ISO/DTS.2 23494-1:~~2022~~2023(E)

Date: 2023-01-05

ISO/TC 276/WG 5

~~Date: 2022-12-05~~

Secretariat: DIN

Biotechnology— Provenance information model for biological material and data — Part 1: Design concepts and general requirements

DTS.2 stage

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Published in Switzerland.

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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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For an explanation of the voluntary nature of standards, the meaning of ISO specific terms and expressions related to conformity assessment, as well as information about ISO's adherence to the World Trade Organization (WTO) principles in the Technical Barriers to Trade (TBT), see www.iso.org/iso/foreword.html.

This document was prepared by Technical Committee ISO/TC 276, *Biotechnology*.

A list of all parts in the ISO 23494 series can be found on the ISO website.

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

Research in life sciences has undergone significant changes during recent years, evolving away from individual projects confined to small research groups to transnational consortia covering a wide range of techniques and expertise. The exchange of research data and biological materials has become essential for the research in life sciences and biotechnology, and consequently interoperability and quality measures of data have become imperative.

At the same time several reports addressing the quality of research papers in life sciences uncovered an alarming number of ill-founded claims. The reasons for the deficiencies are diverse, with insufficient quality and documentation of the biological material used being the major issue.

Hence there is urgent need for standardized and comprehensive documentation of the whole workflow from the collection, generation, processing, and analysis of the biological material to data analysis and statistics. This provenance information serves as a quality indicator and provides information on the reliability thus enabling transparency and comparability of research results.

The purpose of these documents is the standardization of provenance information management for the biotechnology domain in a way that allows for meaningful data integration. To this end, provenance information needs to be prepared in a way that enables interoperability between prevailing tools for data generation, processing, and analysis. While in information technology well-established approaches to provenance information management in general are available (e.g. OPM[1] or W3C PROV[2]), the implementation for the biotechnology domain and related fields in particular is still a pending issue (as discussed in the results from the Electronic Health Records Systems for Clinical Research (EHR4CR) and TRANSFoRm projects in several papers [1], [2][3][4]).

Since data in biotechnology mostly originate from analysis of biological material, it is essential that the provenance information covers the entire process chain, from the source of biological material, throughout processing and analysis of the material, generation, and processing of the data to final analysis and interpretation. With the increasing adoption of data-intensive technologies, such as next-generation sequencing, (NGS), high-throughput mass spectrometry as used for proteomics or metabolomics, or high-throughput microscopy in digital pathology, and their impact on data collection strategies, consistent and comprehensive documentation of data provenance has become a necessity.

In fact, the experimental designs in life sciences have moved from individual experiments with a limited amount of data towards pipelines generating a vast volume of raw digital data using massively parallel acquisition systems demanding for complex data processing workflows to extract biologically relevant information. This trend is particularly evident in next-generation sequencing (NGS), where the actual data acquisition device at the wet lab-digital interface — (i.e. the sequencer—) is completely oblivious to the details of the experiment being performed, with all the specialization pushed to the protocols used by the sample preparation procedure and to the software pipelines processing the data. Software pipelines are continuously changing due to the evolution of analytical algorithms and reference datasets,data sets, which is having a significant effect on result concordance.

In addition, particular issues, relevant to scientific domains utilizing biological material and data obtained from humans, must be considered. This includesThese include aspects of data privacy, ethics, or management of identities. Notably, issues such as withdrawal of an informed consent or communication of incidental findings require the implementation of appropriate mechanisms.

The major objectives for collecting and storing provenance information are summarized belowas follows:

[1] http://openprovenance.org/

[2] http://www.w3.org/TR/prov-dm/

— ~~Retrospective~~retrospective evaluation of experimental results and data analysis with respect to the influence of ~~Standard Operating Procedures (~~standard operating procedures (SOPs) and workflow parameters~~.~~;

— ~~Quality~~quality monitoring of biological materials and data entered in a workflow or analysis pipeline (e.g~~,~~. against reference ranges and tolerances)~~.~~;

— ~~Automation~~automation of quality control procedures (e.g~~,~~. comparisons between different pipelines)~~.~~;

— ~~Profiling~~profiling of sample and data analysis to identify bottlenecks~~.~~;

— ~~Assessment~~assessment of fitness for purpose of biological materials and data for the intended use.

To achieve these objectives, a digitally processable description of provenance information is required.

This overarching ~~standard~~document will be complemented by appropriate vertical standards for specific fields (e.g~~,~~. collection of biological material, data generation, ~~and~~ processing of biological material and data). The basic requirements contained in this document do not impose any limitations to future, domain-specific standards based on this ~~standard~~document.

The standardization of provenance information requires the conceptualization and essential specifications for the generation, management, provisioning~~,~~ and maintenance as described in this document. Not covered ~~here~~in this document are additional fundamental components such as a generic model for provenance information and extensions common to all kinds of provenance information, ensuring security, privacy and ~~nonrepudiability.~~non-repudiability. For particular domains in biotechnology, detailed specifications building on a common provenance model are required, covering provenance information describing:

— the life cycle of biological materials, including acquisition, processing, transport~~,~~ and storage.

— the data generation by analytical methods.

— the data processing and analysis in computational workflows.

This document provides definitions for relevant terms used and specifies fundamental requirements for provenance information generation, management~~,~~ and provisioning.

