



PRINCIPE FELIPE

CENTRO DE INVESTIGACION

---

## Beyond NGS: Bionano Genome Mapping Reveals Structural Variation in Human Cancer and Genetic Disease

---

**Date:** Friday, September 15<sup>h</sup>, 2017 – 12:30h

**Location:** Salón de Actos, Centro de Investigación Príncipe Felipe

**Contact:** [events@cipf.es](mailto:events@cipf.es)

The majority of many complex genomes are made up of repetitive and regulatory elements. Short read sequencing, while very powerful, is unable to map these highly repetitive sequences. As a result, critical information about the genome may be missed when analysis relies on NGS technologies alone. Bionano Genomics' platform for whole genome mapping allows for extremely long read data, resulting in unmatched sensitivity and specificity to detect large structural variation.

Our de novo maps can resolve complex repetitive regions, identify Copy Number Variations, and elucidate genome-wide large structural variation like balanced/unbalanced translocations, inversions, and large indels. Specific examples will be presented of how Bionano's platform is helping provide a molecular diagnosis for patients with Duchenne Muscular Dystrophy, identify potential causative variants for patients with undiagnosed genetic disorders, and how it has been applied to discover large, potentially oncogenic, genomic rearrangements in prostate cancer and leukemia that were missed by NGS alone.

Speaker: Dr. Sven Bocklandt, Senior Application Specialist from Bionano Genomics