



BSI Standards Publication

Genomics informatics — Description rules for genomic data for genetic detection products and services

National foreword

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Foreword

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Introduction

The decreasing cost of sequencing and the gradual in-depth study of genomics have led to the generation of more and more genomic data, but the data quality in genomics is not optimal. From the dimension of data level, there is a lack of data integrity, and medical information has been facing a problem of semantic disunity. These problems have caused great obstacles to downstream applications.

Standardization of data is a prerequisite for data asset management and data storage and applications, which can give better storage for genomic data and enlarge these genomic data used in precision medicine.

This document is based on the actual situation of industry data production, combined with the needs of upstream and downstream industry users. It also takes into account the use made by stakeholders and user friendliness for all common types of genomic data. Solving the problem of data scope and semantic unification can enhance the data association ability, ensure information exchange, improve data flow, improve the data quality from the aspects of data integrity and data validity, and lay a good foundation for subsequent data storage, data application and data sharing.

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