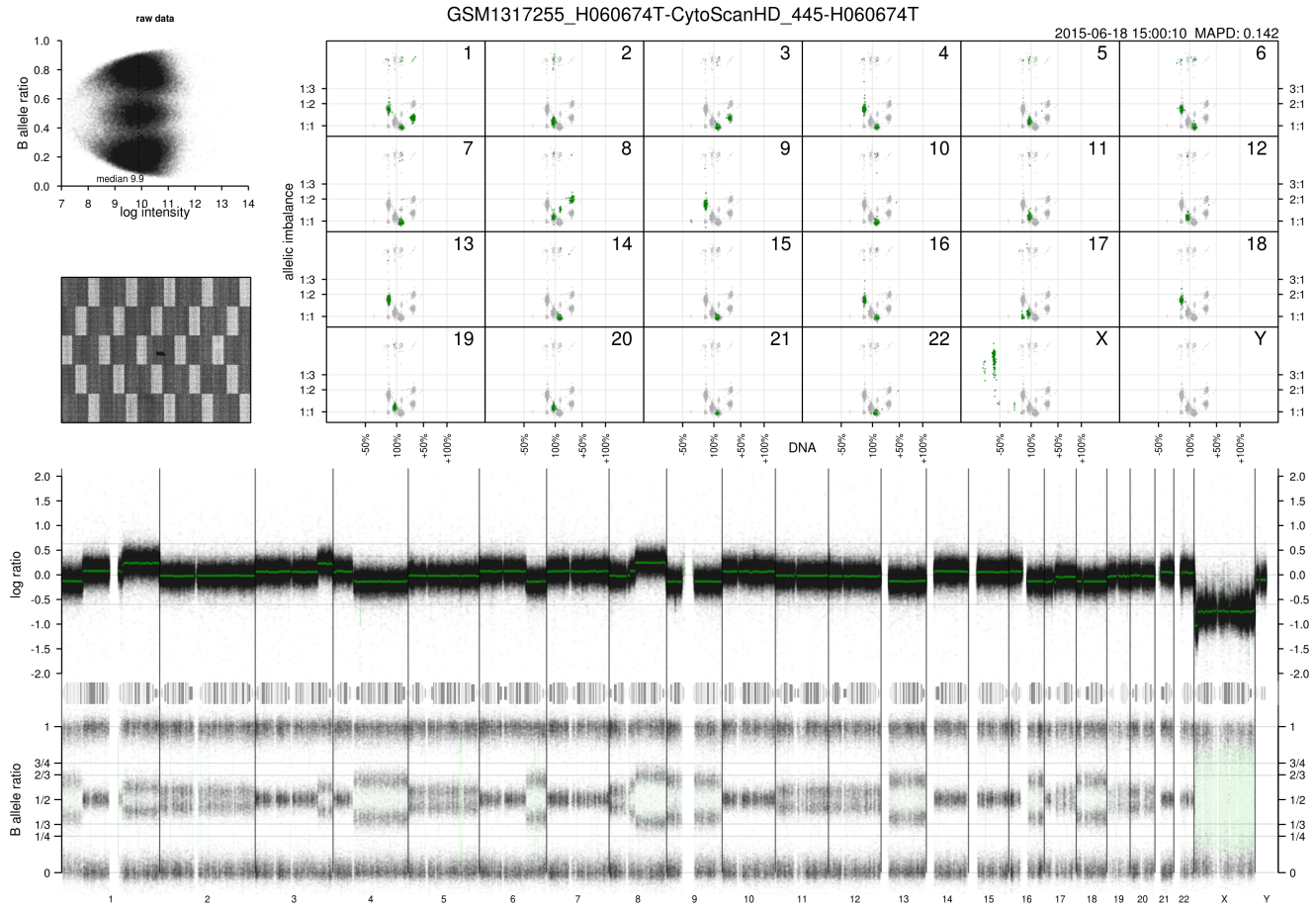
Example 5. A high aneuploid cancer cell line, average copy number is near 4.



