

In figure (g), Y axis shows maternally inherited genotype in the upper line (in black dots) and paternally inherited genotype in the bottom line (in black dots). The middle line shows in red if the rates of maternal/paternal genotypes are larger than 5 and in blue if the rates are smaller than 0.2.

Figure 7. Cryptic AOH reported by Low-pass GS. Figures (a), (c) and (e) show the copy-number distribution in 17C1122, 17C1175 and 17C1176, respectively, while figure (b), (d) and (f) show the distribution of rates of heterozygous SNVs in each of these three samples. X axis in each figure indicate the genomic location. Y axis in figures (a), (c) and (e) indicate the copy-number while in figure (b), (d) and (f) show the rate of heterozygous SNVs. In figure (b), (d) and (f), the candidate regions with AOH reported by low-pass GS are indicated by each pair of a red arrow and the number of windows involved in the bottom. The green dotted line shows the region of *KCTD7* gene across each figure, while in figure (b), the cryptic AOH is highlighted in yellow background.

Figure 8. Correlations of different parameters between GS and Low-pass GS in sample HG00733. (a) The correlation between the parental genomic differences (Y axis) and the rates of heterozygous SNVs (X axis) in GS data. (b) The correlation between the parental genomic differences (Y axis) and the rates of homozygous SNVs (X axis) in GS data. (c) The correlation between the rates of homozygous SNVs (Y axis) and the rates of heterozygous SNVs (X axis) in GS data. (d) The correlation between the rates of heterozygous SNVs (Y axis) indicated by low-pass GS and the rates of heterozygous SNVs (X axis) calculated by GS data. (e) The correlation between the rates of homozygous SNVs (Y axis) indicated by low-pass GS and the rates of homozygous SNVs (X axis) calculated by GS data. (f) The correlation between the rates of homozygous SNVs (Y axis) and the rates of heterozygous SNVs (X axis) in low-pass GS data. In each figure, P value of Pearson correlation coefficient is shown in red.

Figure 9. Observation of decreased rates of heterozygous SNVs in the region with a heterozygous deletion. Heterozygous deletion arr[GRCh37]1q23.1q25.2(158043081_176445395)x1 dn was reported in sample 18C0241. (a) Distribution of copy-numbers among the windows (indicated by black dots) in chromosome 5 in this sample. The X axis shows the genomic location in all figures, while the Y axis indicates the copy-number in figure (a). The large deletion is shown with a pair of line and arrow with affected band in figure (a). Distribution of normalized rates of heterozygous SNVs in low-pass GS (b) and rates of heterozygous SNVs (c) in the same sample both show the decreased rates in the copy number deleted region. In figures (b) and (c), regions with consecutive decreased rates of heterozygous SNVs are indicated by red arrows with the numbers of windows involved in the bottom.

Figure 10. Detection of AOH in chromosome 2 of sample HG00733. (a) Distribution of copy-numbers among the windows (indicated by black dots) in chromosome 2 in this sample. The only deletion is shown by a purple arrow. X axis shows the genomic location in all figures, while Y axis indicates the copy-number in figure (a). Distribution of normalized rates of heterozygous SNVs (b) and normalized rates of homozygous

SNVs (c) cross chromosome 2 by low-pass GS. Distribution of rates of heterozygous SNVs (e) and rates of homozygous SNVs (f) cross chromosome 2 by GS. In figures (b) and (d), AOH (indicated by red arrows with the numbers of windows involved in the bottom) identified by observation of consecutive decreased rates of heterozygous SNVs. In figures (c) and (e), regions with consecutive increased rates of homozygous SNVs (indicated by blue arrows with the numbers of windows involved in the bottom). (f) Distribution of parental genomic differences across chromosome 2. Y axis in figure (b-f) shows the rate of each corresponding parameter. Genomic region of the large AOH seq[GRCh37] 2p23.2p21(29700000_42600000)x2 hmz is shown by a pair of green dotted lines across each figure.

Figure 11. AOH detected within a mosaic trisomy event in sample 16C0836 (a) Copy-number distribution reported by low-pass GS with windows indicated by black dots. The results confirmed approximately 40% increase of the whole chromosome 7 (indicated by a blue line). X axis in figures (a-d) indicates the genomic locations across chromosome 7, while in figure (a), Y axis represents the copy-number. Figures (b) to (d) show the distribution of rates of “germline” heterozygous SNVs (AB), homozygous SNVs and “mosaic” heterozygous SNVs (AAB/ABB), respectively. In figure (b), the candidate regions with AOH detected are indicated by each pair of red arrow and the number of windows, while in figure (c), the windows with increased rate of homozygous SNVs within those regions reported in figure (b) are shown by each pair of blue arrow and the number of windows. In figure (d), the candidate regions with increased rate of “mosaic” heterozygous SNVs are shown by each pair of a blue arrow and the number of windows.

Figure 12. AOH detected in sample aCGH15274. (a) Copy-number distribution reported by low-pass GS with windows indicated by black dots. X axis in figures (a-e) indicates the genomic locations across chromosome 6, while in figure (a), Y axis represents the copy-number. Figures (b) to (d) show the distribution of rates of “germline” heterozygous SNVs (AB), homozygous SNVs and “mosaic” heterozygous SNVs (AAB/ABB), respectively. In figure (b), the candidate regions with AOH detected are indicated by each pair of red arrow and the number of windows, while in figure (c), the windows with increased rate of homozygous SNVs within those regions reported in figure (b) are shown by each pair of blue arrow and the number of windows. In figure (d), the candidate regions with increased rate of “mosaic” heterozygous SNVs are shown by each pair of a blue arrow and the number of windows. In figure (e), Y axis shows maternally inherited genotype in the upper line (in black dots) and paternally inherited genotype in the bottom line (in black dots). The middle line shows in red if the rates of maternal/paternal genotypes are larger than 5 and in blue if the rates are smaller than 0.2.

Figure 13. Rate distributions of different types of SNVs in deletion and duplication. (a) Copy-number distribution reported by low-pass GS with windows indicated by black dots. CNV analysis result shows a deletion seq[GRCh37] del(8)(p23.3p23.2) chr8:g.10134_5523520del and a duplication seq[GRCh37] dup(8)(q22.1q24.3) chr8:g.98620704_146298884dup in case 17BA0551. The X axis in figures (a-d)