Genomic structural variants detection

All genome structural variants are no longer an issue with advanced Optical Genome Mapping (OGM).

Introduction

Structural variants (SVs) are an important source of genetic variation in the human genome, and they are involved in multiple diseases, including cancer and developmental disorders. Optical Genome Mapping (OGM) detects all classes of structural variants ranging from 500bp to whole chromosomes level, including rare variants down to 1% allele fraction.

With years of active user experience, IAB provides a complete sample-to-result solution for SV's detection powered by the Bionano OGM product line.

Method

Over the last few years, OGM established itself as a trusted technology for SV detection in many severe oncological or rare genetic diseases as it proved to be over 99% concordant with traditional techniques like karyotyping, FISH or microarrays. Clinicians all around the globe are now using OGM to diagnose haematological malignancies, solid tumours, Homologous Recombination Deficiency (HRD), or g as a sole solution instead of the traditional approaches.

A single Saphyr system allows for simultaneous high-coverage analysis of 6 samples. The subsequent analysis allows for the sensitive detection of insertions, deletions, inversions, translocations, duplications, copy number variations or chromosomal aberrations, even in highly mosaic samples. As a cherry on top of the pie, Bionano provides software NxClinical for efficient data analysis integration of OGM, microarrays and NGS outputs.

Conclusion

In collaboration with Bionano, IAB affords an effective solution for detecting genomic structural variants of any kind. Optical Genome Mapping is the future that you can meet at IAB.

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