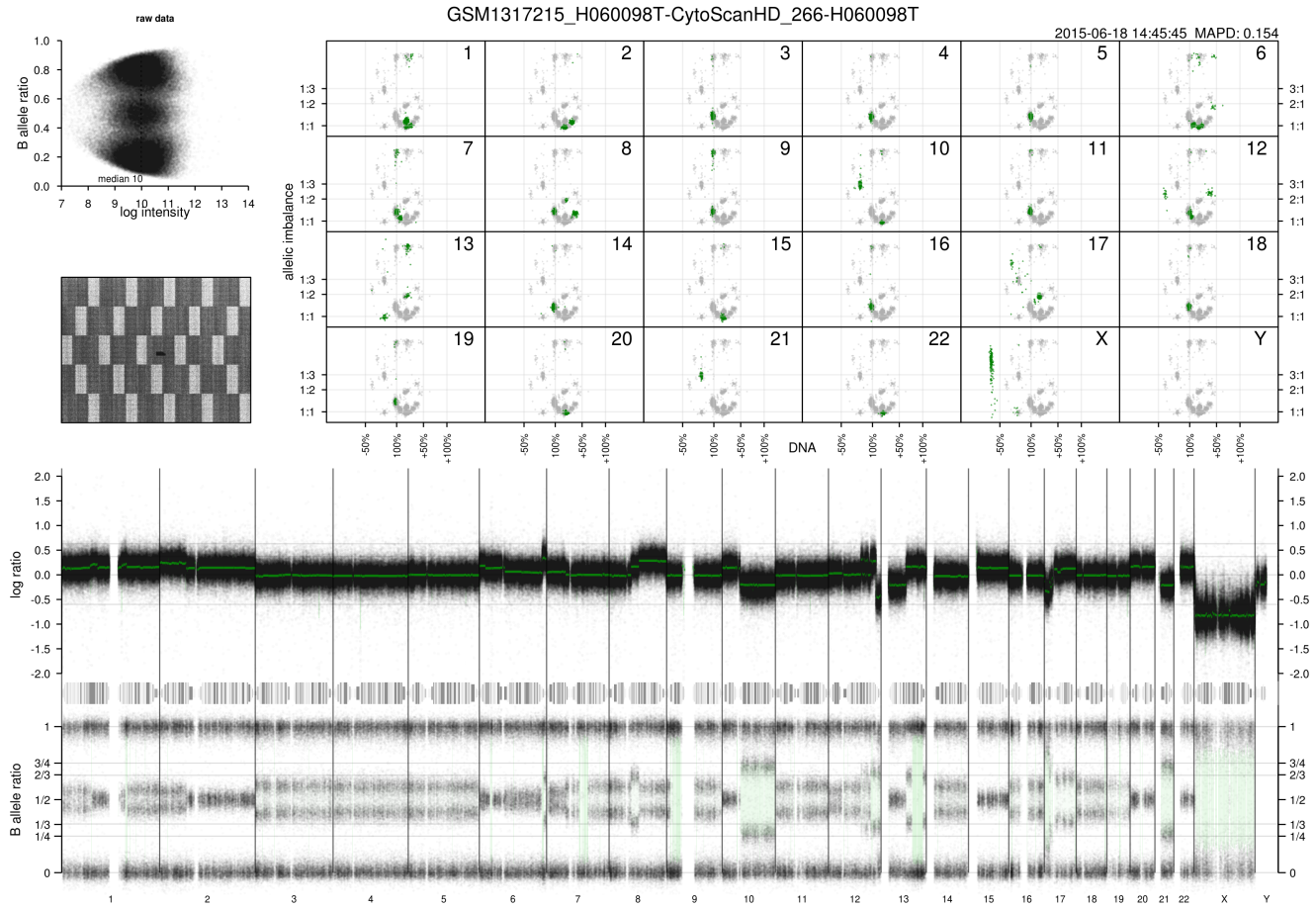
Example 6. Almost entirely homozygous cancer cell line. Averge copy number is unclear.



