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BGI achieves next-gen sequencing analysis of FFPE DNA as low as 200 ng

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BGI, the world's largest genomics organization, reported that it can use next-generation sequencing to analyze DNA as low as 200 ng from formalin-fixed paraffin-embedded (FFPE) samples. This advancement enables researchers to easily identify the genetic details and pathology mechanism of FFPE disease samples, especially for some rare tumors, with higher accuracy and reliability than existing techniques.

FFPE samples are common biological materials for disease diagnoses and scientific research. Because FFPE tissue samples may be stored indefinitely at room temperature, and nucleic acids (both DNA and RNA) may be recovered after decades from the original fixation, they have become an important resource for historical studies in medicine. There are millions of FFPE samples stored worldwide containing significant genetic information for disease and medical research.

With the rapid development of genomics, the ability to sequence FFPE samples opens up large tissue collections from clinical trials for genetic analysis that could help researchers identify novel variations that are linked to disease development. However, during the sample preparation and storage process, formaldehyde can induce modification of nucleotide molecules, such as DNA damage, DNA-protein cross-links (DPC), among others. This hampers further application and development of sequencing in exploring the genetic characteristics of diseases.

Recently, BGI researchers have made a breakthrough on FFPE DNA sequencing by achieving optimization of FFPE DNA library

construction with total DNA degraded to as low as 200 ng. "This is a critical step toward better decoding the potential genetic information of FFPE samples," said Xun Zhao, staff scientist from BGI Department of DNA Sequencing. "In order to fully understand the heterogeneity and special properties of FFPE samples, we hope to conduct more FFPE DNA sequencing projects with collaborators worldwide to further enhance and standardize our technique."

"We expect that next-generation sequencing technologies with FFPE samples could substantially facilitate our understanding of undefined pathological mechanisms and broaden our insights in biomedical research," added Zhao. "This also strengthens the confidence of researchers in pharmaceutical and disease areas, especially when samples are limited."

Provided by BGI Shenzhen