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MEDICAL

MeghaGen is a life sciences company providing both end-to-end and data analysis/archival solutions for all medical applications related to genome-wide screens.

As one of the world's leading companies with unique strength in data analysis, we not only deliver complete solutions for your genome-wide profiling requests, but will be your partner in making our findings meaningful to your practice. Through the impressive research achievements of our highly qualified staff, and our molecular diagnostic network, we are making huge contributions in the area of genome-wide screen in cancer, diabetes and other genetic disorders.

Meghagen's collaborative approach towards molecular diagnosis and reporting helps clinicians preempt diseases; offer better treatment options; help parents make informed life changing decisions; and avoid radical medical choices.

We believe that the best options for patient's quality of life is to leave no-stone-unturned. MeghaGen's cost effective genome-wide screens minimizes difficult/frantic medical choices. Our genome-wide screening services are marked by a special ethical responsibility and assurance of the highest molecular diagnostic standards for our customers.

Our goal is to become the trusted partner for clinical researchers and healthcare professionals in moving the spectacular achievement of sequencing technologies closer to personalized care through rigorous data analysis strategies.

GENOME-WIDE SCREEN IN DISEASES

As the cost of sequencing continues to drop, genome-wide screening of clinical samples is expected to become the norm. In other words, if cost is not a consideration, genome-wide screen should and would be made as routine as blood and urine test. Currently, many single gene-based genetic testing in cancer and other disorders is already proving to be important to patients in receiving the right treatment and to doctors in making informed medical decisions. Parallel to these tests, genome-wide screening for disease-specific variants are offering unprecedented opportunities in rare genetic disorders, in oncology, in diabetes and in patients who respond poorly to drugs.

From genome-wide screen to clinically actionable variants

- Step 1 3,000,000,000 base positions
- Step 2 578842 exomic mutation calls
- Step 3 69780 after a technical quality filter
- Step 4 2798 within the actionable gene loci
- Step 5 130 structure changing variants
- Step 6 12 not so far seen in normal people
- Step 7 2 actionable variants that are also reported among more than 1 million other cancer patients



Depending on clinical applications and cost considerations, genome-wide screening can be limited to somatic variants within tumor hotspots, germline mutations within known cancer-associated loci, structure-changing variants within a panel of more than hundred actionable genes and exome-wide coding variants.

The major challenge is genome-wide screen is making variant calls that are both disease-specific and actionable. With millions of SNPs, SNVs and indels scattered across the normal genomes, identification of disease-specific and actionable variants is like finding a needle in a jungle of normal variants. By annotating structure-changing variants within the exomes (protein coding regions) of cancer samples against fast growing database of more than 38 million normal variants and validating them against variants reported from exomes of more than 1 million cancer samples MeghaGen will filter out those potential disease causing variants that can be associated with treatment options.

Exomes are only 1.5% of the human genome. The impact of variants outside of exome on the expression levels and isoforms of actionable genes are significant. RNA-seq, ChIP-seq and methyl-seq are technologies that can help identify change in expression levels of actionable genes caused by both genetic and epigenetic changes. MeghaGen scientists are experts in the identification of genes that are differentially regulated compared to matched normal from the same patient in a cost effective fashion.

SERVICES OFFERED:

- Genome-wide identification and annotation of single nucleotide polymorphism (SNP), single nucleotide somatic variations (SNV), short insertion/deletion (indels) and other chromosomal abnormalities from targeted and whole exome sequencing efforts for using clinical applications in areas not limited to oncology, genetic disorders, diabetes, and heart disease.
- Disease-specific gene expression and splicing signatures from RNA-seq and reconciling these with the respective genetic variations from the same sample to improve the predictive success.
- Metagenomics characterization of microbial communities of medical importance not limited to gut, skin, cancer and other infectious disease tissues.
- Sequencing and analysis of human pathogens not limited to various strains of tuberculosis bacilli, HIV, and other opportunistic novel organisms.
- Epigenetic profiling of actionable genes.
- Data archival service for future retrieval and comparison to help our patients with new drugs discovered in the future and for comparing samples screened from the same family or patients with very similar cancer phenotype to gain better understanding of the mechanism.

