A tale of two platforms: An evaluation of the Roche GS Junior and Illumina® MiSeq next-generation sequencing instruments for forensic mitochondrial DNA analysis

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ABSTRACT

Next-generation sequencing (NGS) refers to a suite of technologies that enable rapid and high-throughput generation of large amounts of identical sequence information. These techniques are being increasingly employed in forensic science to accelerate investigations by enabling cost-effective, rapid generation of large amounts of detailed sequence information from forensic samples. This study compares the Roche GS Junior and Illumina® MiSeq NGS platforms, as well as the commercial DNA Investigator™ kits for mitochondrial DNA (mtDNA) analysis. Novel methods were employed to remove sequencing artifacts associated with both platforms that are typically removed through deconvolution analysis. The study demonstrates that the Roche GS Junior is capable of producing a thorough deconvolution of forensic casework samples, whereas the Illumina® MiSeq is not, as evidenced by unexpected variants observed in data obtained using both instruments.

SEQUENCING CHEMISTRY

NGS LIBRARY PREPARATION

DATA ANALYSIS – CLC GENOMICS WORKBENCH

EXPECTED VARIANT FREQUENCIES – WHOLE GENOME DATA

UNEXPECTED VARIANTS IN HV DATA – SUMMARY

EXPECTED VARIANTS IN HV DATA – SUMMARY

ALIGNMENT DIFFERENCES

WHOLE GENOME SAMPLES – COVERAGE MAPS

REFERENCES

UNEXPECTED VARIANTS IN HV DATA – SUMMARY

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CONTACT

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