

Optical genome mapping (OGM) and long-read sequencing (LRS) to resolve findings in leukemia research



September 17, 2024, 1-2 PM CET



Live Webinar

Join us for a live webinar on September 17, 2024, to hear from Eddy de Boer from the University Medical Centre Groningen, Netherlands, on how optical genome mapping and long-read sequencing are helping to resolve findings in leukemia analysis.

'Leukemias are genetically heterogeneous, and their analysis, therefore, includes various classical methods. Optical genome mapping (OGM) may replace these as it detects different types of structural aberrations simultaneously and additionally detects much smaller aberrations (500 bp vs 5–10 Mb with karyotyping). However, its resolution may still be too low to define relevance of aberrations when they are located between two OGM labels or when labels are not distinct enough. Cas9-directed long-read sequencing (LRS) has the potential to confirm new OGM findings that may not be completely resolved. We confirm LRS was able to show OGM calls were located between 0.2 and 5.5 kb of the OGM-estimated breakpoints, confirming the high reliability of OGM. Furthermore, we show examples of redefinition of aberrations between labels that enable judgment of medical relevance. Our results suggest that Cas9-directed LRS can be a relevant and flexible secondary technique in assessment workflows, including OGM'.



Speaker

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