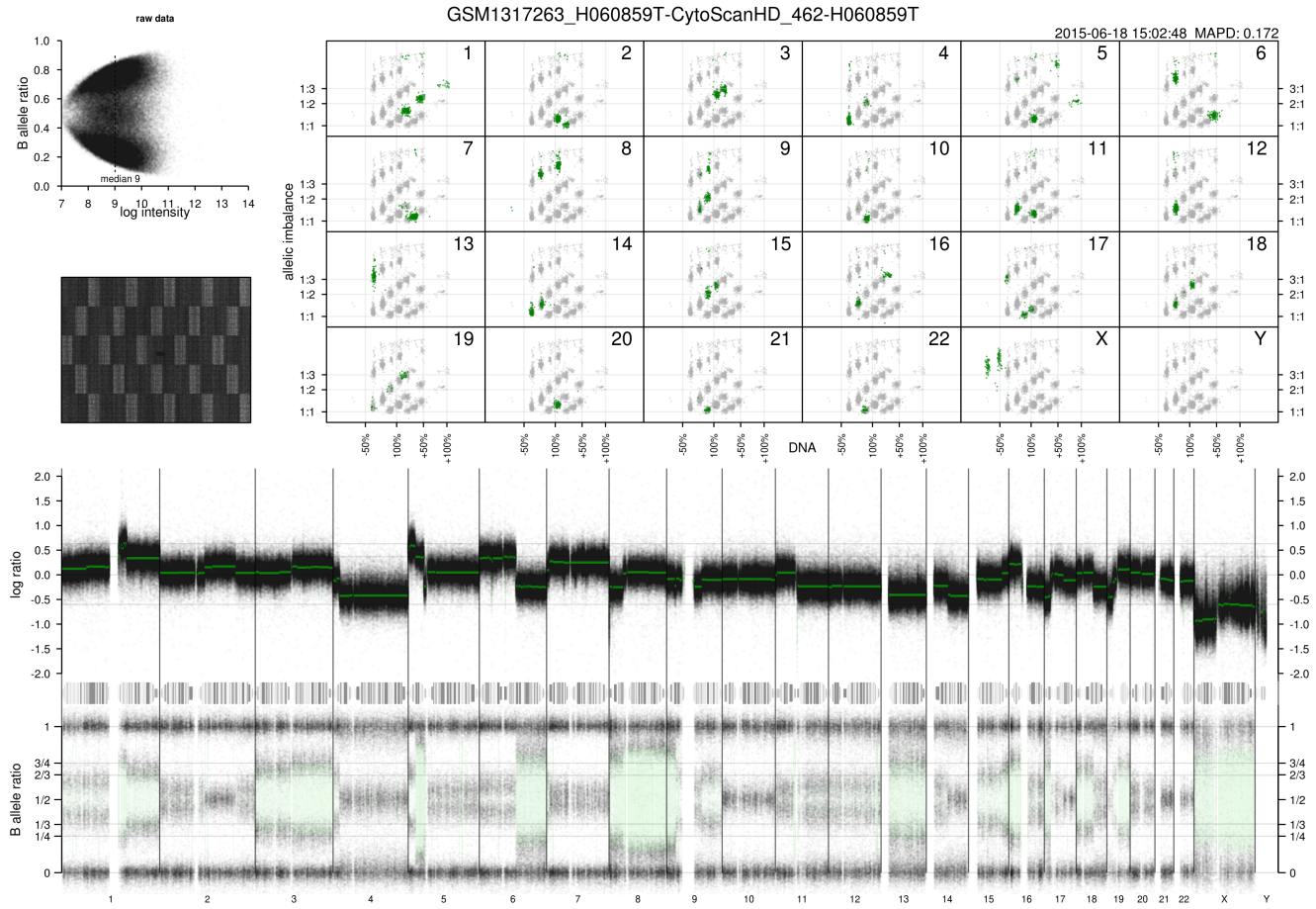
Example 7. Heterogeneous cancer cell line. Copy numbers appear to differ between cell populations, making the interpretation difficult.



