

GO EXPONENTIAL

Accelerate the pace of discovery with the power of Linked-Reads

In a world of incremental improvements, true breakthroughs are born from massive leaps forward. The genomics community is not about shifting paradigms but shattering them. With Linked-Read sequencing data, we can achieve a comprehensive understanding of genomic variation. Power your next discovery with Chromium™ Solutions that uncover the genome and exome data you've been missing. Now, you can resolve ambiguous single nucleotide variants, obtain phasing and haplotype information, identify structural variants, and assemble genomes without breaking a sweat.

Biology is challenging. Research is a race. Get there faster.
Learn more at go.10xgenomics.com/linked-reads



#GoExponential

Biology is complex. But within that complexity lies answers that can help you solve the world's biggest challenges. As members of the scientific community, we are driven to unlock the potential of genomic discovery. It's a drive shared by everyone who shatters convention to uncover exponential possibility.

10x Genomics exists to take your thinking to the next level through the development of novel genomics applications. 10x GemCode™ Technology integrates innovative microfluidics, chemistry, and informatics to enable the discovery of genomic data on a massive scale. Our Chromium™ System and Solutions for single cell, V(D)J, genome, exome, and *de novo* assembly, each with its own custom-built software suite, provide comprehensive approaches that amplify your ability to push the boundaries of biological research.

From phased genomic structural variation studies and single-cell gene expression analysis, to immune repertoire profiling and beyond, there are no limits to what we can discover together.

#GoExponential at 10xGenomics.com



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LINKED-READS

FIND THE ANSWER TO WHAT'S BEEN MISSING



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LINKED-READS

FIND THE ANSWER TO WHAT'S BEEN MISSING

Organisms and biology are immensely complex, and to truly understand disease mechanisms, heritability, potential diagnoses, and development of therapies, we need access to more genomic information. We build upon the existing next generation sequencing framework with Linked-Reads by constructing long-range information from short-reads to provide access to hidden and inaccessible information. With Linked-Reads, researchers now have the power to resolve ambiguous single nucleotide variants (SNVs), provide phasing and haplotype information, identify structural variants (SVs), and assemble genomes without the need for a reference sequence.

What are Linked-Reads?

Linked-Reads are a new sequencing data type that provide long-range information from short-read sequencing data. Linked-Read generation starts out by partitioning high molecular weight genomic DNA (HMW-gDNA) with barcoded gel beads, primers, and sequencing adapters to generate gel beads in emulsion (GEMs) that act as micro-reaction chambers. Isothermal incubations create short, barcoded fragments which, once sequenced, can be linked back to their original parent molecule—hence, Linked-Reads. This gives context to where the short-reads originated from in the genome and allows for the reconstruction of the original long input DNA.



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1

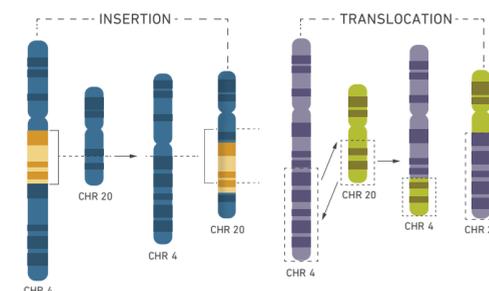
INTERROGATE UNMAPPABLE REGIONS

Single nucleotide variants potentially affect an individual's susceptibility to disease and response to treatment. Using Linked-Reads, researchers can obtain long-range information from short-read sequences, lending resolving power to previously inaccessible variants. An important example is detecting whether an SNV occurs in a medically relevant gene or in a paralogous pseudogene.

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FIND STRUCTURAL VARIANTS

Genomic SVs range in size from small insertions and deletions to larger genomic rearrangements like chromosomal inversions and gene duplications.¹ The ability to reliably detect clinically actionable SVs has the potential to unlock new therapeutic targets and may help advance the way we treat certain diseases, such as cancer.² With Linked-Reads, novel algorithms based on barcode-overlap and physical barcode-coverage facilitate the accurate and reliable detection of SVs across megabases of DNA.



GLOSSARY

Short-read sequencing: generating numerous short (50-150 bases) DNA fragments
Long-read sequencing: generating longer (multiple kilobases) DNA fragments to assemble a longer contiguous sequence
Linked-Reads: short-reads tagged with molecular barcodes based on origin DNA fragment (multiple megabases)
Haplotype: a group of genes within an organism inherited from a single parent
Paralog: genes that are sequentially similar, having descended from a common ancestral sequence, but perform different functions
Pseudogene: a DNA segment sequentially similar to a known gene but possessing suboptimal functionality to varying degrees

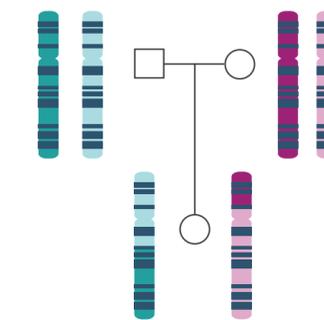
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RESOLVE HAPLOTYPES

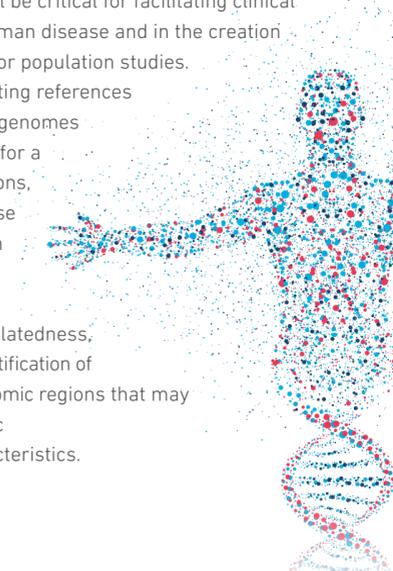
Given the diploid nature of the human genome, identifying changes that occur on maternal and paternal chromosomes is critical for understanding gene expression changes in genetic disease research, especially for autosomal recessive inherited disorders.³ Inherited variants can be present in *cis* or *trans*, and if functionally linked, may manifest in human disease. Linked-Reads enable researchers to phase variants⁴ and facilitate large-scale haplotype reconstruction,⁵ which may improve our understanding of genetic disease.



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EASILY ASSEMBLE GENOMES

Linked-Reads enable *de novo* assembly independent of a reference sequence, allowing for deeper studies of organisms and individuals without reference bias or contrived "consensus" sequences.⁶ The creation of true diploid *de novo* assemblies will be critical for facilitating clinical research of human disease and in the creation of references for population studies. Improving existing references in non-human genomes is also needed for a variety of reasons, including for use in conservation biology and establishing evolutionary relatedness, as well as identification of conserved genomic regions that may explain specific species' characteristics.



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