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DATA OFFICE FOR CLINICAL & TRANSLATIONAL RESEARCH

Targeted Sequencing in the Michigan Genomics Initiative Cohort

About this guide

This guide provides information about the MIPS targeted sequencing in the Michigan Genomics Initiative (MGI) cohort.

What variants were included in the sequencing panel?
What methods were used to sequence the specimen?
In what format is the data available?
How do I access the data?
How do I cite the data?
Where can I get help with genetic data?

High Level Summary

A custom targeted sequencing panel was designed for 151 genes using single molecule molecular inversion probes or smMIPS.

Example Use Case

Researchers could examine a specific gene variant rather than the whole genome.

What variants were included in the sequencing panel?	ABCA1, ACTA2, ACTC1, ACVR1, ADAMTSL4, ADH1B, AKT1, ALDH2, ANGPTL3, APC, APOA5, APOB, APOC3, APOE, APP, ARAP1, ATP7A, BRCA1, BRCA2, BTBD11, BTK, C19orf80, C2CD4A, C2CD4B, CACNA1S, CAMK1D, CBS, CHRNA5, CHSY1, COL3A1, COL5A1, COL5A2, COMT, CYP2A6, DAB2IP, DHDDS, DHX38, DNAH17, DNMT3A, DPT, DSC2, DSG2, DSP, EFEMP2, EGFL7, ELN, FAAH, FBLN4, FBLN5, FBN1, FBN2, FN1, FOXE3, GATA4, GATA5, GCH1, GCKR, GLA, GNB4, IGF2BP2, IPO8, IRX3, IRX5, JAZF1, KCNH2, KCNN1, KCNQ1, LDLR, LMNA, LOX, LOXL, LOXL2, LOXL3, LOXL4, LRP1, LRRK1, MAP3K4, MAS1, MAT2A, MATR3, MEN1, MFAP5, MLH1, MSH2, MSH6, MUTYH, MYBPC3, MYH11, MYH7, MYH9, MYL2, MYL3, MYLK, NF1, NF2, NKX2-5, NOTCH1, NPC1L1, OPRM1, PCSK9, PHACTR1, PKP2, PLOD1, PMS2, PNPLA3, PNPLA5, PRKAG2, PRKG1, PSEN1, PSEN2,, PTEN, RB1, RET, RPGRIP1L, RYR1, RYR2, SCN5A, SDHAF2, SDHB, SDHC, SDHD, SERPINA10, SERPINB2, SHC1, SKI, SLC2A10, SMAD3, SNAPIN, SORT1, STK11, TCF7L2, TGFB1, TGFB2, TGFB3, TGFBR1, TGFBR2, TM6SF2, TMEM43, TNNI3, TNNT2, TP53, TPM1, TRPV1, TSC1, TSC2, VEGFA, VHL, VPS13C, WT1, YY1AP1, ZBTB42
What methods were used to sequence the specimen?	A custom targeted sequencing panel was designed for 151 genes using single molecule molecular inversion probes or smMIPS. Coding exon coordinates were retrieved from the UCSC Genome Browser “knownGene” table (build GRCh37/hg19) and padded by 5 bp in each direction to include 5 splice sites. For each sample, approximately 9 ng of purified smMIPS probes were combined with 250 ng genomic DNA. The captured material was amplified by PCR using barcoded primers. The resulting PCR products were pooled for one lane of paired-end 150 bp sequencing on an Illumina HiSeq 4000 instrument at the University of Michigan Sequencing Core. Reads were aligned to the human genome reference (build GRCh37/hg19) using bwa mem and a custom pipeline (available at https://github.com/kitzmanlab/mimips) was used to remove smMIPS probe arm sequences and remove reads with duplicated molecular tags. Variant calling of MIPS sequencing results for both single nucleotide variants and insertions/deletions was performed using the GotCloud pipeline. An iterative filtering process was performed after variant calling to remove variants with a depth < 10, then samples with call rates < 0.6, followed by variants with a call rate < 0.8, and finally samples with call rates < 0.9.

In what format is the data available?	The data are available in BAM files (*.bam). These files are compressed binary versions of a tab-delimited text file that contains sequence alignment data.
How do I access the data?	Once the cohort is created through DataDirect, please put in a request for genetic data through the Precision Health Research Scientific Facilitators.
How do I cite the data?	Clinical Implications of Identifying Pathogenic Variants in Individuals with Thoracic Aortic Dissection (https://pubmed.ncbi.nlm.nih.gov/31211624/)
Where can I get help with genetic data?	Contact the Precision Health Research Scientific Facilitators at PHDataHelp@umich.edu