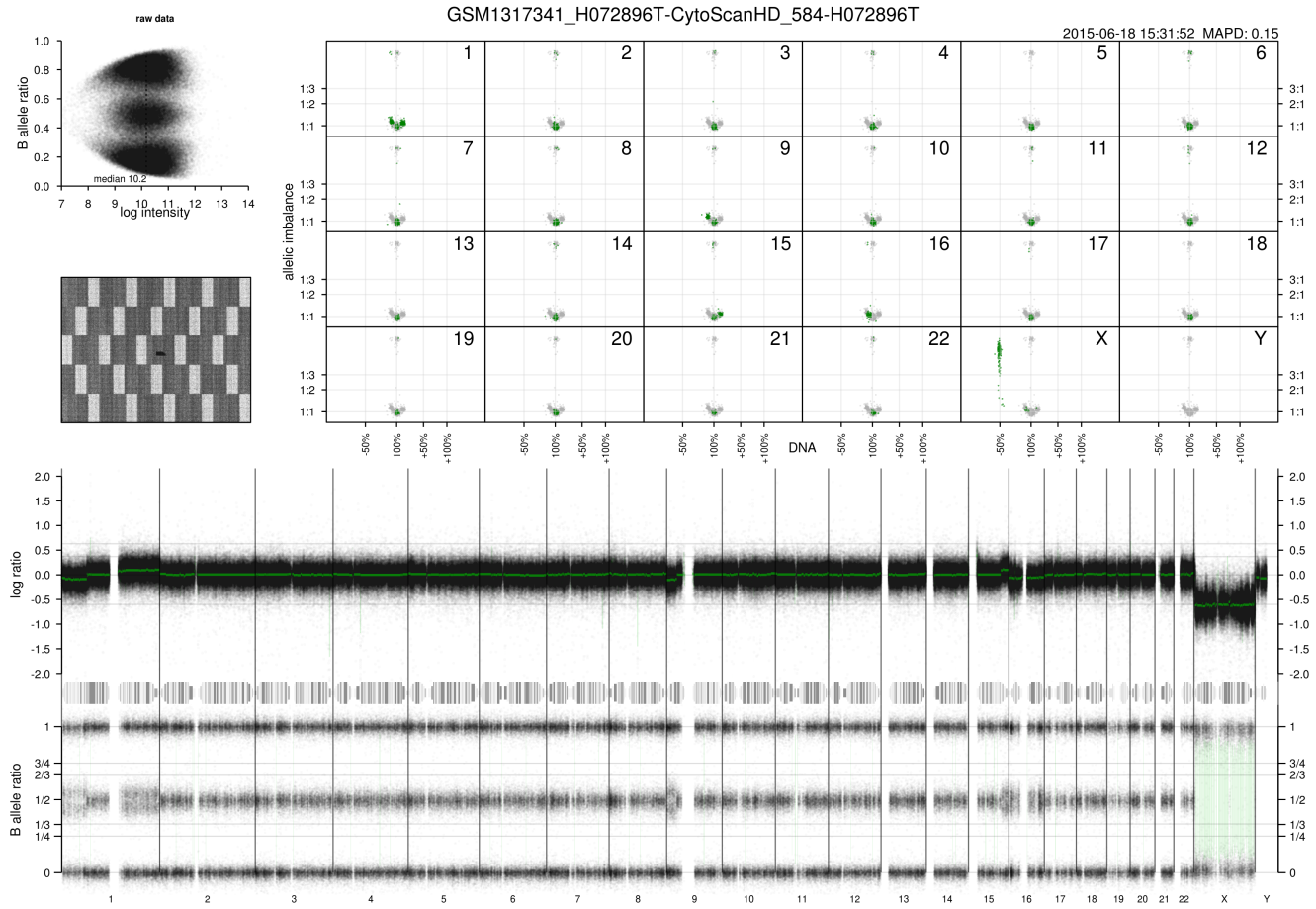
Example 8. Cancer tissue sample with relatively high normal cell content and an average copy number near 3 in the cancer cell population. There are 2 (2+0) copies of chromosome 13, 3 (2+1) copies of chromosome 2, and 3 (2+1), 4 (3+1) and 6 (5+1) copies along chromosome 8.



