



High Throughput Genomics Core (HTGC)

The High Throughput Genomics Core (HTGC) is a CAP/CLIA-certified state of the art genomics facility located in the center of Shadyside in Pittsburgh, PA. The facility houses a high-throughput, clinical-grade, 9,000 ft² next generation sequencing (NGS) laboratory with an integrated bioinformatics cloud infrastructure. The HTGC staff includes two laboratory directors, laboratory staff, bioinformaticians, and IT support personnel.

The HTGC provides numerous laboratory and data analysis services to local research, commercial, and clinical interests. Our flexible, modular approach to project management allows samples to enter and leave the workflow at any stage, so investigators requisition only the services they require. Some clients complete highly customized portions of an experiment at their own or other facilities while realizing significant cost savings by completing sequencing and analysis at the HTGC. The HTGC works closely with UPMC in addition to other University of Pittsburgh's genetics "core" laboratories to facilitate inter-lab sample transfer and batching to maximize efficiency and minimize cost for these projects. Other clients utilize our end-to-end workflow beginning with nucleic acid extraction and ending with comprehensive bioinformatics analysis or clinical reporting, including customizations available throughout the process. In other cases, we generate clinical-grade data for downstream analysis and treatment decision-making by collaborators.

Executive Leadership

Anantha Shekhar, MD, PhD

Senior Vice Chancellor for the Health Sciences
Dean, John and Gertrude Petersen School of Medicine
University of Pittsburgh

Paul G. Wood, MS

Director for Core Facilities, OORHS
University of Pittsburgh

Mark W. Geraci, MD

Associate Vice Chancellor for Interdisciplinary Research,
Health Sciences
Vice Dean for Research, School of Medicine
Professor of Medicine
University of Pittsburgh

Laboratory Leadership

Fen Guo, PhD, FACMG, FCCMG

UPMC Clinical Genomics and High Throughput Genomics
Core Laboratory Director

Lindsey Kelly, PhD, Laboratory Manager

Dara Kozak, BS, Director of Operations

Gopi Vyas, MS, Bioinformatics Supervisor

Our Teams

In addition to the leadership team, the HTGC is powered by a team of molecular genomics specialists, bioinformaticians, and a software engineer.



Our Services

Tests*

We can perform any combination of the following:

1. DNA/RNA Extraction
 - From blood, saliva, buccal swab, fresh, frozen, or FFPE tissue, buffy coats, cell pellets or PBMC, stool, urine, lymphatic and cystic fluid. Other sample types may be available on request
2. Quantity, Concentration, and Quality Checks (extracted DNA/RNA, prepared libraries)
 - Using Fragment Analyzer, Qubit, Synergy, Tecan, or qPCR
3. RNA and DNA Library Preparation
 - We prepare mRNA, Total-RNA, miRNA, Whole Genome, and Whole Exome libraries
4. Next Generation Sequencing Services
 - We sequence various types of libraries including mRNA, Total-RNA, single cell-RNA, single cell-ATAC, 10x genomics speciality, Nanostring specialty, ATAC-seq, Amplicon, Whole Genome*, and Whole Exome* libraries within investigator-defined parameters and output targets
5. Pharmacogenomics*

In collaboration with the Pharmacogenomics Center of Excellence in the University of Pittsburgh School of Pharmacy, we can return the following from blood, saliva, buccal swabs, or extracted DNA sample types:

 - Research results spanning more than 4600 markers in nearly 1200 genes (returning raw CEL files and/or VCF from ThermoFisher's PharmacoScan Assay)
 - Clinical-grade reporting on a subset of validated, actionable genotypes for medication management and inclusion in the medical record
6. Axiom Microarray Services
 - We offer the Axiom PangenomiX and PangenomiX Plus array. The PangenomiX Plus array cover pathogenic variants and modules for PGx, neurological disorders, ancestry, SARS-CoV-2, and immunity panels; ACMG 73 update; HLA typing; copy number fixed regions; PGx content; ~800,000 markers
 - Our platforms are also able to run the previous releases: Axiom Precision Medicine Research Array (PMRA) & Axiom Precision Medicine Diversity Array (PMDA).

***Laboratory-Developed Test validated for clinical use available**

Tests under development

The following tests are currently being developed and we will be accepting samples for them soon!

