MEDGENOME | ACTIA Inherited Genetics

CAP ACCREDITED COLLEGE of AMERICAN PATHOLOGISTS

ExomeMAX

Coverage Matters

Clinical Presentation

Bridge The Gap

Molecular Diagnosis

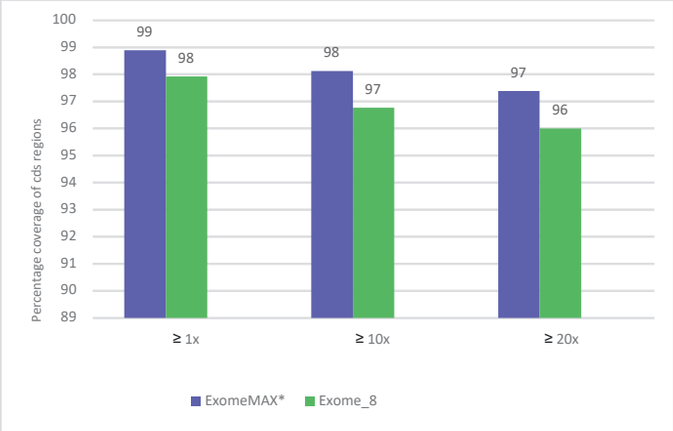
It is an inclusive design augmented by experts to provide better coverage of disease-associated genes to improve diagnostic outcomes. It provides a comprehensive genetic evaluation of inherited disease genes in this era of constantly evolving genotype-phenotype associations.

Features

- Coverage of alternate gene transcripts (MANE, GENCODE, REFSEQ)
- Coverage of Non-Coding Pathogenic variants (HGMD/ClinVar)
- 100% Coverage of Mitochondrial genome
- Variant analysis powered by ML algorithm and reviewed by certified Clinical Geneticist
- Extensive Validation, CAP Proficiency testing
- Specialised probes for superior detection of SNVs and CNVs
- Panel Size 47 Mb, Genes ≥ 20,000, Exons >2,10,000
- Industry leading average gene coverage of ~99%

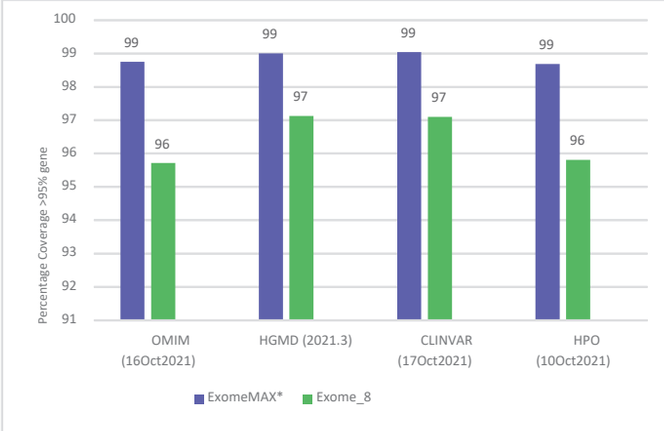
ACTGACTGACTGACTGACTGACTGACTGACTGACTGACTGACTGACTGACTGACTGACTGACTGACTGACTGACTG

At 20x coverage ExomeMAX has ~4.5% better coverage than off the shelf exomes



*Metrics based on 11-12Gb data
#Powered by Twist

Diagnostic yield can increase by 30% with improved coverage of exomes.



*Metrics based on 11-12Gb data

Indications for prescribing

- In cases where Clinical findings or family history is suggestive of an underlying genetic etiology
- To screen for genetically heterogeneous diseases
- To detect an undiagnosed genetic disease (diagnostic odyssey) in a patient
- To facilitate medical intervention and/or treatment
- To confirm the suspected genetic diagnosis
- To guide reproductive planning and assessment of recurrence risk
- To determine a prognosis (based on family history)

Validation

Requisite quality controls throughout the workflow from the laboratory sample processing till interpretation ensuring consistency, validity and accuracy

- Analytical sensitivity is NIST reference standard NA12878 is 99% SNPs and 96.2% for Indels.
- 100% disease causing variants (n= 50)
- CNV pipeline evaluated by orthogonally validated Copy number variants
- ML enabled analysis using MedGenome proprietary tool
- MedGenome database is backed by one of the largest South Asian Variant
- Results reviewed by certified clinical geneticist

ExomeMAX is clinically relevant for a range of disorders across

Genetic Counselling

FREE pre & post test genetic counselling with our expert and certified genetic counsellors

Talk to the Experts 1800 103 3691 diagnostics@medgenome.com www.medgenome.com