Xromate®

Simplify your copy number variations (CNVs) analysis

Xromate™ CNV-seq

Background

Karyotyping and chromosomal microarray analysis (CMA) are widely used for detecting copy number variations (CNVs) in prenatal diagnosis. In recent years, advancements in next-generation sequencing (NGS) technology have led to the development of CNV-seq, an efficient CNVs analysis method utilizing low-coverage whole-genome sequencing. Compared to CMA, CNV-seq provides a higher detection yield, requires less initial DNA, and is more applicable to low-level mosaicism. [1-2]

Parameters	Description
Technology	Low-coverage whole-genome sequencing, single-end
Platform	Illumina or Salus SBS sequencing system
Sample type	gDNA from peripheral blood, chorionic villus, amniotic fluid, cord blood/tissue, product of conception, etc.
Analysis offered	Genetic disorders caused by chromosomal aneuploidies and CNVs larger than 100 kb in size with mosaicism

Xromate™ CNV-seq

Xromate™ CNV-seq utilizes low-coverage whole-genome sequencing to analyze CNVs. It is widely used for prenatal diagnostics, miscarriage analysis, and investigating genetic causes in patients. Since 2014, Xromate™ has processed over 300,000 cases, with dozens of institutions implementing it in their laboratories.

EZ-GALO™ - Rapid PCR-Free Library

Featuring proprietary EZ-GALO™ technology, Xromate™ CNV-seq utilizes a streamlined single-tube reaction (enzymatic digestion, repair, and ligation into a single reaction tube) to minimize human error arising from solution transfers. Its PCR-free design avoids amplification bias, ensuring exceptional reproducibility and eliminating the diagnostic 'grey zone' for clear, definitive results.

Rapid and Efficient Workflow

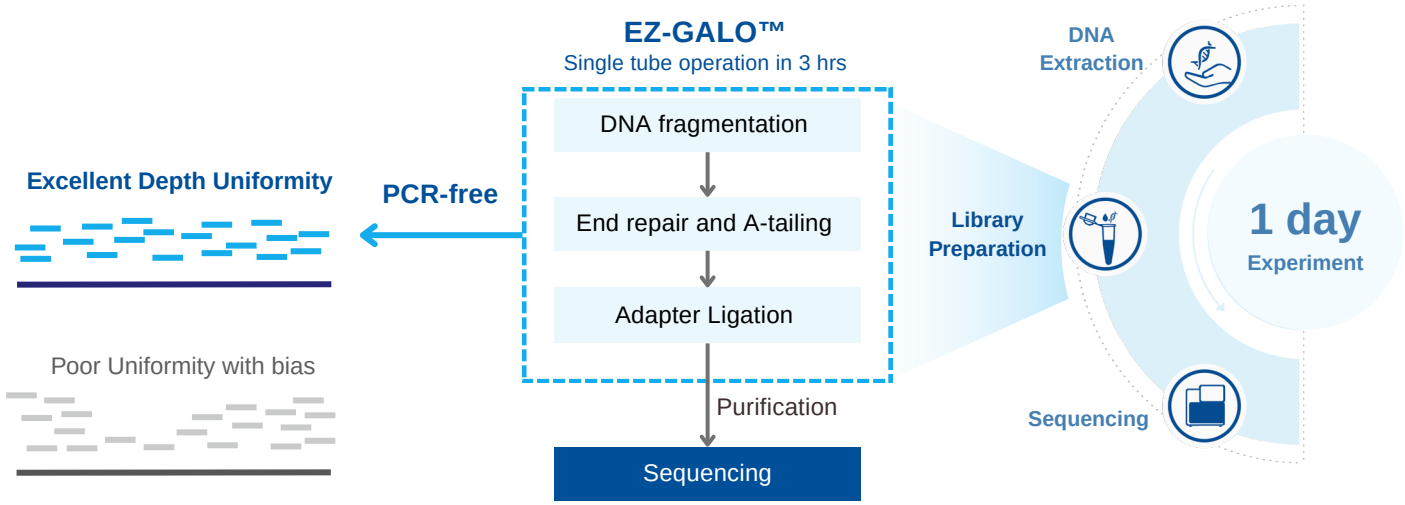
Powered by patented technology: EZ-GALO™ — Rapid PCR-free library, and CNVisi™— Integrated analysis and reporting system

Reliable Results

- Minimized amplification bias with PCR-free library
- ~ 93% of sequencing data are Q30 [3]
- High sensitivity for low-level aneuploidy (> 10%)

Practical

- Requires only 10 ng of initial DNA — ideal for low-yield prenatal samples
- Compatible with mixed sequencing batches with other testing services



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CNVisi™- Integrated Analysis and Reporting System

Copy Number Variation Integrated System of Interpretation (CNVisi™) is an all-in-one system for quality control, data analysis, annotation, interpretation, and reporting, providing an efficient solution for rapid report management.

Accurate

Utilizing information from over 20 public databases and our internal CNVs database of million Chinese individuals, CNVisi™ automatic interpretation achieves 99.6% pathogenic classification accuracy [4]

Time Saving

CNVisi™ provides clear annotation and evidence based on ACMG guidelines. It reduces manual effort by over 90% in reporting time and simplifies interpretation complexity

Efficient

Trained on 200,000+ CNV reports, CNVisi™ helps draft report language to further cut manual work

An Ideal Prenatal Diagnosis Strategy: OeXome™ + Xromate™

With a low initial DNA quantity requirement, Xromate™ CNV-seq can be combined with OeXome™ whole exome sequencing (WES) using just 60–90 ng of DNA in one sample. [5] Compared to the sequential prenatal diagnosis workflow (karyotyping, CMA, and WES), the OeXome™ + Xromate™ strategy effectively reduces total turnaround time, facilitating early intervention with comprehensive detection in a single step.

In a cohort study [5], couples with singleton fetuses displaying structural anomalies or increased nuchal translucency (NT) via ultrasound were recruited for Xromate™ CNV-seq and OeXome™ WES using products of conception (POC) samples. Among 959 trios successfully tested:

- 227 were identified with a pathogenic or likely pathogenic alteration (CNVs or variants), of which 84.14% were *de novo*
- 99 fetuses were carrying CNVs
- 118 fetuses were carrying variants
- Remaining were identified as double diagnoses
- Overall diagnostic rate for combined strategy was 23.67%

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