NEW YORK GENOME CENTER® | Collaboration Research Discovery

Long Read Sequencing

NYGC offers long-read sequencing services on both Oxford Nanopore Technologies (ONT) PromethION 24 and PacBio Revio platforms. Both technologies generate long sequencing reads that enable researchers to resolve complex genomic features beyond the capabilities of traditional short-read sequencing, including structural variants, repetitive regions, haplotypes, and full-length transcripts.

The optimal platform depends on your research objectives, sample type, and desired analytical outcomes. NYGC's scientific project management team works with researchers to recommend the appropriate sequencing approach, balancing read length, accuracy, application requirements, and data analysis needs.

NYGC includes high molecular weight (HMW) nucleic acid extraction as part of our standard long-read sequencing workflows to support optimal read length and sequencing performance. Researchers may also submit compatible HMW DNA or RNA prepared in their own laboratory. For projects using client-provided HMW nucleic acid, pricing will be adjusted.

Oxford Nanopore Technologies (ONT) PromethION 24

ONT sequencing uses nanopore technology to directly read native DNA or RNA molecules as they pass through a nanopore, enabling highly flexible sequencing applications. ONT supports real-time data generation, ultra-long reads, direct RNA sequencing, and detection of modified bases such as DNA methylation.

ONT long-read sequencing is well suited for applications requiring:

- Ultra-long read lengths for complex genome characterization
- Direct RNA sequencing and full-length transcript analysis
- Native DNA analysis and modified base detection
- Flexible sequencing strategies with real-time data availability

Submission Requirements

The following are the standard minimum DNA input requirements for sequencing PacBio genomes. If you have low-input or low-quality samples, please contact your Project Manager before submitting, as alternative workflows may be available. Alternative submissions, such as cells, may also be supported.

Mammalian Whole Blood

- At least 2 mL of whole blood in EDTA (Lavender Top) tubes

Tissue

- A minimum of 2 x 2 x 2 mm of OCT-embedded or fresh frozen tissue is required (0.5 cm x 0.5 cm x 0.5 cm is optimal) or at least 10 scrolls of 5-10µm in thickness each

HMW DNA

- Minimum of **10 µg** of unamplified, RNase-treated, high molecular weight DNA
- Submitted in **50-100 µL** of EB (Elution Buffer)
- DNA should be extracted using the Qiagen MagAttract HMW DNA Kit or similar product

HMW RNA

- Minimum of **1 µg** of total RNA or **100 ng** poly-A RNA

Standard Analysis & Deliverables

Analysis includes:

- Base Calling performed using Dorado with the specific base calling mode (Fast, High Accuracy, or Super High Accuracy)
- Read alignment, retaining modified bases if requested
- Additional analysis dependent on assay type

Deliverables:

- Base called and unaligned BAM files
- Aligned read CRAM files
- Alignment QC reports
- Additional deliverables dependent on assay type

PacBio Revio

PacBio Revio generates highly accurate HiFi reads through circular consensus sequencing (CCS), combining long-read sequencing with high per-base accuracy. HiFi reads provide reliable characterization of complex genomic regions and enable highly accurate variant detection, genome assembly, and transcript isoform analysis.

PacBio Revio is well suited for applications requiring:

- High-accuracy long-read whole-genome sequencing
- Comprehensive structural variant detection
- Phasing and haplotype resolution

- De novo genome assembly
- Full-length transcript characterization through Iso-Seq and Kinnex workflows

Submission Requirements

The following are the standard minimum DNA input requirements for sequencing PacBio genomes. If you have low-input or low-quality samples, please contact your Project Manager before submitting, as alternative workflows may be available.

Mammalian Whole Blood

- At least 2mL of whole blood in EDTA (Lavender Top) tubes

Tissue

- A minimum of 2 x 2 x 2 mm of OCT-embedded or fresh frozen tissue is required (0.5 cm x 0.5 cm x 0.5 cm is optimal) or at least 10 scrolls of 5-10µm in thickness each

HMW DNA

- Minimum of **3 µg** of unamplified, RNase-treated, high molecular weight DNA
- Submitted in **10–50 µL** of EB (Elution Buffer)
- DNA concentration determined by **PicoGreen** (or equivalent fluorescence-based assay)
- DNA should be extracted using the Qiagen MagAttract HMW DNA Kit or similar product

HMW RNA

- Minimum of **1 µg** of total RNA

Standard Analysis & Deliverables

Analysis includes:

- Base Calling and Phasing, if applicable
- Read alignment, including available modified bases
- Additional analysis dependent on assay type

Deliverables:

- Base called BAM files
- Sequencing QC reports
- Additional deliverables dependent on assay type

Frequently Asked Questions – Long-Read Sequencing

What information is needed to start a project?

To begin planning a long-read project, we typically request:

- Number and type of samples
- Sample source and preservation method
- Sequencing Platform
- Desired sequencing coverage
- Required timeline
- Data delivery requirements
- Any planned downstream analyses

How are projects initiated?