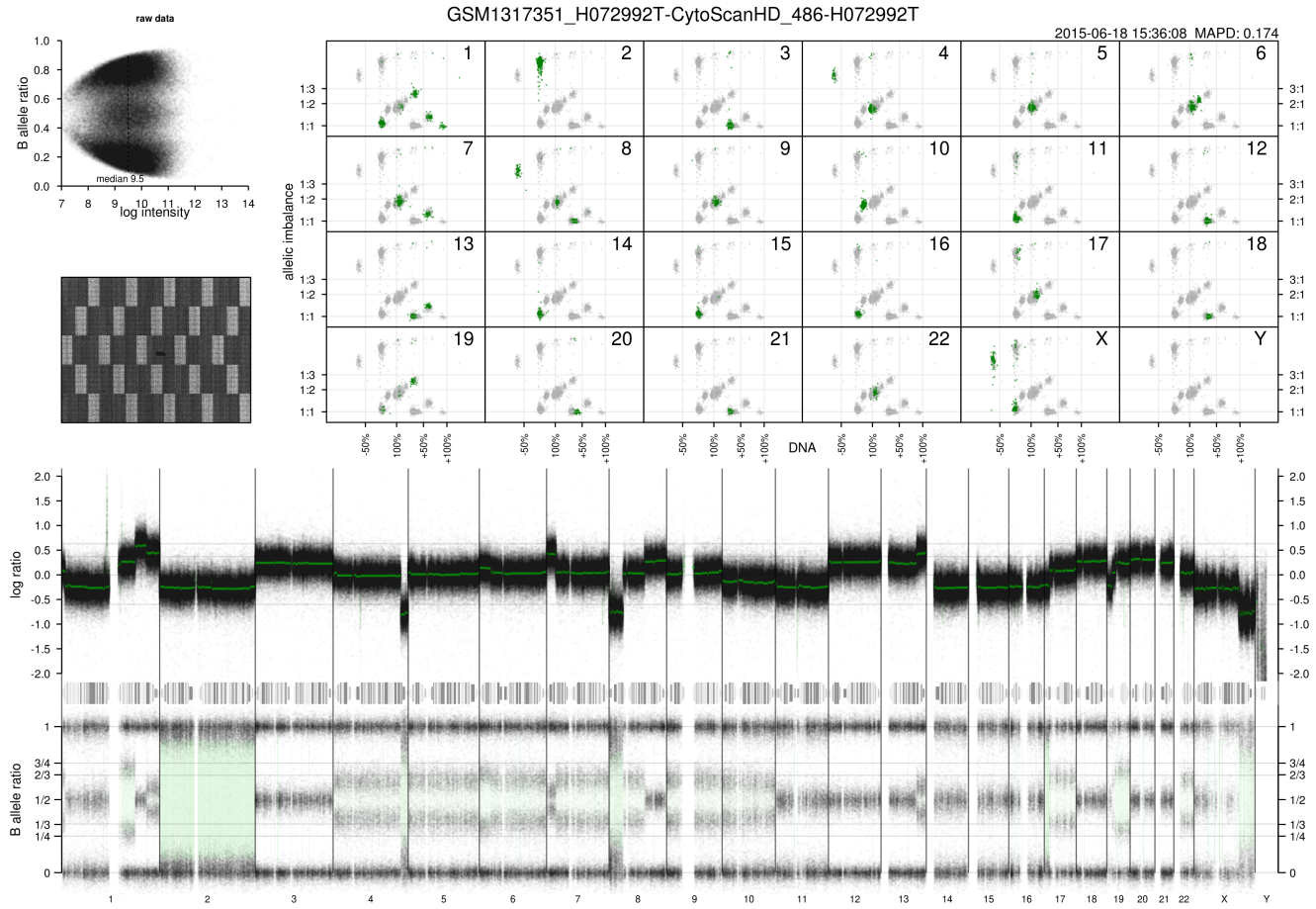
Example 9. Cancer tissue sample with an average copy number near 3 in the cancer cell population. Despite moderate tumour cell content, complete (copy-neutral) LOH events can be observed regionally on e.g. chromosomes 7, 9 and 13. Such a pattern is often seen in consanguineous patients, therefore that LOH is most likely constitutional rather than somatic. For 2 (2+0) copies (e.g. chromosome 21), an allelic imbalance of 1:3 is observed. With the normal cells assumed to have 1+1 copies, the tumour cell content must be very near 50%.



Example 10. High aneuploid cancer tissue sample. Average copy number is about 4.5 The X chromosome has 1 (p-arm) or 2 (q-arm) copies in the tumor cell population. As there is only one copy of X per cell in the normal cell pupolation, it's segment clusters deviate from those of the other chromosomes (lower DNA abundance and higher allelic imbalance).



Example 11. Cancer tissue sample with low tumour cell content and few somatic copy number alterations. Average copy number in the tumour cells is likely near 2 with deletions affecting 1p, 9p and 16, and duplications affecting 1q and 15q.



Example 12. Cancer tissue sample with average copy number near 3 and some evidence of tumour cell heterogeneity. It is possible that chromsome 10 appears in 1 copy per cell in part of the tumour cell population and 4 (2+2) copies in remaining tumour cells.