IAB Exome

We designed IAB Exome with the aim to balance the level of coverage of relevant regions at minimised sequencing capacity. IAB Exome combines the advantages of Illumina DNA Prep with Enrichment - CE-IVD certified library preparation kit and Twist Alliance VCGS panel plus Twist Mitochondrial panel to get standardised coverage and data output for human Whole Exome Sequencing (WES).

Introduction

Clinical researchers and geneticists frequently look for the best human WES solution to achieve robust coverage of the entire exome and/or various virtual panels.

IAB exome was designed to ensure that not only standard protein-coding regions (exons) are covered, but the additional information related to the selected gene regulation regions, introns and mitochondrial DNA (mtDNA) can be generated; these are beneficial for genomic analysis focused on cancer, complex diseases, rare diseases, Mendelian disorders, carrier screening etc.

IAB Expertise

The long-term acquired expertise of IAB team is based on precise quality control (QC) of all the steps – starting with DNA isolate through library preparation and NGS library QC essential for proper settings of sample pooling and subsequent sequencing processing, sequencing QC ending with data quality QC and reliable interpretation.

Protocol

Sample

Human genomic DNA: input 100 ng; DNA concentration: ≥ 10 ng/µl; Minimum sample volume: 20 µl

Quality Control (DNA)

- DNA purity: A260/280 ~1,6-2,0
- DNA quantity: C > 10 ng/µl in total volume > 20 µl (min. 200 ng gDNA)
- DNA integrity: Intact DNA with no degradation

Normalization of the samples

Pre-enrichment part

- QC – Pre-enrichment library quantification (≥ 300 ng/library)
- Plexing - 12 libraries (Fig 1)

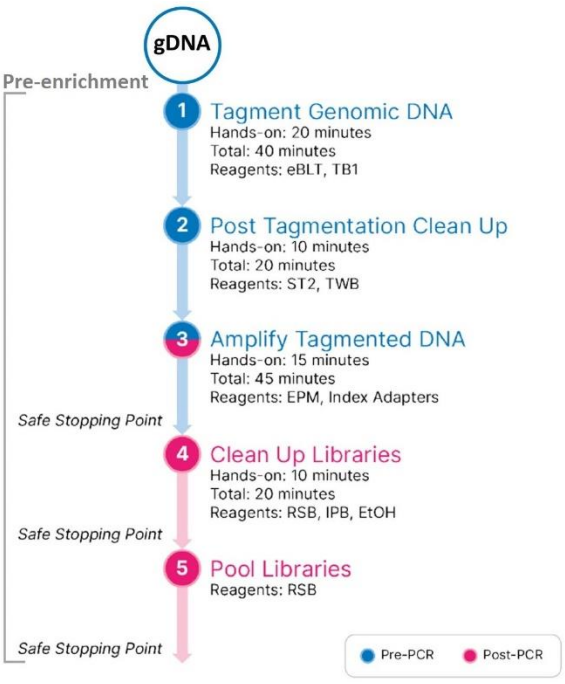

Figure 1

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Hybridization

- Twist Alliance VCGS
- Twist Mitochondrial Panel

Post-enrichment

- Capture probes with streptavidin beads (Fig 2)

Library QC

- Quantification
- Quality check by capillary electrophoresis
- Normalization of libraries based on molarity

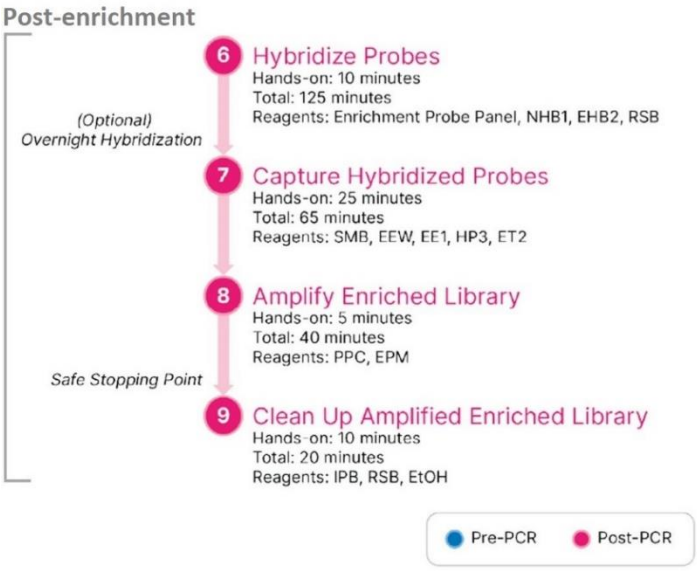

Figure 2

Sequencing

- NovaSeq X Plus (Illumina, USA) (Fig 3)
- Pair-end sequencing 2×150 bp
- %Q30 > 75% (full NovaSeq X Plus specifications)
- Coverage target - Ø100x/sample

Figure 3

Results

Raw sequencing data (FASTQ files) are included in the Wetlab report. For each sequencing service provide complete information of service workflow and outputs (Sample QC metrics, library preparation, NGS library QC and sequencing metrics). (Fig 4, Fig 5, Fig 6)

Data analysis: We offer the complete solution for secondary and tertiary analysis of WES data. Secondary analysis usually includes QC, trimming, reads mapping and variant calling. DRAGEN Bio-IT Platform (Illumina) provides accurate, comprehensive and efficient analysis for all these steps. Emendgene (Illumina) covers the whole tertiary analysis for us, including variant annotation, prioritisation and reporting.

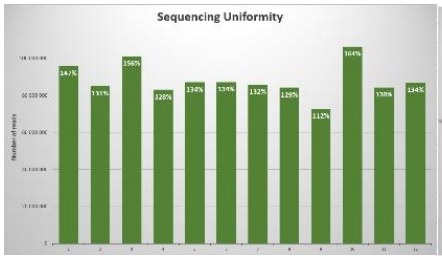

Figure 4

Figure 5

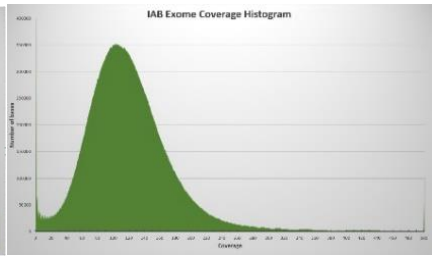

Figure 6

Conclusions

The IAB Exome **targets clinically relevant exome regions**, allowing the identification of desired germline variants (SNP, InDel, CNV) at a high level of uniformity.

About IAB

The experienced team follows IAB Exome workflow with strict quality procedures to perform the essential QC steps on your samples and generate reliable and repeatable results.

Certification

- *ISO 9001:2016*
- *ISO 13485 ed.2:2016*

The IAB Exome is validated using the Coriell NA12878 genomic reference standard

TWIST

Twist Bioscience is well known for its best-in-class performance NGS panels with a major focus on maximising the coverage of clinically relevant content of various databases (RefSeq, CCDS, GENECODE, ACMG Guidelines, etc.) while simultaneously minimising the sequencing requirements.

Illumina

Illumina is a global leader in sequencing and array technologies that are fueling groundbreaking advancements in life science research, translational and consumer genomics, and molecular diagnostics.

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