MEDGENOME | ACTIA Inherited Genetics

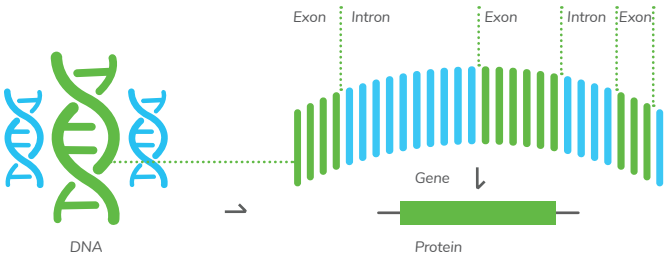
CAP ACCREDITED
COLLEGE of AMERICAN PATHOLOGISTS

Whole Exome Sequencing

Whole Exome Sequencing is a Next Generation Sequencing (NGS) based technique which enables the identification of disease causing variants in coding sequences.

~85% of known disease-causing mutations are in the coding sequences.

Validated bioinformatics & robust data interpretation system at MedGenome Labs provides an efficient platfom for clinical diagnosis.



Indications for Testing

- Confirm clinical diagnosis for genetically heterogeneous disorder
- Understand complex and atypical clinical presentation
- Identify etiology in inherited developmental disease/disorder with or without dysmorphism
- Confirm genetic diagnosis specially in case with inconclusive or negative results from disease specific panels
- Enable reproductive decisions, prognosis and disease management

Why MedGenome

- Clinical grade pipeline | Deep annotation | End to end automation | ML based variant
- Unmatched experience of 350,000+ exomes
- CAP accredited; Extensively validated pipeline
- South Asia largest database with 5,000,000+ variants
- Consistent proficiency testing through "External Quality Monitoring Programs"
- Two levels of analysis and review by clinical geneticist

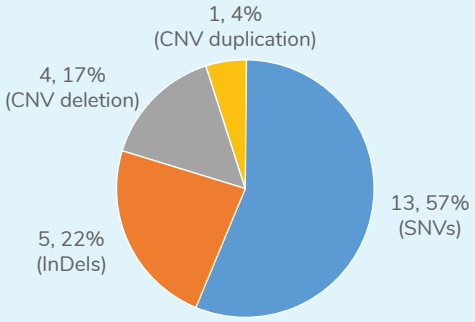
Validation

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

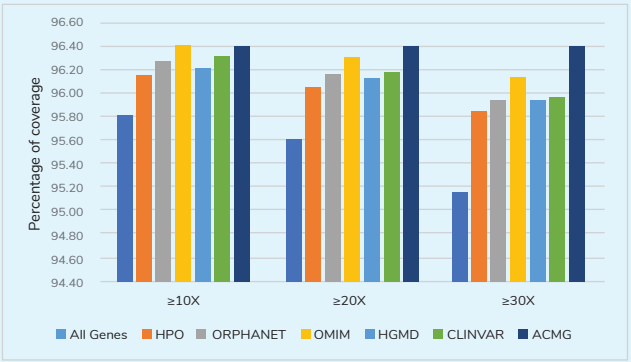
Analytical sensitivity on NIST reference standard NA12878 is >99% SNVs and >95% for InDels

CNV pipeline evaluated by orthogonally validated sample with known Copy number variants

100% concordance of disease causing variants (n=23)



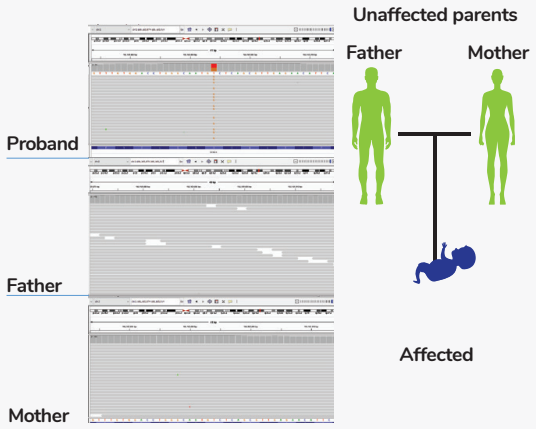
Twist exome assay enables deep coverage >95% of gene targets with 20x coverage.



TRIO-Exome

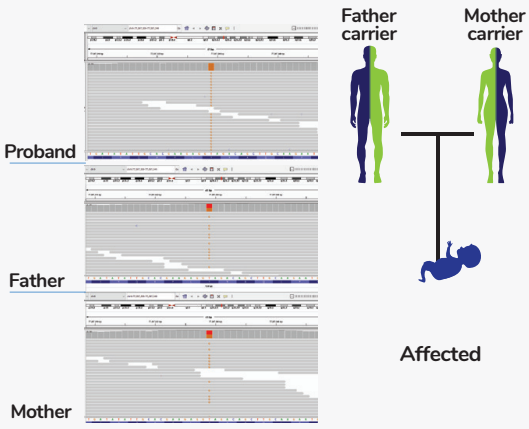
NGS based Trio Exome Analysis offers a powerful approach for the identification of causal mutations for inherited diseases including denovo variants.

Figure 1



Denovo Mutation in proband

Figure 2



The homozygous variant observed in the proband was detected in the heterozygous state in both the parents.

Note: Triplet repeat expansions, translocations cannot be detected by this methodology. Genetic changes present outside of the targeted region will not be detected

Test Sample Requirements

Individual samples/ trios/ families

Blood (3-5ml in EDTA tubes)
TAT : 21 Workinng Days

Test Requisition Form: Detailed clinical information and family history

Expert Genetic Counseling before and after the test is available for your patients.