

共催：日本人類遺伝学会第 66 回大会 第 28 回日本遺伝子診療学会大会 合同開催
イルミナ株式会社

ランチョンセミナー 3

Acute Care Genomics: Accelerating the diagnosis of rare diseases on a national scale

日時：2021 年 10 月 14 日（木） 12:10～13:10

会場：第 4 会場（パシフィコ横浜 会議センター 3F 「303」）
およびライブ配信

座長：慶應義塾大学医学部小児科

武内 俊樹 先生

演者：Associate Professor Sebastian Lunke

Victorian Clinical Genetics Services (VCGS),
Murdoch Children's Research Institute, Melbourne, Australia;
Australian Genomics Health Alliance, Australia

講演要旨：

Introduction: Ultra-rapid genomic testing is emerging as a key diagnostic tool for critically unwell children with suspected monogenic conditions. Health system wide implementation of ultra-rapid genomic testing requires scalable and reliable laboratory processes backed by robust clinical pathways.

Aims: To develop and implement a health-system wide ultra-rapid genomics pathway for critically ill children.

Methodology: Ultra-rapid testing was implemented at VCGS across successive cohorts totalling over 300 patients using a process of continuous review and improvement, stakeholder engagement, and education.

Results: Continuous improvement has informed changes to key laboratory processes, including sequencing approach, bioinformatics and analysis processes, a move to dedicated analysis teams of scientists and clinical geneticists, as well as of out-of-hours sample processing. These revised processes have been applied to a total of over 300 cases, with an average TAT of under 3 calendar days (range 2-5 days), using both exome and genome sequencing. Backed by whole of system change, the implemented model meets demand from over a dozen neonatal and paediatric intensive care units, is scalable to process over 500 cases per year in our laboratory, and is being adopted by other Australian diagnostic laboratories.

Conclusion: Implementation of ultra-rapid genomic testing in existing diagnostic laboratory environments is feasible at a national scale but requires whole of system change and is critically dependent, close clinical integration and adequate resourcing.

《ご注意》

本セミナーはハイブリッド方式（現地+ライブ配信）にて開催いたします。当日、密を避ける観点から会場での参加登録は
ございません。必ず事前に学会 HP からオンライン参加登録をお願いいたします。

<https://www.jshg-jsgdt2021.org/online.html>

※整理券の配布はございません。

パシフィコ横浜 会議センター 3F 301 + 302 にてブースも出展していますので、ぜひお立ち寄りください。

■ セミナーに関するお問合せ先 contactJPN@illumina.com

イルミナ株式会社 〒108-0014 東京都港区芝 5-36-7 三田ベルジュビル 22 階 jp.illumina.com

illumina®