Accelerating the Transformation in How We Analyze the Human Genome

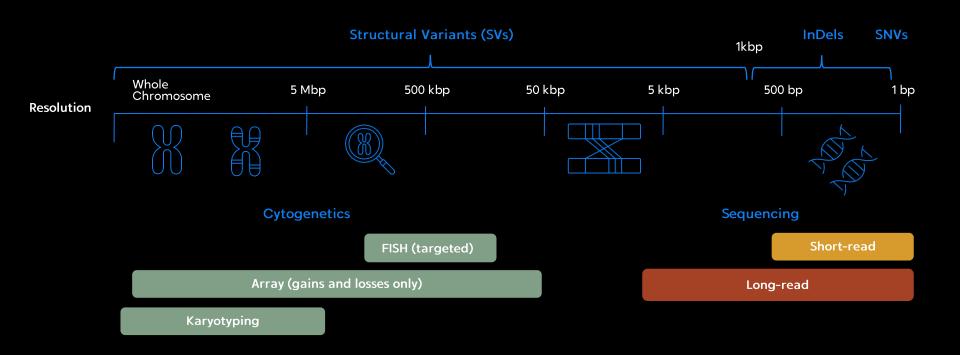
Damla Senol Cali, Ph.D.
Staff Software Engineer, Hardware Acceleration
dsenolcali@bionano.com

BIO-Arch Workshop @ RECOMB'23
April 14, 2023
Istanbul, Turkey





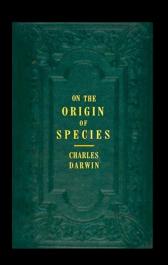
The Unmet Need in the Genome Continuum



Two Decades of NGS Have Not Closed the Gap



NGS is Unable to Reliably Tackle SVs for Fundamental Reasons

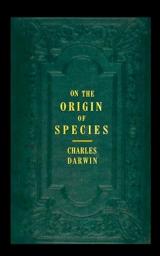


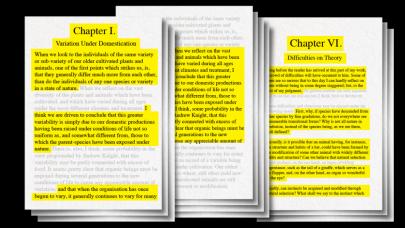






The OGM Difference is Context from Ultra Long Reads





Chapter I. Variation Under Domestication When we look to the individuals of the same variety or sub-variety of our older cultivated plants and animals, one of the first points which strikes us, is, that they generally differ much more from each other, than do the individuals of any one species or variety in a state of nature. When we reflect on the vast diversity of the plants and animals which have been cultivated, and which have varied during all ages under the most different climates and treatment, I think we are driven to conclude that this greater variability is simply due to our domestic productions having been raised under conditions of life not so uniform as, and somewhat different from, those to which the parent-species have been exposed.

under nature. There is, also, I think, some probability in the

view propounded by Andrew Knight, that this variability

may be partly connected with excess of food. It seems

pretty clear that organic beings must be exposed during

any appreciable amount of variation; and that when the

to vary for many generations. No case is on record of a

several generations to the new conditions of life to cause

organisation has once begun to vary, it generally continues

variable being ceasing to be variable under cultivation. Our

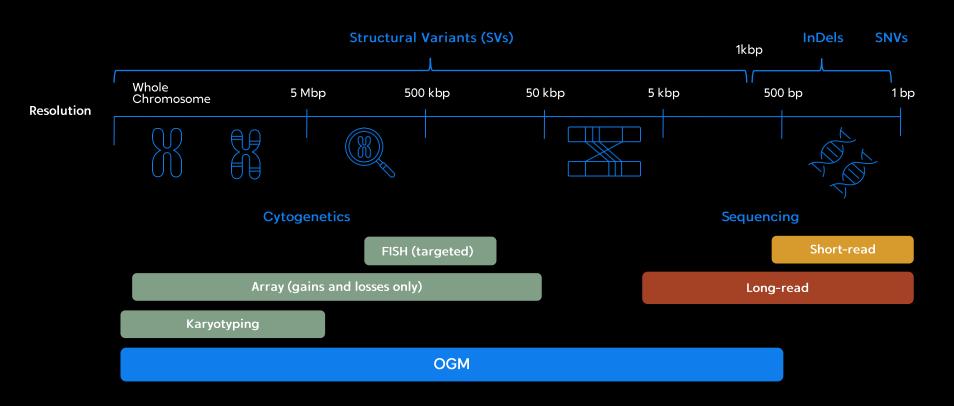
oldest cultivated plants, such as wheat, still often yield new

