

BGI develops whole exome sequencing analysis of FFPE DNA samples to boost biomedicine

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BGI Tech Solutions announced today that they have achieved whole exome sequencing analysis of total degraded DNA as low as 200 ng from formalin fixed paraffin embedded (FFPE) samples. This advancement enables researchers to efficiently uncover the genetic information from FFPE disease samples such as cancers and infectious diseases, with the advantages of high reliability, accuracy and fast turnaround time.

FFPE samples are the most common biological materials for disease diagnoses and clinical studies. Especially in cancer research, millions of FFPE archival [cancer tissue](#) samples provide an enormous and invaluable repository of information, which hold a wealth of data for the discovery of biomarkers, drug development as well as diseases diagnosis and treatment.

However, during the FFPE sample preparation and storage process, the effect of formaldehyde on [nucleic acids](#) is detrimental, which can induce modification of nucleotide molecules, such as [DNA damage](#), DNA-protein cross-links (DPC), among others. This may lay problems for researchers to get enough high-quality DNA from these FFPE samples to comprehensively explore the genetic characteristics of diseases, especially for some rare tumors.

FFPE samples are a unique sample type with a lot of challenges, and

researchers from BGI Tech have optimized the [DNA extraction](#), library construction and sequencing pipelines of FFPE [DNA samples](#). At present, DNA as low as 200 ng from FFPE samples can be used for whole exome sequencing. To insure the accuracy and quality of sequencing, researchers evaluated the FFPE DNA sequencing results and demonstrated that FFPE exome sequencing could maintain the equivalent accuracy and reliability with the normal DNA sample sequencing.

It is reported that ~85% of genetic diseases are related with exome variations. Whole exome sequencing is a robust innovative technique that selectively sequences the coding regions of a genome and can be used to identify novel genes associated with rare and [common diseases](#) such as cancer, diabetes, and obesity. However, currently traditional exome sequencing has higher requirement for the quality and the amount of input of DNA samples. Zhao Lin, Director of Products R&D Department of BGI Tech, said, "Our whole exome sequencing technology with FFPE DNA sample is an important step toward better and quickly decoding the genetic information underlying FFPE diseases samples. I believe this advancement will strengthen the confidence of researchers in pharmaceutical and disease areas, especially when samples are limited. In order to accelerate biomedical research, we expect to conduct more FFPE sequencing projects with collaborators worldwide."

Provided by BGI Shenzhen

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