mfine
/diagnostics

ExomeFirst

Simultaneous Detection of Mutations and Copy Number Variations | Greater Diagnostic Yeild | Lesser Cost

Twist Exome 2.0 Comprehensive Whole Exome spike-in with Mitochondria

Customized to provide the value of multiple clinical panels wrapped into one and enable the detection of rare and inherited diseases, as well as germline cancers.

FEATURES

>95%
SENSITIVITY
In CNVs detection spanning two or more exons

38.2MB
Panel Size

>20K
Genes Analysed

>85%
OF BASES
are sequenced with a quality score of ≥Q30

100%
NGSE-A, NGS-A
compliance in CAP proficiency testing

99%
SPECIFICITY
is guaranteed for all reported variants

99%
Clinvar variants covered

99.3%
Targeted Regions Covered At 20x

100%
COVERAGE
Of Mitochondrial Genome

01 **Superior coverage from major genetic databases**
like RefSeq, CCDS, GenCode, Clinvar and ACMG73

02 **RefSeq transcripts analyzed**
across the OMIM morbid set of genes

03 **DRAGEN 4.2.4 SNV, CNV and SV calling powered**
by enhanced machine learning and multigenome graph

04 **Coverage of alternative transcripts**
from GENCODE, REFSEQ

05 **New targeted callers for higher genotyping accuracy**
HBA 1/2, LPA and RH, SMN, CYP21A2 and CYP2D6

06 **Change in genome build**
from GrCh37 to GrCh38

07 **Deep intronic variant coverage Addition of spike-in probes**
for clinically relevant non-coding pathogenic and likely pathogenic variants

Use of additional Variant Callers :

LOFREQ* : Fast and sensitive variant-caller for inferring SNVs and Indels
MUTECT : Accurate and sensitive cancer mutation detection
PINDEL : Detection of breakpoints of large deletions
DELLY : Integrated structural variant (SV) prediction method

Variant Prediction Tools:

M-CAP | InMeRF | Mupro
i-mutant | Consurf | MutPred2
| REVEL | Alpha Missense