varieties: our oldest domesticated animals are still capable

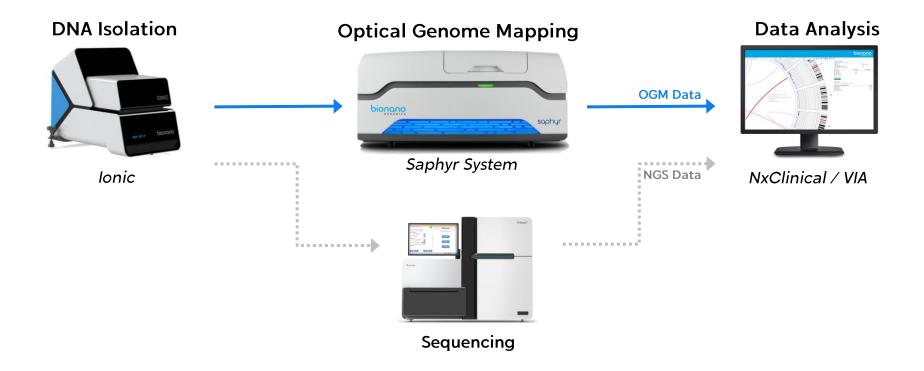
of rapid improvement or modification.

It has been disputed at what period of life the causes of variability, whatever they may be, generally act; whether during the early or late period of development of the embryo, or at the instant of conception. Geoffroy St. Hilaire's experiments show that unnatural treatment of the embryo causes monstrosities; and monstrosities cannot be separated by any clear line of distinction from mere variations But I am strongly inclined to suspect that the most frequent cause of variability may be attributed to the male and female reproductive elements having been affected prior to the act of conception. Several reasons make me believe in this; but the chief one is the remarkable effect which confinement or cultivation has on the functions of the reproductive system; this system appearing to be far more susceptible than any other part of the organisation, to the action of any change in the conditions of life. Nothing is more easy than to tame an animal, and few things more difficult than to get it to breed freely under confinement, even in the many cases when the male and female unite. How many animals there are which will not breed though living long under not very close confinement in their native country! This is generally attributed to vitiated instincts; but how many cultivated plants display the utmost vigour, and yet rarely or never seed! In some few such cases it

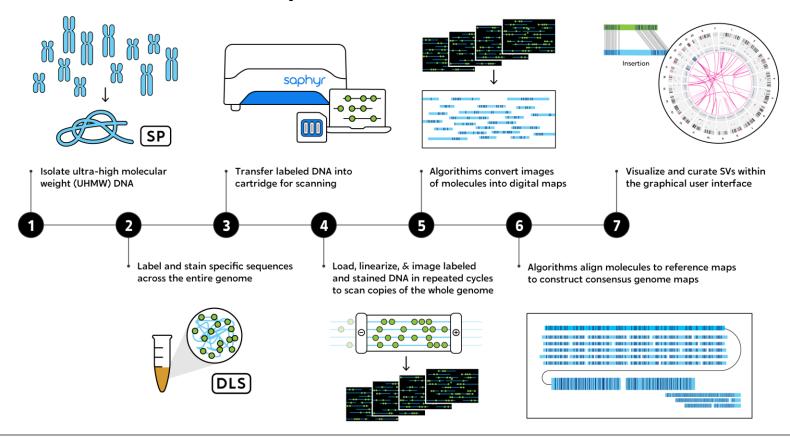
We Believe Optical Genome Mapping (OGM) Closes the Gap



Transforming Genomic Analysis with OGM + NGS



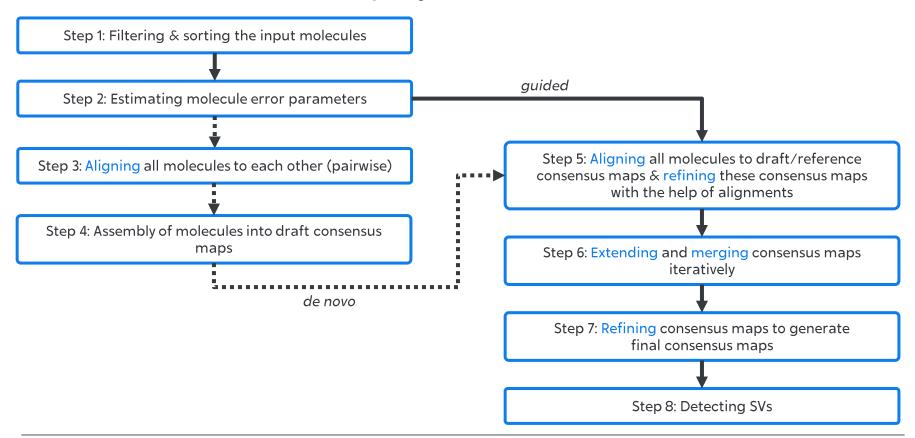
The OGM Workflow is Sample to Answer for SV Detection





Saphyr® System for Structural Variation Detection | Bionano

De Novo & Guided Assembly Pipelines with OGM Data



Future Evolution in How the World Sees the Genome

Enhancing throughput, usability, time to results and analysis integration across genomics applications



