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PHARMACEUTICAL INDUSTRY STANDARD OF THE PEOPLE'S REPUBLIC OF CHINA

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Gene mutation in tumor tissue detection kit (high-throughput sequencing)

肿瘤组织基因突变检测试剂盒(高通量测序法)

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Foreword

This document is in accordance with the provisions of GB/T 1.1-2020 "Guidelines for standardization work Part

1.Structure and drafting rules for standardization documents" Drafting is required. Please note that some of the contents of this document may involve patents. The issuing organization of this document does not assume the responsibility for identifying patents. This document is proposed by the State Food and Drug Administration. This document is under the jurisdiction of the Medical High-throughput Sequencing Standardization Technology Management Unit. This document was drafted by: Beijing Jiyinga Medical Testing Laboratory Co., Ltd., China Food and Drug Inspection Institute, Beijing Medical Devices Medical Equipment Inspection Institute (Beijing Medical Biological Protection Equipment Inspection and Research Center), Peking Union Medical College Hospital, Cancer Hospital of Chinese Academy of Medical Sciences, Affiliated Cancer Hospital of Nanjing University, Nanjing Shihe Gene Biotechnology Co., Ltd., Zhenyue Biotechnology Jiangsu Co., Ltd., Xiamen Feishuo Biotechnology Biotechnology Co., Ltd., Shanghai Sildi Biomedical Technology Co., Ltd., and Beijing Youxun Medical Equipment Co., Ltd. The main drafters of this document are. Yi Xin, Jia Zheng, Wang Ruixia, Liang Zhiyong, Ying Jianming, Zhou Xiaoyan, Shao Yang, Zheng Shan, Chen Yan, Guan Li, Shan Guangyu, Yi Yuting, Chu Yuxing, Yuan Fuqiang, Yu Ting, Qu Shoufang, Huang Jie.

The tumor tissue gene mutation detection kit (high-throughput sequencing method) in this document refers to a method for converting nucleic acid fragments into a sequenceable library. The kit mainly comprises deoxyribonucleoside triphosphates, ligase, polymerase, linker, primers, probes and the like. It is important to emphasize that an IVD test system is a combination of components that complete all stages from sample processing to final result reporting. For example, the tumor tissue gene mutation detection (high-throughput sequencing) detection system includes not only the kits referred to in this document, but also nucleic acid When designing, developing and verifying extraction kits, gene

sequencers, sequencing reagents, data analysis software, etc., complete and definite The detection system is analyzed and the performance is evaluated to complete the evaluation of each part of the detection system. In the process of clinical application, with the development of technology and the deepening of clinical research, there are more and more database construction methods, data analysis software, databases, etc. We suggest that developers take the initiative to update the products on the market based on their in-depth understanding of product design, production process and quality control. Continuous improvement and innovation of products. According to the relevant requirements of in vitro diagnostic reagent production quality management, when the above changes may affect product safety When evaluating the risks that may be caused by the changes, and taking measures to reduce the risks to an acceptable level when necessary, the relevant Regulatory requirements. This document is based on the application and validation data of high-throughput sequencing technology and is intended for manufacturers of in vitro diagnostic reagents and medical testing laboratories. etc. for use. Tumor tissue gene mutation detection kit (High-throughput sequencing)

1 Scope

YY/T 1946 is the product standard for the tumour tissue mutation panel by high-throughput sequencing, the NGS kit that drives targeted therapy selection in oncology. It sets the terms and definitions for these kits, specifies the requirements, the labelling and the accompanying information, and describes the corresponding test methods. An NGS panel is the hardest kind of IVD to standardise, because the product is a wet chemistry kit and a bioinformatics pipeline together, and the result is a variant list whose accuracy depends on both. Fixing the requirements at kit level is how a Chinese reviewer can compare panels that differ in gene content. For a manufacturer registering a tumour NGS panel in China, this is the standard the analytical validation is measured against.

This document defines the terms and definitions of the tumor tissue gene mutation detection kit (high-throughput sequencing method) (hereinafter referred to as the "kit"). It specifies relevant requirements, labels and instructions for use as well as packaging, transportation and storage, and describes the corresponding test methods. This document applies to kits that use high-throughput sequencing based on probe capture or multiplex PCR for the detection of tumors. Single nucleotide variants (SNVs), insertion/deletion variants (Indels), Copy number variations (CNAs), gene fusions, tumor mutation burden (TMB), and/or microsatellite instability (MSI). The kit is also suitable for paired detection of tumor tissue and control samples. This document does not apply to kits that use whole exome sequencing and single molecule sequencing technology to detect tumor gene mutations.

Note. The control sample may be whole blood or adjacent tissue sample, which is mainly used to distinguish germline mutations in tumor FFPE tissue sample testing.

2 Normative references

The contents of the following documents constitute the essential clauses of this document through normative references in this document. For referenced documents without a date, only the version corresponding to that date applies to this document; for referenced documents without a date, the latest version (including all amendments) applies to This document.

GB/T 191 Pictorial markings for packaging, storage and transportation

GB/T 29791.2 Information provided by the manufacturer of in vitro diagnostic medical devices (labeling) Part

3 Terms and definitions

The following terms and definitions apply to this document.

3.1 High-throughput sequencing Different from traditional dideoxy (Sanger) sequencing, a technology that can perform parallel sequencing of a large number of nucleic acid molecules at one time, Usually one sequencing reaction can produce no less than 100Mb of sequencing data. [Source: GB/T 40664-2021, 3.8]

3.2 Single-nucleotide variants; SNVs A type of variation caused by the substitution of a single nucleotide base at the same position in the genome.

3.3 Insertion and deletion alterations; Indels A type of variation that occurs when one or more bases are inserted or deleted in the genome.

Note. Usually refers to 1 to 50 bases.