



Illumina Whole Exome Sequencing and Bioinformatic Methods

Illumina whole exome sample enrichment was prepared using the [Twist Exome 2.0 Plus Comprehensive Spike-in](#) and the [Twist Mitochondrial](#) panels. Prepared libraries were then loaded onto an Illumina NovaSeq X Plus sequencer in one or more multiplexed shared-flow-cell runs, producing 2x151bp paired-end reads.

Demultiplexing, quality control and adapter trimming was performed with bcl-convert¹ (v4.2.4). Sequencing statistics are included in the '*DNA Sequencing Stats.xlsx*' file.

Additional resources

- Exome enrichment reference: [Twist Exome 2.0 hg38 reference](#)
- Sample data sets and run outputs: <https://basespace.illumina.com/s/GBEdTs1IKRCM>
- Example Baseline files: <https://basespace.illumina.com/s/X8j2Lr7cKJXu>
- TruSight Oncology Analysis Files: <https://basespace.illumina.com/s/CXP94TyjB9Tr>

¹ bcl-convert: A proprietary Illumina software for the conversion of bcl files to basecalls.