Center for Pediatric Genomic Medicine (CPGM) services

The CPGM is currently able to provide next generation sequencing and analytic services to investigators including sequence alignment and variant detection for whole genome (WGS), whole exome (WES), small gene panels and RNA sequencing. In addition, variant results can be characterized by the RUNES pipeline and viewed through the VIKING software developed by the Center.

All projects involving next generation sequencing (NGS) will include the option of receiving clinical-grade interpretations of samples according to the American College of Genetics and Genomics (ACMG). Other options include receiving raw data files, which includes BAM files of aligned sequences, VCF files of nucleotide variants, variant files (VCF and/or CSV) with RUNES annotations and VIKING/variant warehouse access.

CPGM sequencing, bioinformatic pipeline and interpretation workflow

All sequencing operations at the Center are performed in a custom-designed, CLIA-accredited laboratory. The CPGM currently has methods for DNA and RNA isolation from blood, tissue, Formaldehyde Fixed-Paraffin Embedded (FFPE) (DNA only), and buccal samples if previous isolation has not been performed. Pricing is roughly $600.00 for each RNA-seq or WES sample ($1800.00 for trio). The Center has five Illumina HiSeq and two MiSeq sequencers. Primary analysis is performed by Illumina software. Currently, sequences are aligned to the reference nuclear and mitochondrial genomes (GRCh37) with BWA or GSNAP (Genentech), and variants are identified and genotyped with the GATK.

The CPGM's standard bioinformatic pipeline includes: 1)RUNES (Rapid Understanding of Nucleotide variant Effect Software) for nucleotide variant annotation and 2) VIKING with SSAGA (Symptom- and sign-assisted genome analysis for enhanced variant analysis) for enhanced variant analysis. RUNES incorporates data from existing pathogenicity prediction software and several variant databases to assign a composite ACMG pathogenicity score. Annotated RUNES variants are then analyzed in VIKING. To facilitate variant analysis, VIKING allows dynamic sorting, selection and prioritization of displayed variants with a menu of filters. These include inheritance pattern, variant frequency, genotype, and the ACMG pathogenicity category. Additionally, the variants displayed can be changed by altering the SSAGA information. SSAGA is a clinicopathological correlation tool that maps a subject's clinical phenotype to genes previously associated with those phenotypic features.

RNA sequencing is performed using total stranded RNA and the Illumina sequencers above. The standard bioinformatic pipeline can be used for gene expression or variant calling.

Custom bioinformatics analysis services can be provided on a project-by-project basis for $45/hour. A minimum of 1 week of analyst time is suggested.

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