BSI Standards Publication

Genomics informatics — Data elements and their metadata for describing the microsatellite instability (MSI) information of clinical massive parallel DNA sequencing

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National foreword

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ISO copyright office
CP 401 • Ch. de Blandonnet 8
CH-1214 Vernier, Geneva
Phone: +41 22 749 01 11
Email: copyright@iso.org
Website: www.iso.org

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Foreword

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The procedures used to develop this document and those intended for its further maintenance are described in the ISO/IEC Directives, Part 1. In particular, the different approval criteria needed for the different types of ISO documents should be noted. This document was drafted in accordance with the editorial rules of the ISO/IEC Directives, Part 2 (see www.iso.org/directives).

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This document was prepared by Technical Committee ISO/TC 215, *Health informatics*, Subcommittee SC 1, *Genomics informatics*.

Any feedback or questions on this document should be directed to the user's national standards body. A complete listing of these bodies can be found at www.iso.org/members.html.

Introduction

Massively parallel sequencing is a high-throughput analytical approach to nucleic acid sequencing that allows whole genomes, transcriptomes, and specific nucleic acid targets. These advanced technologies have been used in the clinical field, and clinical sequencing has been applied to realize personalized medicine and precision medicine. ISO/TS 20428[1] has been developed for clinical usage.

In the field of cancer treatment, various treatment strategies were performed differently from traditional anti-cancer chemotherapies. One of those strategies is the control of human immune system that maintains the action to extract cancer cells. Recent outcomes of clinical trials show that this immune therapy is efficient for some patients who have a specific molecular character of their tumor mass, such as PD-L1 or CTLA4 surface protein expression[2]. As a result, these molecular characters are used as biomarkers for selecting patients. In colon cancer, according to several clinical trials, it is reported that the status of MSI (microsatellite instability) is regarded as a biomarker that drugs based on immuno-therapy are more efficient for the patient with MSI-H (high)[3].

The status of MSI can be calculated and reported by small nucleotide deletion on a specific region of human genome reference with NGS sequencing[4]. According to US FDA, four NGS sequencing products were approved for companion diagnostics. Among these products, three NGS sequencing provide MSI status and value on their NGS sequencing report. CLIA-certified labs or equivalent level agencies in countries also are servicing the MSI status from their methods[5]. It is forecasted that more clinical NGS sequencing will be approved to report MSI.

However, there is no standard for describing MSI status, value, and metadata. ISO/TS 20428 focuses on only DNA variations compared with the reference genome. According to some research results, MSI status and the way to describe it are different even if using the same sequencing data. This makes it difficult for clinicians and researchers not only to use MSI status results for clinical decisions but also for secondary analyzing purposes when receiving from more than one sequencing lab. Related metadata should be essential to expand the usage of MSI status results.

In this document, the data elements and their standardized metadata for MSI status in electronic health records will be described. The clinical report for MSI will provide helpful information on bioinformatics analysis to help clinical decisions.

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1 Scope

This document identifies data elements and metadata to represent the information about microsatellite instability (MSI) for reporting the value of the biomarker using clinical massive parallel DNA sequencing.

This document covers information about the MSI test result and related data, such as used resources, data generation condition, and data processing information which are helpful to clinical diagnosis and research.

This document is not intended

— for defining experimental protocols or methods for calculating the value of microsatellite instability (MSI),

— for the other biological species than human resource, or

— for the Sanger sequencing methods.

2 Normative references

The following documents are referred to in the text in such a way that some or all of their content constitutes requirements of this document. For dated references, only the edition cited applies. For undated references, the latest edition of the referenced document (including any amendments) applies.

ISO 8601 (all parts), *Date and time — Representations for information interchange*

ISO/TS 22220:2011, *Health informatics — Identification of subjects of health care*

ISO/TS 27527:2010, *Health informatics — Provider identification*

3 Terms and definitions

For the purposes of this document, the following terms and definitions apply.

ISO and IEC maintain terminology databases for use in standardization at the following addresses:

— ISO Online browsing platform: available at https://www.iso.org/obp

— IEC Electropedia: available at https://www.electropedia.org/

3.1
biological specimen
biospecimen
specimen
sample of tissue, body fluid, food, or other substance that is collected or acquired to support the assessment, diagnosis, treatment, mitigation or prevention of a disease, disorder or abnormal physical state, or its symptoms

[SOURCE: ISO/TS 20428:2017, 3.34]