

Table 1. Key factor analysis for simulated single exon CNV events. Results presented are for 971 identified (*true positives*) and 29 missed (*false negatives*) simulated single exon deletions with 50% of original sequencing reads removed. Data distribution plots for each factor are provided in Supp Figures S6-S13.

| Factor | True positives | False negatives |
|--|--------------------------------|-------------------------------|
| <i>Sample independent factors</i> | | |
| Exon size (<i>n</i> nucleotides) | 183 (12-5692; 280.9) | 122 (11-347; 80.7) |
| GC content (%) | 50.2 (28.0-76.1; 9.6) | 54.3 (26.2-76.1; 17.4) |
| Distance to neighbouring exons (<i>n</i> nucleotides) | 4186 (64-871570; 29256) | 2621 (359-30814; 5580) |
| <i>Test sample dependent factors</i> | | |
| Total read depth (<i>n</i> non-duplicate reads) | 1701 (62-71020; 3408) | 280 (0-1756; 339) |
| Normalized total read depth (rpkm) | 4024 (348-16508; 1942) | 1447 (0-5414; 1401) |
| Intra-sample variability (rpkmCV, %) | 39.8 (0-102.2; 15.6) | 52.6 (21.6-88.5; 17.3) |
| Nucleotides with insufficient coverage (%) | 0.14 (0-54.1; 1.9) | 29.8 (0-100; 40.4) |
| <i>Reference sample dependent factors</i> | | |
| Inter-sample variability (rpkmCV, %) | 6.1 (0.1-39.5; 4.8) | 22.0 (0-88.6; 20.8) |

CV, coefficient of variation; *rpkm*, reads per kilobase per million. Reported statistics: mean(min-max; *sd*).