

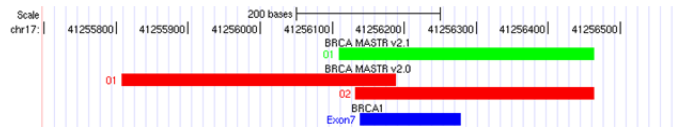
## Changes in BRCA MASTR v2.1 compared to BRCA MASTR v2.0

### Rationale

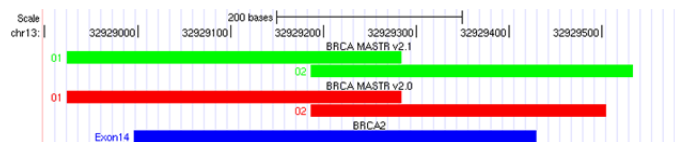
Amplicon BRCA1\_ex07\_01 had the tendency to show low read counts. Detailed analysis revealed this was caused by the combination of a polymorphic GAA-repeat and the presence of a 12bp long poly-A stretch. We opted to ban the polymorphic GAA-repeat from any amplicon and found a solution in which both amplicons for BRCA1 exon7 were replaced by 1 amplicon encompassing the entire exon. Please note that the total number of amplicons is now reduced to 93.

For reasons of compatibility changing the primer for BRCA1 exon7 forced a secondary change in the primers of amplicon BRCA2 exon14. The detailed changes are outlined below.

- BRCA1 exon7:
  - BRCA1\_ex07\_01<sup>(v2.0)</sup> is removed from Plex3
  - One primer of BRCA1\_ex07\_02<sup>(v2.0)</sup> is changed, the new amplicon encompasses the entire exon: BRCA1\_ex07\_01<sup>(v2.1)</sup> (Plex4)
  - Figure:

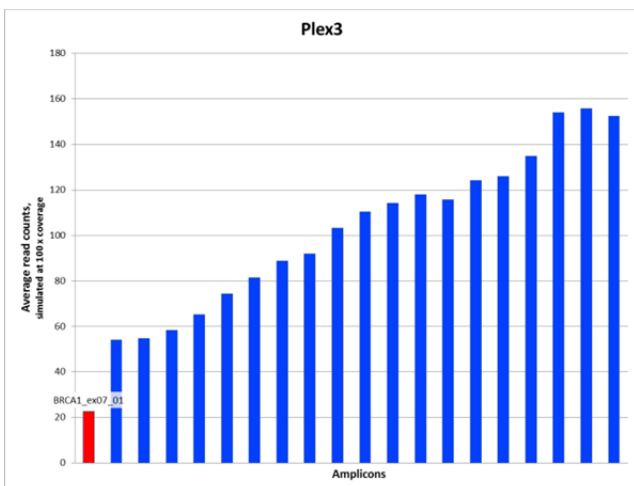


- BRCA2 exon14:
  - One primer of BRCA2\_ex14\_02<sup>(v2.0)</sup> is changed (30 bp more into the intron): BRCA2\_ex14\_02<sup>(v2.1)</sup> (Plex4)
  - Figure:



- Resulting read counts:

### BRCA MASTR v2.0



### BRCA MASTR v2.1

