

Fact Sheet: Changes between the 2018 report update (2018-01) versus the 2016 report (2016-02.2)

General for all platforms:

platforms in the 2018 report update: CMA, Illumina HiSeq[®] 2500, Illumina NextSeq[®] 550 and Illumina HiSeq X[™]; platforms in the 2016 report: CMA, Illumina HiSeq[®] 2500 and Illumina HiSeq X[™]:

- Overhead cost proportion: 22.3% vs. 23%
- No inflation adjustments were made for prices except labour from previous years for the 2018 report update; labour prices were adjusted by an annual 1.5% increase to 2017/2018. On the other hand, all prices were inflated for the 2016 report.

Exome and Genome analyses:

- Addition of filtration steps to analyze ~300-400 variants in order to flag variants of interest for both primary and secondary variants (impacts labour costing)

Whole Exome Sequencing:

- Addition of a new platform (NextSeq[®] 550)
- Exome output per month (NextSeq[®] 550): 80 (64-96)
- Exome output per month (HiSeq[®] 2500): 83 (70-95) vs. 87.5 (75-100)
- Bioinformatics
 - Pipeline: no change (Genome Analysis Toolkit (GATK))
 - Decrease of 1 step (temporary BAM files) for file storage
 - Additional 2 steps (extra step in recalibration; indel realignment) for computation use
 - Price per node: \$26,804 vs. \$9560

Whole Genome Sequencing:

- Elimination of platform HiSeq[®] 2500
- Inclusion of trio in addition to probands on the same platform (HiSeq X[™])
 - Genome output per month 72 (64-96)
 - Proportion for follow-up testing (Sanger sequencing) is 10%
 - Small and large equipment volume: ½ of proband model
- Genome output per month 72 (64-96) vs. 150 (10% variability assigned)
- Bioinformatics
 - Pipeline: GATK vs. HAS
 - Additional 4 steps (recalibration; post-recalibration merge; CNV detection; SV detection) for computation use
 - Price per node: \$26,804 for all steps in 2018. In 2016, annotation was done on the old node (\$9560). Otherwise, same as 2018.