GPDx
Genomic Precision Diagnostic

Igenomix®
WITH SCIENCE ON YOUR SIDE

PRODUCT/CATEGORY	Technology	Description	Advantages	Disadvantages
1 Genome/Exome	Next Generation Sequencing (NGS)	• Process that determines the order of nucleotides, building blocks of DNA in an individual's genetic code. • Whole Genome Sequencing (WGS) sequences both the entire protein coding and the non-coding regions of the genome. • Whole Exome Sequencing (WES) sequences all the protein coding regions in the genome (exons).	• Cost-effective solution for the diagnosis of complex and unsolved cases • Helps determine whether a couple is at risk of having a child with a genetic condition as well as prognostic information regarding the disease • Help determine if a disease-causing variant is inherited and helping to prevent any further genetic disorder • Faster turnaround time for high sample volumes • Higher sensitivity to detect low-frequency variants	• Expensive equipment • Bioinformatic processing of data • Less cost-effective for sequencing low numbers of targets (1-20 targets) • Time-consuming for sequencing low number of targets (1-20 targets)
2 Chromosomal Microarray (CMA)	Chromosomal Microarray (CMA)	• Molecular cytogenetic method for the analysis of copy number variations (CNVs) • Gold standard for the detection of CNVs	• Detects structural aberrations by providing information on thousands of targets in a single experiement detecting copy number changes at the gene, chromosome and genome level. • Provides comprehensive genetic testing for the most commom chromosomal conditions as well as large number of severe genetic conditions not detected by traditional chromosome analysis.	• CMA cannot detect rearrangements (i.e balanced translocations, small deletions, sequence variants, mosaicisms etc)
3 Precision Panels	Next Generation Sequencing (NGS)	• Testing multiple genes associated with a particular disorder, thus, creating genetic panels based on WGS. • Panels include all relevant pathogenic and likely pathogenic variants within coding regions, regulatory sequences, and deep intronic regions described in Human Gene Mutation Database (HGMD).	• Allow the utilization of strong diagnostic hypothesis to reduce the cost while benefitting from the power and upside of WGS • Fast, thorough, and cost-effective diagnosis for patients with distinctive clinical features • Option of creating customed precision panels based on genetic demand of the patient	• Expensive equipment • Bioinformatic processing of data • Less cost-effective for sequencing low numbers of targets (1-20 targets) • Time-consuming for sequencing low number of targets (1-20 targets)
4 Gene Analysis	Multiplex ligation-dependent probe amplification (MLPA)	• Multiplex PCR method that detects abnormal copy numbers of up to 50 different genomic DNA or RNA sequences associated with a disease • MLPA can also detect DNA methylation changes (MS-MLPA)	• Most reliable and cost-effective method to detect known deletion/duplications and specific CNVs. • Is able to discern between point mutations, as well as duplication/deletion of genes • Small alterations to the MLPA protocol can allow for a variety of applications	• Is not able to detect large CNVs, sequence variants or mosaicisms • MLPA is very sensitive to impurities
	Expansion (EXP)	• Gold standard assessment for many repeat expansion diseases • PCR-based screening of repeat lengths	• Run test with fewer requirements (no controls, no size thresholds)	• Limited to only repeat expansions • Will eventually be substituted by WES and WGS
	NGS	"	"	"
	Sanger/MiniSeq sequencing (SANG)	• Also known as "chain termination methos" is a method for determining nucleotide sequences in DNA	• Fast, cost-effective sequencing for low number of targets (1-20 targets) • Verification sequencing for site-directed mutagenesis	• Low sensitivity • Low discovery power • Not as cost-effective for high number of targets (>20 targets) • Low scalability due to increasing sample input requirements