Research & Pharma Solutions

Exome Sequencing on the NovaSeq™ X Plus

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The NovaSeq™ X Plus instrument employs the same general principles for sequencing libraries of short DNA molecules as the NovaSeq™ 6000 instrument. However, there are differences in the sequencing chemistry (e.g., modified nucleotides, polymerases) and the image acquisition system.

We already conducted several tests with different datasets and specifications to show that the sequencing results of the NovaSeq™ X Plus and the NovaSeq™ 6000 are comparable. These results are available in our NovaSeq™ X Plus Performance Flyer. We now want to focus specifically on comparing the performance of the two instruments with respect to exome sequencing.

We sequenced our WES Classic (12 Gb) product on both instruments using the well-characterized Genome in a Bottle (GIAB) sample HG001 as a reference. Subsequently, the sequenced samples were analyzed. The coverage is comparable for both instruments (120.8 for NovaSeq™ 6000 and 121.3 for NovaSeq™ X Plus).

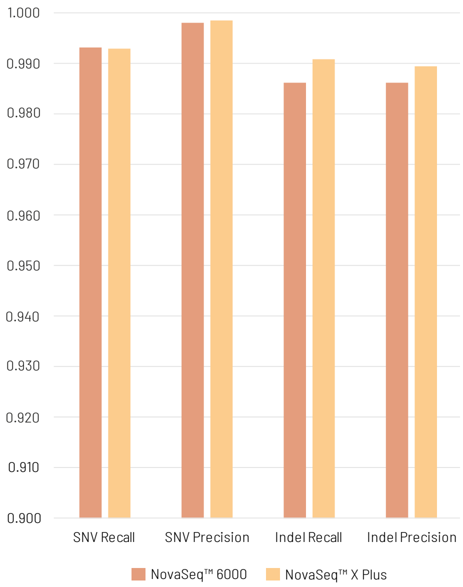

Figure 1 | Variant Calling Comparison. Recall and precision values for SNVs and indels are compared between the two instruments.

For this GIAB sample, so-called truth sets for high-confidence calls of single-nucleotide variants (SNVs) and small insertions and deletions (indels) are available. By comparing the predicted variant calls to the truth sets, variant calling parameters such as recall (also known as sensitivity), and precision (also known as positive predictive value) can be calculated. As shown in figure 1, variant calling parameters are comparable between the two instruments. For indel calling, recall and precision are slightly (but not significantly) higher with the NovaSeq™ X Plus instrument.

The NovaSeq™ X Plus instrument allows for highly accurate variant calling in the same way as the NovaSeq™ 6000. Thus, both instruments are equally suited for exome sequencing.