日本人類遺伝学会 第70回大会

InheriNext

InheriNextの評価と独自の解析プロセス LS 10

日時 ｜ 2025年12月19日（金）12:50～13:50

会場 ｜ 第6会場（パシフィコ横浜会議センター 3F 313＋314）

座長 ｜ 山本 俊至（東京女子医科大学大学院 医学研究科 先端生命医科学系専攻 遺伝子医学分野 / ゲノム診療科）
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共催 ｜ 日本人類遺伝学会第70回大会、Compass株式会社

Approach to Improve Accuracy and Efficiency in Variant Pathogenicity Evaluation: InheriNext Experience in Cardiomyopathy

演者 ｜ 宮下 洋平（国立循環器病研究センター　遺伝情報管理室）

Cardiomyopathy is a representative cardiovascular disease with a strong genetic predisposition. Numerous genes are implicated in disease onset, and many remain to be fully clarified. Consequently, comprehensive genomic analysis is rapidly expanding in both research and clinical practice. At the National Cerebral and Cardiovascular Center (NCVC), we have established clinical genetic testing for hypertrophic cardiomyopathy as well as the nationwide genome consortium study of cardiomyopathies (Grand-Star Next). However, the number of variants identified through large-scale genomic sequencing is vast, and manual curation by dedicated curators alone results in a heavy workload. Furthermore, clinical practice requires accurate and timely reporting, highlighting the need for novel approaches to support variant evaluation and streamline workflows.

We evaluated the feasibility of using the InheriNext system, developed by Compass Corporation, as a primary screening tool for cardiomyopathy cases. Its performance was compared with expert manual curation to determine whether efficiency and accuracy could be balanced.

This session will present validation results on the accuracy of variant evaluation using the InheriNext system. In addition, we will discuss how best to clarify the respective roles of automated systems and expert curators, and how to establish standardized workflows that ensure both efficiency and reliability in variant interpretation and reporting.

Streamlining Genetic Diagnosis of Neuromuscular Diseases: Efficient Variant Annotation via Lab-Industry Collaboration

演者 ｜ 吉岡 和香子（国立精神・神経医療研究センター 神経研究所 疾病研究第一部）

Neuromuscular disorders are often rare and require highly specialized expertise, yet diagnostic facilities remain limited. To address this, we provide central pathological evaluation for over 70% of muscle biopsies performed nationwide and serve as a hub for genetic diagnosis, covering most hereditary myopathies beyond those already reimbursed by the national insurance system. To date, we have established a custom-designed sequencing panel that covers all exons and exon–intron junctions of 115 genes in which pathogenic variants have been identified in at least two Japanese patients. In addition, we apply whole-genome sequencing not only to screen mutations in known disease genes but also to discover novel genes in unresolved cases.

A critical challenge in this work is the interpretation of variants, which requires integration of population data, computational predictions, and disease databases. Efficient management depends on well-designed annotation files, yet these must be continuously updated, representing a significant burden for our team that lacks dedicated bioinformaticians. Optimizing the process thus becomes essential under constrained human resources. In this luncheon seminar, we will present our collaborative efforts with Compass Corporation to streamline annotation file generation, our validation of the inheriNext scoring system, and perspectives on how such tools can further improve diagnostic workflows and accelerate discoveries in rare neuromuscular disease research in Japan.

＊今大会ではランチョンセミナーの事前予約をおこないます。事前予約は参加登録マイページより可能です。

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