1. Lipidomics (from plasma)
2. Metabolomics (from plasma and urine)
3. Metagenomics (from stool and saliva)
4. Targeted Proteomics (from plasma)
5. Illumina Protein Prep (from plasma)
6. TruSight Oncology 500
7. Infinium EPIC Methylation Array (using Illumina iScan System)

Bioinformatics and Data Analysis

We provide a range of custom and standard bioinformatics analyses including but not limited to:

1. Whole Exome or Genome Germline and Somatic/Tumor Variant Calling
2. Whole Genome Copy Number Variant and Tumor Purity
3. RNAseq Feature Counts with/without Variant Calling
4. RNAseq Differential Gene Expression
5. 10x Genomics Cellranger Primary Analysis

Raw or formatted sequencing data files (bcl, fastq, BAM, VCF and CEL) files for use in external analysis are also available for relevant test types.

Project Management

We support investigators with:

1. No-cost planning and setup consultations with our in-house experts
2. Translation of research findings to development of clinical testing
3. Ongoing support, troubleshooting, customization, and experimental design throughout the process
4. Validating outside or creating in-house custom workflows and analyses for novel projects
5. Clinical-grade reports appropriate for medical decision-making and inclusion in the medical record
6. Letters of Support and other grant/publication support documents as needed
7. Project-based invoicing to any funding source

Our Facility

Laboratory Space

The laboratory is subdivided into three separate rooms. First, the sample accession and management room contains 2 workstations to scan the barcodes of each sample into the laboratory information management system (Clarity LIMS by Illumina). Within the Clarity LIMS each sample will be assigned to the appropriate workflow and therefore can be tracked electronically throughout the entire laboratory and analysis process. The sample management room also contains one Vantage robot by Hamilton and one Tecan Fluent 780 for liquid handling, two Chemagic 360 instruments by Perkin Elmer for 96-well or 24-well plate-based DNA and RNA isolation, one Synergy by BioTek and one Qubit by Invitrogen for quantification, and one Fragment Analyzer by Agilent for measuring RNA quality.

Second, the library preparation and qPCR room contains two Hamilton Vantage robotics for automated library preparation, two Eppendorf epMotion 5075t NGS robotic handlers for automated library preparation and sample preparation for Mass Spec protocols, two Fragment Analyzers for library size and quality determination, one Tecan to measure library concentration, and one LightCycler by Roche for qPCR to measure library concentration as required, and two Chemical Fume Hoods.

Third, the sequencing room contains two NovaSeq X Plus, one Novaseq 6000, and one MiSeqDX by Illumina for sequencing. This room also holds the Biomek FXP liquid handler by Beckman Coulter for the automated processing of 96-well format ThermoFisher microarray plates and the GeneTitan by ThermoFisher for integrated hybridization, staining and scanning of the PharmacoScan and other array Axion varieties. It also contains one iScan System including a Tecan Freedom EVO 150 robotic handler, two Stellar Mass Spectrometers with Vanquish Neo UHPLC systems, and two Orbitrap Exploris 240 Mass Spectrometers with Vanquish Flex UHPLC systems.

The HTGC also contains ample storage for samples and reagents in 4°C, -20°C, and -80°C units; biosafety cabinet and dead air hoods, and other small lab equipment required for optimized high-throughput processing.

Equipment List

Equipment list:

- 3 – Vantage liquid handlers by Hamilton
- 2 – epMotion 5075t NGS by Eppendorf
- 1 – Chemagic 360 24-head automated nucleic acid extraction instrument by Perkin Elmer
- 1 – Chemagic 360 96-head automated nucleic acid extraction instrument by Perkin Elmer
- 1 – Synergy by BioTek
- 1 – Qubit by Invitrogen
- 3 – Fragment Analyzer by Agilent
- 1 – LightCyclers by Roche
- 1 – Tecan Infinite 200 Pro by Tecan
- 1 – Tecan Fluent 780 by Tecan
- 1 – Tecan Freedom EVO 150 by Tecan
- 2 – NovaSeq X Plus by Illumina
- 1 – Novaseq 6000 by Illumina
- 1 – MiSeqDX by Illumina
- 1 – Biomek FXP liquid handler by Beckman Coulter
- 1 – iScan System by Illumina
- 2 – Stellar Mass Spectrometers with Vanquish Neo UHPLC by ThermoFisher
- 2 – Orbitrap Exploris 240 Mass Spectrometer with Vanquish Flex UHPLC by ThermoFisher
- 1 – GeneTitan by ThermoFisher
- Various small lab equipment:
 - Plate and hand-held barcode scanners
 - Plate, large capacity, and micro-centrifuges
 - Vortexers
 - Label printers integrated to the laboratory information management system (Clarity LIMS by Illumina)
 - 4°C, -20°C, and -80°C units
 - Biosafety cabinet
 - Dead air hoods
 - Chemical fume hoods
 - Thermocyclers