スイーツセミナー1

次世代ゲノム医療の情報基盤：

インハウスNGSデータ管理とロングリードシーケンスの最前線

2025年
12月18日（木）15:55–16:55

会　場　第5会場（パシフィコ横浜 会議センター 3F 311+312）

座　長　上村 泰央（株式会社ジーンベイ）

共　催　株式会社 ジーンベイ

SS1-1

松本 直通 先生　横浜市立大学 遺伝学教室

Recent Topics in Comprehensive Rapid Variant Search Systems for In-House NGS Data and Long-Read Sequencing

Since 2009, our laboratory has consistently pursued the identification of causal variants in rare diseases through NGS analysis. Over this period, the accumulating exome data, the discovery of new disease-associated genes, and turnover among analysts have made it challenging to determine who, when, and how to analyze unsolved exome cases—posing a critical operational issue for our team.
To address these challenges, we introduced GeneBay's GenaGenomeManager (GGM), which has markedly improved our workflow. As of July 11, 2025, the system houses gVCF files from 18,696 samples (9,838 probands and 3,919 trios), enabling rapid generation of per-gene variant lists and candidate somatic variants with low variant allele frequencies from across the entire dataset. These capabilities have substantially accelerated both internal efforts and collaborative gene discovery research domestically and internationally. At the upcoming Sweets Seminar, we will showcase some of our findings achieved through GGM.
Moreover, since 2015, our laboratory has also employed long-read sequencing to investigate the genetic basis of rare diseases. To date, we have completed genome sequencing for over 1,500 cases, demonstrating its effectiveness in resolving cases that remained unsolved by short-read exome sequencing. Selected results from these analyses will also be presented.
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SS1-2

上村 泰央　株式会社ジーンベイ

GPU-Powered NGS Data Analysis and In-House Variant Database for Next-Generation Genomic Medicine

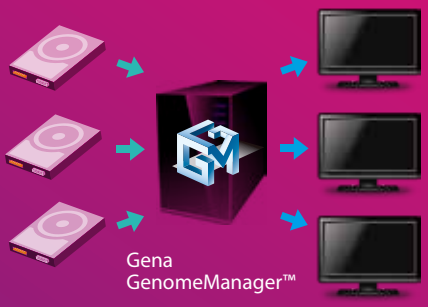
As next-generation sequencing (NGS) becomes integral to clinical practice, the demand for in-house data analysis, variant data accumulation, and integration with clinical information is rapidly increasing. To address these needs, we developed a GPU-equipped NGS data analysis system and a storage-based NGS analysis platform, Gena Genome Manager (GGM).
Leveraging NVIDIA Parabricks, our GPU system processes FASTQ to VCF files using the GATK best practices pipeline in as little as 40 minutes, enabling rapid and accurate variant calling. The resulting VCF files can be easily imported into GGM, allowing the construction of an in-house variant database. GGM further enables the integration of clinical metadata, facilitating comprehensive analysis to derive meaningful insights from genomic variants. This infrastructure empowers researchers to efficiently manage and interpret large-scale genomic data in a clinical context. We present this system as a robust solution for advancing precision medicine through integrated genomic and clinical data analysis, offering scalability and flexibility for next-generation medical advancements.

企業展示のお知らせ

1. ストレージ型 NGS 解析システム／GGM

増え続けるNGSデータの管理、閲覧、解析をトータルにサポート

- 1000サンプル規模で登録可能な、サーバー体型システム
- 将来的なデータ増加に備えて、拡張可能なストレージ
- ユーザー数制限なし、各ユーザのデスクトップコンピュータから何人でも使用可能
- ワークスペースごとに設定可能なユーザアクセス制限による、データの機密保持



2. GPU 搭載ナノポアデータ解析システム

ナノポアベースコール解析および各種解析パイプラインのデモ

- 解析用途に適したスペックのシステムを提供
- 面倒なデバイスドライバやソフトウェアの設定はおまかせ
- さまざまなオプションが選択でき、必要最小限の構成が選択可
- 解析手順書が付属しているため、直ちに解析の実行が可能

3. ナノポアシーケンス受託解析

Oxford Nanopore Technologies社の認定サービスプロバイダーとして、
最新ナノポアシーケンサーを用いた受託解析を提供

- PromethION 専用セルを最大48セル搭載
- 1セルあたりのスループット約30Gb～120Gb
- リード長：断片化の有無により異なる（数kb～数百kb）
- クオリティ：最頻値Q18～Q26（塩基配列精度98.5%～99.7%）

日程（予定）
12月18日（木）9:00－19:00
12月19日（金）9:00－19:00
12月20日（土）9:00－16:00

会場（予定）
パシフィコ横浜 会議センター
3F 301＋302

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