**Presentation title:**

Reveal the hidden corners of the genome with optical genome mapping technology - sharing our insights from the Czech Republic

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Chromosomal aberrations play a crucial role in the risk stratification of hematological malignancies. Identifying these aberrations accurately allows for more precise risk assessments, facilitating better prognostic predictions and treatment planning. This can lead to improved patient management strategies, including the selection of appropriate therapies and monitoring protocols.

Optical genome mapping (OGM) is a cutting-edge technology developed for genome-wide detection of structural variants (SVs) as well as copy number variations (CNVs). OGM aims to address the limitations of existing cytogenetic techniques offering a higher resolution than karyotyping, allowing whole-genome analysis, unlike FISH, and the detection of balanced chromosomal abnormalities missed by chromosomal microarray analysis (CMA).

IAB laboratories, providing OGM services, in collaboration with clinical institutions, evaluated the outcomes of conventional cytogenomic techniques such as FISH, karyotyping, and CMA, as well as NGS, alongside OGM analysis. We focused on the ability of OGM to enhance cytogenomic diagnostics and perhaps even replace routinely used techniques including karyotyping, FISH, and CMA. Our study cohort comprised 10 patients with acute myeloid leukemia (AML), 10 with multiple myeloma (MM), and 10 with chronic lymphocytic leukemia (CLL).

OGM has effectively identified the majority of the cytogenetic abnormalities seen in routine cytogenetics including aneuploidies, hyperdiploidies, and structural variants. OGM also detected balanced translocations identifying partners of driver genes not routinely sought by other techniques. Additionally, OGM has proven its capability in uncovering additional cytogenetic abnormalities, particularly complex rearrangements. OGM missed cytogenetic abnormalities in minor subclones (VAF < 5%), and those having breakpoints located in areas poorly covered by OGM (centromeric and telomeric regions). OGM has demonstrated the ability to detect additional clinically relevant SVs missed by standard of care methods, owing to its significantly higher sensitivity and resolution.

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**Biography**

Regina Bezdekova Fillerova spearheads the research and development activities at the Institute of Applied Biotechnologies aimed at advancing personalized medicine and improving patient outcomes.

With over 15 years of experience in academia and industry, Dr. Fillerova has made significant contributions to the field of genetics, particularly in the translation of research findings into practical diagnostic solutions. Her innovative work has led to the creation of several diagnostic kits.

Dr.Fillerova holds a Ph.D. in Molecular Genetics from Palacky University, where her research focused on the genetic predispositions of infectious diseases in cooperation with Orthopedic and Pulmonary clinics. She has authored numerous peer-reviewed publications.

In the last five years, she has been intensively devoted to the optimisation and implementation of the method of optical genome mapping into clinical practice, especially in the field of hemato-oncology and postnatal genetics.