Once project scope and service requirements have been finalized with your NYGC Project Manager, NYGC will provide a formal service quote outlining the agreed-upon deliverables, pricing, and project details. To begin project execution and submit samples, NYGC requires a signed copy of the quote and an approved purchase order (PO). For large-scale projects, additional discussions regarding customized terms and conditions may be accommodated.

What is the expected turnaround time?

All turnaround times can be found on our '[main research services page](#)'.

What is a standard submission size?

We are open to both small-scale and large-scale long-read projects. Volume discounts can apply.

How should samples be submitted?

For U.S.-based submissions, once a project has been quoted and a purchase order has been received, we provide NYGC-barcoded tubes in a matrix rack, an electronic sample manifest, and an overnight return shipping label for sample submission. Note that the provided tube orientation is critical, and tubes should not be rearranged.

Your Project Manager will coordinate international submissions, including shipping requirements and documentation.

Materials submitted for nucleic acid extraction, including blood, tissue, and cell samples, can be shipped directly to NYGC. Your Project Manager will provide the appropriate shipping address and an electronic sample manifest to complete prior to shipment. Please provide at least 24 hours' advance notice before shipping samples to ensure proper receipt and processing. To maintain sample integrity and ensure successful delivery, samples should arrive during NYGC business hours, as the laboratory is closed on weekends and holidays.

What DNA or RNA input requirements do you recommend?

We recommend submitting high-quality genomic DNA or RNA that meets our standard quantity and quality requirements. Specific requirements may vary depending on sample type and requested sequencing depth. Our team will review your sample information during project planning to confirm suitability before submission.

Will you check DNA/RNA quality and quantity before library preparation?

Yes. All projects include an initial quality control (QC) assessment to confirm DNA/RNA quantity and evaluate DNA/RNA quality. QC results, including DNA/RNA integrity metrics, will be shared with you for review before proceeding with library preparation.

What sequencing coverage options are available?

We offer a range of sequencing depths depending on study goals. Our team can help determine the appropriate coverage based on your research objectives, sample type, and downstream analysis needs.

What quality metrics will be provided?

We provide sequencing and QC metrics to help evaluate data quality, including metrics related to sequencing performance, alignment, coverage, and sample quality.

What species are supported?

Our standard analysis pipelines support human, mouse, and PDX samples. Library prep kits typically are tested only on mammalian samples. Projects on other species may be accepted and would require custom analysis.

How is data returned?

We support data delivery to a variety of client-managed endpoints and storage environments. Before delivery, we can perform a test transfer to confirm connectivity and ensure efficient transfer of large sequencing datasets.

What data formats are provided?

Data delivery formats can be customized based on project requirements. Common deliverables include standard alignment files and associated quality metrics. Additional analysis outputs can be discussed during project planning. Delivery of raw data types typically incurs an additional cost.

Do you provide secondary analysis?

Yes. Our standard sequencing services include secondary analysis options, as described in the Analysis and Deliverables sections above. These services provide processed data and analytical outputs designed to support downstream research needs. For projects requiring additional customization, our bioinformatics team offers consulting services to address specific

research questions, develop tailored analytical approaches, and generate publication-ready visualizations and reports.

For more collaborative engagements involving deeper scientific partnership, custom analyses, or method development, collaboration agreements may be established. Please contact our Project Management team at ProjectManagement@nygenome.org to discuss your project needs.

Can samples be submitted anonymously/de-identified?

Yes. **Samples submitted for research sequencing services must be de-identified prior to submission.** Sample identifiers should not contain personal health information (PHI) or other identifying information. As part of our sample intake process, submitted sample information is reviewed for compliance with these requirements. If identifying information is detected, the sample record will be flagged and the submitting party will be notified so that the information can be corrected. Any records containing unintended identifying information will be removed and handled according to established data management procedures.

For clinical sequencing services, please contact us at https://www.nygenome.org/clinical-genetics/.

How are samples tracked throughout the workflow?

NYGC uses a comprehensive laboratory information management system (LIMS) and integrated informatics infrastructure to provide secure, end-to-end tracking of every sample throughout the sequencing process. From initial sample receipt through library preparation, sequencing, quality control, and data delivery, each sample is tracked with complete process traceability. Our production laboratory has used the LIMS platform since 2015, with integrations across laboratory workflows, instrumentation, robotics, sequencing platforms, and reporting systems. This enables automated tracking of sample status, workflow progression, and quality metrics while supporting thousands of samples across hundreds of active projects. Sequencing operations are supported by additional monitoring and reporting tools that provide real-time visibility into sequencing performance, data generation, quality control metrics, and workflow status. Automated pipelines track key processing steps, including demultiplexing, alignment, quality assessment, and issue resolution, ensuring that potential challenges are identified and addressed promptly. Before data delivery, NYGC's Project Management and Computational Biology teams review sequencing performance, coverage metrics, and project-specific quality requirements to confirm that deliverables meet established standards. This integrated approach provides clients with confidence that their samples are handled securely, tracked throughout the workflow, and delivered with reliable, high-quality results.