POSTER 44

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EVALUATION OF FLUIDIGM SNPTRACE™ PANEL, WITH A COMPARISON TO DNA QUAL AND BIOANALYZER FRAGMENT ANALYSIS, FOR DNA QUALITY ASSURANCE WITHIN THE TUMOUR BANK PROGRAM

ABSTRACT

Translational research is the key component to apply basic science findings into practical medicine. High quality human biological specimens are the foundation of accurate and reproducible research result and will maximize the productivity of translational research. To assure that the best samples are provided to translational research, quality assurance (QA) programmes are an essential part of biobanks' routine.

To identify the most feasible technology for regular biobank QAP daily practice, two technologies promoted from use in biobanking QAP have been compared, the Fluidigm SNP trace panel (Fluidigm, USA) and DNAqual (Eurobio, France). 93 samples, including 80 DNA samples from 4 donors and 13 DNA samples isolated from bone marrow aspirates (BMA) smeared on slides, were examined using both technologies. BMA slides had been stored at the room temperature and exposed to air for 2-14 years. In addition, to create a range of possible conditions occurring in biobank daily practice, DNA samples from donors were treated with multiple freeze-thaw cycles (0-20), varied snap delay period (0-21 days), radiation, sonication, heat, UV and mixed contamination (MC) respectively. DNA qualities have been verified using Bioanalyzer DNA 7500 Kit (Agilent, USA).

The results showed that the Fluidigm panel could detect 50% MC between samples accurately. The sensitivity of the contamination detection need to be further tested. It also reported the degraded DNA qualitatively instead of quantitatively. By contrast, DNAqual can provide the quantitative DNA quality index although it is not designed for detecting MC at all. The accuracy of the index need to be further validated.

Inaddition, the consistency rate of the Fluidigm panel is 100% within the plate due to its qualitative feature, but it is not optimal to compare the results across the plates, whereas due to its qPCR feature, DNAqual can compare the readings across runs. All these results will be further validated and elucidated.

These results form the basis of a better biospecimen QA solution for the Australian biobanking community. Furthermore, we will discuss the ongoing implementation of these new QA platforms into daily biobank practice and how these will benefit translational research involving genomics.

POSTER 45

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A BETTER WAY - AMPLISEQ EXOME FOR CLINICAL RESEARCH

ABSTRACT

Many laboratories are actively engaged in implementing large-scale sequencing in clinical and diagnostic settings. There is significant debate about the best approach (gene-specific panels, WES, WGS), each of which has benefits and disadvantages. In addition to accuracy and utility, other practical considerations must be taken into account such as cost, scalability, practicality of implementation and infrastructure requirements.

In many settings, WES may strike the appropriate balance between these factors.Since its development, WES has made incremental advances but for the most part is still based upon hybridisation to biotinylated probes. Recently a completely new way to enrich exomes was developed based on massively parallel PCR (AmpliSeq). In combination with the Ion Torrent Chef and Proton sequencer, the process is rapid, flexible, scalable in a variety of settings and highly reproducible. it is now possible to routinely and economically sequence 100X+ coverage exomes).