

LONG-READ WHOLE GENOME SEQUENCING LRS



Genetics and
Rare Diseases
Diagnosis Center

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Traditional genetic tests (such as exome sequencing or short-read genome sequencing) analyze DNA by reading it in small fragments and then assembling these pieces. In the first step, DNA is fragmented into small segments. These fragments are individually sequenced using specialized instruments, and computer programs reconstruct the data by aligning each fragment to its correct position. The results are then compared with a reference human genome to identify variants associated with disease. This method allows us to reliably access the majority of genetic information. Exome sequencing in particular has provided diagnostic opportunities to millions of individuals worldwide. However, these techniques may not fully detect large structural variations, repetitive regions, or complex variants within the DNA. For this reason, Long-Read Whole Genome Sequencing (LRS) has begun to be used, especially in patients who remain undiagnosed with other methods.

Another major advantage of this technology is its ability to detect certain epigenetic disorders and methylation abnormalities. In addition to capturing all the information obtained through short-read whole genome sequencing, long-read sequencing can also identify chromosomal rearrangements (such as translocations, inversions, and deletions), repeat expansion disorders, enable accurate HLA interpretation, allow haplotype analysis (distinguishing the genes inherited separately from the mother and the father), and detect methylation abnormalities. As accessibility to this technology increases in the coming years, it is highly likely that it will replace many existing sequencing technologies.



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For detailed information
about Long-Read Whole
Genome Sequencing,
please scan the QR code.

