ICS 110
CCS C44

YY

PHARMACEUTICAL INDUSTRY STANDARD OF THE PEOPLE'S REPUBLIC OF CHINA

YY/T 1968-2025

Software of human embryo preimplantation genetic testing for chromosome aneuploidy analysis

胚胎植入前染色体非整倍体分析软件

Issued on: June 18, 2025 **Implemented on: July 1, 2026**

Issued by: National Medical Products Administration

Table of Contents

Foreword

This document complies with the provisions of GB/T 1.1-2020 "Standardization Work Guidelines Part

1.Structure and Drafting Rules of Standardization Documents". Drafting. Please note that some content in this document may involve patents. The issuing organization of this document assumes no responsibility for identifying patents. This document was proposed by the National Medical Products Administration. This document is under the jurisdiction of the unit responsible for the standardization of medical high-throughput sequencing technology. This document was drafted by: Shandong University, Suzhou Beikang Medical Device Co., Ltd., Beijing Zhongyi Kangwei Medical Device Co., Ltd., and BGI Genomics. Biotechnology (Wuhan) Co., Ltd., Suzhou Yuanyi Medical Technology Co., Ltd., Renji Hospital Affiliated to Shanghai Jiao Tong University School of Medicine, and Sun Yat-sen University Affiliated Hospital It belongs to the Sixth Hospital and the China National Institutes for Food and Drug Control. The main drafters of this document are. Gao Yuan, Kong Lingyin, Fei Jia, Xiang Jiale, Ma Jinlong, Sun Yun, Liang Xiaoyan, Zhang Chao, Qu Shoufang, Zou Yang, and Gao Ming. Gao Xuan and Chen Zijiang.

Currently, several preimplantation genetic testing kits for aneuploidy and their accompanying analysis software have obtained medical device certification in the domestic market. Medical device registration certificates, and all comply with the relevant laws and regulations of local and national medical device software and preimplantation genetic diagnosis of aneuploidy. The analytical software and its supporting reagents have high performance requirements for analytical algorithms and software, and a large amount of clinical data has been accumulated, fully validating the software's safety. Sexuality and effectiveness. The field of assisted reproduction is becoming increasingly mature, but there is still no matching analysis software in China for preimplantation genetic testing (PGT) for aneuploidy. Standards for this component, as an essential part of the entire detection system for in vitro diagnostic reagents, require the formulation of standards for preimplantation genetic diagnosis (PGD) aneuploidy. Analyze recommended industry standards for software to guide software development, maintenance, and production management to meet registration regulations and supporting requirements. The

performance requirements of reagents for analytical software are outlined to further standardize the industry's overall requirements for preimplantation genetic testing (PGT) chromosomal aneuploidy analysis software. This is provided for reference by various companies. Preimplantation chromosome aneuploidy analysis software

1 Scope

YY/T 1968 is the standard for the software that decides which embryo gets transferred. It specifies the requirements for preimplantation genetic testing software used for chromosome aneuploidy analysis, known as PGT-A, and describes the corresponding test methods. It applies to the analysis of low-depth high-throughput sequencing data from preimplantation embryo biopsies, the workflow that turns a few cells into a call on whether an embryo carries the right number of chromosomes. The stakes make the software unusual among IVD tools: the output is not a diagnosis to be confirmed later but a selection decision that is acted on immediately and cannot be revisited. For a manufacturer of PGT analysis software seeking Chinese registration, or a laboratory validating a pipeline, this is the standard the algorithm performance evidence is measured against.

This document specifies the requirements for preimplantation genetic testing (PGT) aneuploidy analysis software and describes the corresponding experimental methods. This document applies to preimplantation low-depth high-throughput gene sequencing data generated from compatible testing kits and gene sequencers. The analysis aims to determine whether the embryo has chromosomal aneuploidy, large segment deletions, duplications, or other abnormalities.

2 Normative references

The contents of the following documents, through normative references within the text, constitute essential provisions of this document. Dated citations are not included. For references to documents, only the version corresponding to that date applies to this document; for undated references, the latest version (including all amendments) applies. This document.

YY/T 1657 Preimplantation Genetic Chromosomal Aneuploidy Detection Kit (Sequencing Method)

3 Terms and Definitions

The following terms and definitions apply to this document.

3.1 In the in vitro fertilization-embryo transfer process, preimplantation genetic diagnosis (PGD) software is used to analyze the preimplantation chromosome aneuploidy of matched embryos. Analysis of low-depth whole-genome sequencing data generated by chromosome aneuploidy detection reagents and high-throughput sequencers was performed to obtain

embryonic chromosomes. The analysis results indicate whether the data is aneuploid or contains large fragment deletions or duplication anomalies.

Note. In this document, large deletions and duplications refer to chromosomal copy number variations (CNVs) larger than 4Mb, while abnormal fragments smaller than 4Mb are considered normal. Chromosomal abnormalities of 4Mb or more should be reported as needed clinically.

3.2 Effective reads Data obtained from high-throughput sequencing, after quality control, alignment, and deduplication, is aligned to unique locations on the genome and can be used for analysis. The number of effective reads.

Note. The unit prefix for the effective data volume in this document is M, where 1M represents 1,000,000. [Source: YY/T 1657,3.2]

3.3 genome coverage The effective data obtained from high-throughput sequencing covers the proportion of the physical length of the genome to the total length of the reference genome. [Source: YY/T 1657,3.3]

3.4 Copy number variation Deletions or duplications occurring in the genome.

Note. The size of abnormal segments in this file is expressed in Mb, where 1Mb represents a segment of 1,000,000 bp in length. [Source: YY/T 1657,3.4]