Enhancing Throughput

Sample prep kits yielding cleaner DNA and enhanced stability



Simplified Sample Prep

Enhanced labeling kits + SW to accelerate analysis



Time to Result

The power of NVIDIA in OGM



Analysis Integration

SW that integrates OGM, NGS, and CMA and speeds interpretation

Future Evolution in How the World Sees the Genome

Enhancing throughput, usability, time to results and analysis integration across genomics applications



Enhancing Throughput

Sample prep kits yielding cleaner DNA and enhanced stability



Simplified Sample Prep

Enhanced labeling kits + SW to accelerate analysis



Time to Result

The power of NVIDIA in OGM



Analysis Integration

SW that integrates OGM, NGS, and CMA and speeds interpretation

High Throughput Saphyr

Addressing the needs of high-volume labs and low-cost regions



The next step in the evolution of the OGM workflow:

- Up to 96 cancer or 338 constitutional samples per week
- Designed to scale with multiple systems orchestrated for higher throughputs -"workcell"
- Compatible with current Bionano reagents and software

Future Evolution in How the World Sees the Genome

Enhancing throughput, usability, time to results and analysis integration across genomics applications



Enhancing Throughput

Sample prep kits yielding cleaner DNA and enhanced stability



Simplified Sample Prep

Enhanced labeling kits + SW to accelerate analysis



Time to Result

The power of NVIDIA in OGM



Analysis Integration

SW that integrates OGM, NGS, and CMA and speeds interpretation

Bionano's Next-Generation Compute Solution

- Bionano's Optical Genome Mapping (OGM) is a scalable genome analysis workflow elevating cancer and complex disease research
- Decreasing the cost of genome analysis is the main goal to be accessible to more people and get answers quickly and accurately
- Our next-generation compute solution:



Provides vital data processing capabilities for our high-throughput
OGM instruments



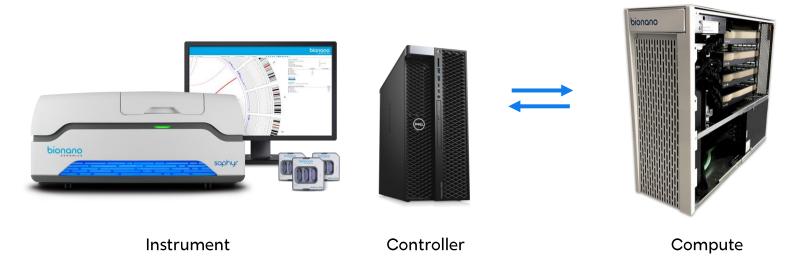
Significantly reduces time and cost of the analysis of OGM data to meet the higher throughput need



Accommodates clinical testing needs at scale by reducing the pipeline processing time by ~8×

Overview of our New Compute Solution

- Next-generation NVIDIA technology to accelerate OGM analysis
- A standalone workstation with multiple NVIDIA GPUs networked to the instrument controller
- An NVIDIA CUDA optimized Bionano Solve pipeline that accelerates alignment, refinement, and structural variation detection



Bionano and NVIDIA Supercharging OGM Analysis



- Bionano, the pioneer of OGM solutions, and NVIDIA, the pioneer of accelerated computing, combine their industry leading technologies for the next-generation accelerated OGM analysis solution
- NVIDIA's RTX 6000 Ada Generation enables HPC power into the lab, and ready for the cloud!
- NVIDIA GPUs provide:
 - Thousands of parallel CUDA cores enable extreme parallelism
 - Efficient floating point-based calculations in parallel with high processing power
 - Much higher performance per \$ (vs. CPU)
 - Vertical and horizontal scaling opportunities with either additional workstations or datacenter solutions with higher count of GPUs

We are accelerating the transformation in how we analyze the human genome!



Bionano & NVIDIA:

Accelerating Analysis for Fast Time to Results



Technological solution to support higher throughput



New high-performance algorithms from Bionano



Powered by NVIDIA RTX™ 6000 Ada Generation GPUs



Analysis of highly complex cancer whole genomes in less than 2 hours



Workflow tailored for a small lab and IT footprint

We are Hiring!

Senior Software Engineer - Hardware Acceleration

At Bionano, we are committed to unlocking understanding of genome biology to advance the promise of genomics in areas including cancer and human disease, agricultural bioengineering and genome discovery. Our optical genome mapping and analysis tools help researchers see true genome structure to fill in what's missing from sequencing-based data.

At Bionano, we are invested in the success of our customers and users around the world, and are dedicated to supporting them with the tools, resources and support they need to achieve their goals and make a real impact on improving quality of life for all.

Apply Now

Application Deadline

May 23, 2023

Department

Systems Dev

Employment Type

Full Tim

Location

SD - San Diego

Workplace type

Onsite

Watch for More: Bionano Workshop @ AGBT'23



Thank you.

bionano

Accelerating the Transformation in How We Analyze the Human Genome

Damla Senol Cali, Ph.D.
Staff Software Engineer, Hardware Acceleration
dsenolcali@bionano.com

BIO-Arch Workshop @ RECOMB'23
April 14, 2023
Istanbul, Turkey



