Xromate®

Simplified your copy number variations (CNVs) analysis

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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	Whole-genome sequencing, single-end
Platform	illumina or Salus sequencer
Sample type	gDNA, peripheral blood, chorionic villus, amniotic fluid, cord blood/tissue, product of conception, and other
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

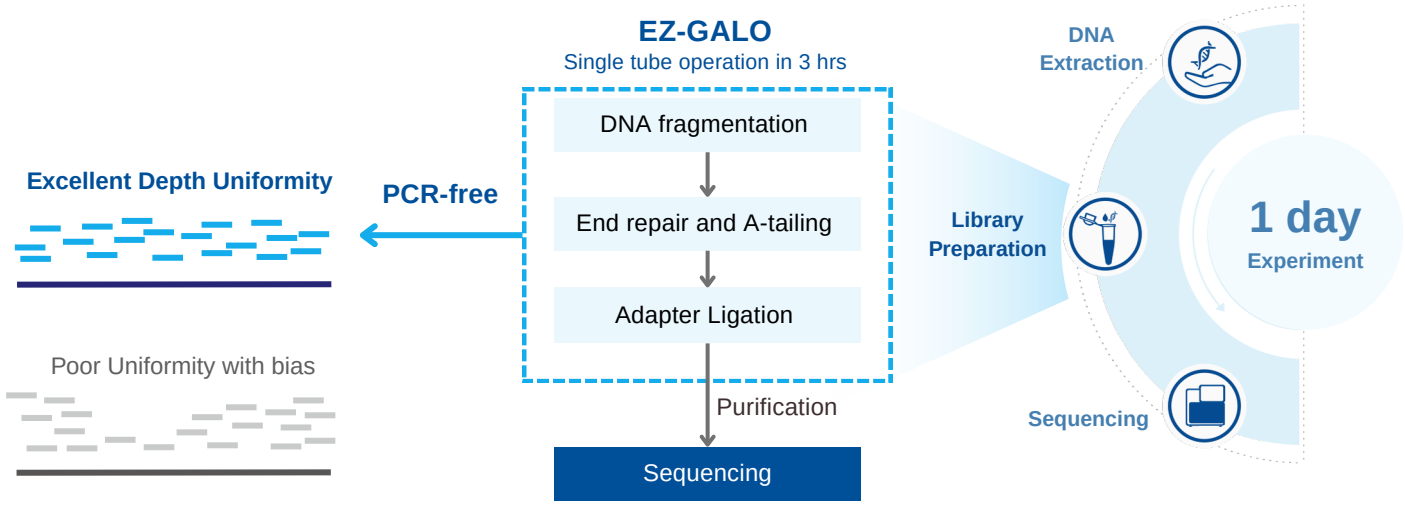
Powered by patented technology: EZ-GALO — Rapid PCR-free library, and CNVisi — Integrated analysis and reporting system

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
- Mixed sequencing batch 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

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

A 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 [4], 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 with double diagnoses
- Overall diagnostic rate for combined strategy was 23.67%

References:
[1] Ma N, Xi H, Chen J, et al. Integrated CNV-seq, karyotyping and SNP-array analyses for effective prenatal diagnosis of chromosomal mosaicism. *BMC Med Genomics*. 2021;14(1):56.
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[3] Zhou X, Chen X, Jiang Y, et al. A Rapid PCR-Free Next-Generation Sequencing Method for the Detection of Copy Number Variations in Prenatal Samples. *Life (Basel)*. 2021;11(2):98.
[4]. Chen S, Liu C, Luan X, et al. Application and clinical utility assessment of natural language processing-based software for copy-number variants interpretation. *J Transl Med*. 2025;23(1):1052.
[5 Chen X, Jiang Y, Chen R, et al. Clinical efficiency of simultaneous CNV-seq and whole-exome sequencing for testing fetal structural anomalies. *J Transl Med*. 2022;20(1):10.

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