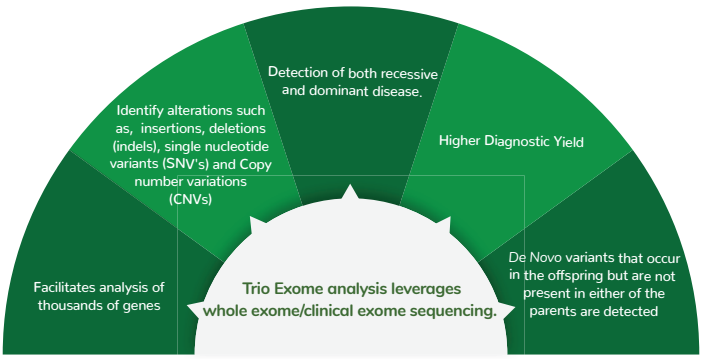
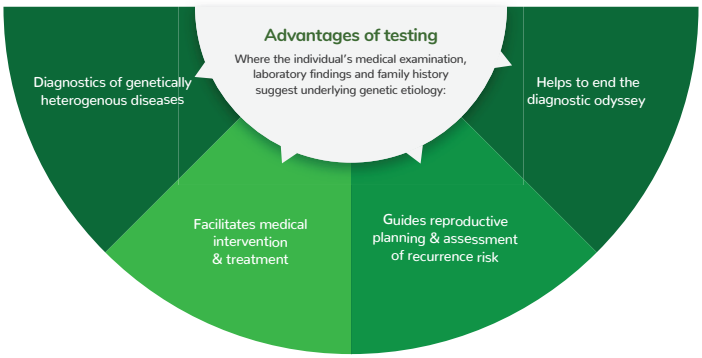
MEDGENOME | ACTIA

Trio Exome Analysis

A powerful approach for the identification of mutations for inherited diseases.





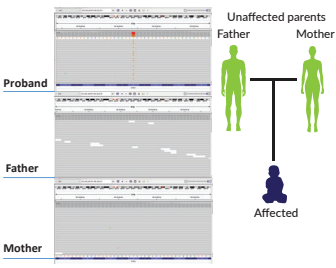
Test Features:

- Comprehensive QC including relatedness (Mendelian QC)
- Combined variant calling for increased accuracy of variant detection (in low coverage region)
- Better diagnostic yield compared to single proband analysis
- Ability of ascertain inheritance of clinically relevant variants i.e. compound heterozygous and *de novo* variants

	Clinical Exome (V3 -MedGenome)	*Whole Exome (Agilent SSV5)*
Panel Size	~29MB	~50MB
No of Genes	8342	22,713
Panel Coverage	80-100X	80-100X
Coverage at > 20X	>=95%	>=95%

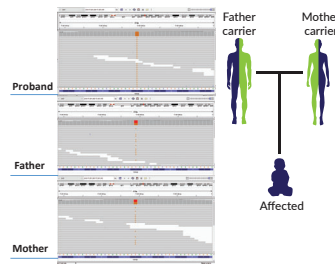
Examples:

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

MedGenome Offers	Sample Requirements	TAT
TRIO Analysis	Peripheral blood OR Purified genomic DNA	4 Weeks

Get in touch

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Locations

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