Dr. Guy Froyen from the Molecular Diagnostics group of the Jessa hospital performed comparative analysis of three software packages for mutation detection in tumors.

Statement Dr. Guy Froyen:

In the Molecular Diagnostics group of the Jessa hospital we are currently implementing targeted NGS for the detection of actionable mutations in solid as well as hematological tumors with diagnostic, prognostic or therapeutic value.

For data analysis, visualization and management we performed a comparative analysis of three software packages starting from the Fastq or Bam files generated by the MiSeqReporter after targeted capture with TruSeq or TruSight panels, run on a MiSeq instrument (Illumina).

Compared to the two competitors' software modules, SeqNext (SeqPilot) from JSI performed best when taking into account the price, detection efficiency, speed, flexibility, robustness, data storage needs and customer service.

SeqNext was able to unequivocally detect and annotate all SNVs and indels including crucial deletions up to 52 bp (in CALR) and insertions of 51 bp (in FLT3). The number of analysis settings are multitude and once the different parameters have been set in consultancy with the helpful and friendly JSI specialists, the analysis can easily be performed by non-bioinformaticians, which is very important for hospitals where such highly specialized persons are absent. Customized data reporting is included and can be integrated in LIMS. A onetime license fee with acceptable yearly maintenance costs makes the software affordable to all laboratories.

Guy Froyen, PhD
Jessa Ziekenhuis
Campus Salvator, Klinische Biologie
Laboratorium voor Moleculaire Diagnostiek (LMD)
MT1 - lokaal 1.37
Salvatorstraat 20
3500 Hasselt
http://www.jessazh.be