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ExomeXtra® Sequencing with CNV Backbone

Why We Need a Higher Output for the New ExomeXtra®

The new ExomeXtra® Sequencing products include a new and unique CNV backbone that enables genome-wide analysis and high-resolution detection of copy number variations (CNVs) for clinical applications. The CNV detection in non-coding regions is unique in exome sequencing, providing an increased probability of finding disease-causing mutations outside of exomic regions. Due to this new CNV backbone, more targets need to be enriched and sequenced, resulting in a higher output of 18 Gb instead of 12 Gb.

The higher output of 18 Gb:

- increases the coverage
- decreases the noise level
- slightly increases the number NGS calls
- increases the sensitivity of the CNV calls

Figure 1 shows the highly significant decrease in coverage for an output of 12 Gb compared to 18 Gb for both the exomic regions as well as the CNV backbone targets. The coverage can decrease even further for samples with poor quality, increasing the risk of a high false positive CNV calling rate. The lower output increases the noise level and decreases the number and sensitivity of the SNV calls. Thus, we highly recommend using the validated output of 18 Gb for the new ExomeXtra® Sequencing products.

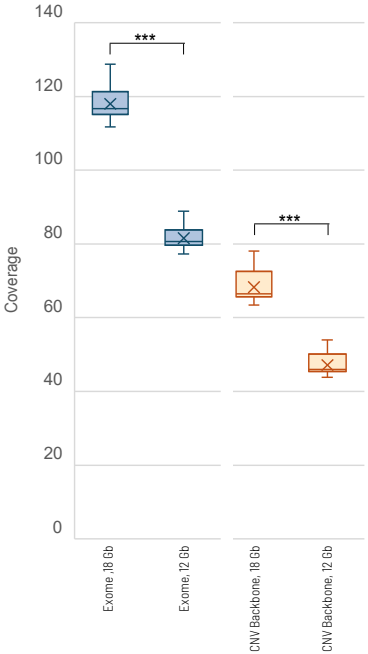

Figure 1: Exome and CNV Backbone coverage for an output of 18 Gb and 12 Gb.

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