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Biotechnology — Provenance information model for biological material and data — Part 1: Design concepts and general requirements

1 Scope

This document specifies a general concept for a provenance information model for biological material and data and requirements for provenance data interoperability and serialization.

The provenance information model covers any information relevant to the quality and fitness ~~-~~for ~~-~~ purpose of the biological material generated throughout the preanalytical phase of the materials ~~lifecycle~~life cycle from collection to analysis, data originating from analytical procedures applied to the biological material and results from further mathematical processing of the data.

This document is applicable to organizations, authorities~~,~~ and industries ~~(~~that are:

a) collecting, processing, or distributing biological material for research;

~~(~~b) generating, collecting, ~~analyzing,~~analysing ~~or~~ storing data on biological material.

This document does not apply to biological material and data used for other than research or in fields that are regulated by national, regional~~,~~ or international laws, such as medical diagnosis and therapy or food production.

~~This standard does not touch legal, governmental, or organizational regulations or ethical rules.~~

NOTE International, national, or regional regulations or requirements can also apply to specific topics covered in this document.

2 Normative references

~~There are no normative references in this document.~~

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.

United Nations Treaty Collection. Nagoya Protocol on Access to Genetic Resources and the Fair and Equitable Sharing of Benefits Arising from their Utilization to the Convention on Biological Diversity [online]. Available from: https://treaties.un.org/pages/ViewDetails.aspx?src=IND&mtdsg_no=XXVII-8-b&chapter=27&clang=_en

3 Terms and definitions

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

ISO and IEC maintain ~~terminological~~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
~~**automatic**~~
~~pertaining to a process or equipment that, under specified conditions, functions without human intervention~~

~~[SOURCE: ISO/IEC 2382:2015, 2121282]~~

common provenance model
~~**3.2**~~
CPM
~~**Common Provenance Model**~~
extension of *PROV-DM* (3.12) for generating, maintaining~~,~~ and provisioning *provenance information* (3.13) on biological material and data

~~**3.3**~~
3.2
described activity
activity~~,~~ performed on a *described object* (3.3)

Note 1 to entry: Examples for activities performed on physical objects can be biobanking activities as specified in ISO 20387:2018, 3.6~~,~~. Examples for activities performed on digital objects can be data analytics as specified in ISO/IEC 20546:2019, 3.1.6.

3.~~4~~ 3
described object
physical or digital object~~,~~ which is being documented by *provenance information* (3.~~5~~ 13)

3.4
finalization event
time instance, at which generated and assembled *provenance information* (3.13) is transformed into *finalized provenance information* (3.5)

3.~~6~~ 5
finalized provenance information
provenance information (3.13) transformed into a representation specified by the ~~Common Provenance Model (CPM)~~*common provenance model* (3.1), and which is prepared to be conserved or archived and which is considered as being immutable

~~NOTE~~ Note 1 to entry: Finalized provenance information is a subset of provenance information.

3.~~7~~ 6
interoperability
capability to communicate, execute programs, or transfer data among various functional units in a manner that requires the user to have little or no knowledge of the unique characteristics of those units

[SOURCE: ISO/IEC 20944-1:2013, 3.6.1.24]

3.~~8~~ 7

machine-readable
pertaining to data in a form that can be automatically input to a computer

[SOURCE: ISO/IEC/IEEE 24765:2017, 3.2300~~]~~, modified — Example deleted.]

3.~~9~~ 8
opaque provenance component
part of the *finalized provenance information* (3.5) that is findable but not directly accessible and is stored as an opaque object at the respective *responsible subject* (3.15)

3.~~10~~ 9
persistent identifier
unique identifier that ensures permanent access for a digital object by providing access to it independently of its physical location or current ownership

[SOURCE: ISO 24619:2011, 3.2.4~~]~~, modified — Abbreviated term and note to entry deleted.]

3.~~11~~ 10
preparation
activities, taking place in a laboratory after acquisition, to make biological material ready for further use in the life cycle, storage or distribution

Note 1 to entry: These activities can include, e.g.~~,~~, centrifuging, homogenizing, purifying, fixing, stabilizing, replicating, filtering, sorting, culturing, vacuum drying, freeze drying, freezing and thawing, tissue sectioning, fractionating, dispensing/aliquoting, cryopreserving ~~etc~~.

[SOURCE: ISO 20387:2018, 3.37]

3.12
PROV-DM
~~Conceptual~~conceptual data model forming a basis for the W3C provenance (PROV) family of specifications~~.~~

[SOURCE: W3C PROV-DM:2013~~][3]~~[5]]

3.13
provenance information
information that documents the history of a *described object* (3.3) and related *described activities* ~~containing~~(3.2), and that contains information about the origin or source of the described object, any changes that ~~may~~can have taken place since it was originated, and who has had custody of it since it was originated

3.14
provenance provider
institution or its organizational unit that is responsible for the transformation of *provenance information* (3.13) into *finalized provenance information*~~,~~ (3.5) and its storage, and which can be either a *responsible subject* (3.15) or ~~which is~~ acting on behalf of a responsible subject

3.15
responsible subject
institution, or the organizational unit it is part of, which is responsible for providing and assembling *provenance information* (3.13) documenting a *described activity* (3.2) on a *described object* (3.3) it is involved in

3.16

serialization
process of translating data structures or object states into a format that can be stored or transmitted and reconstructed later

4 Requirements and recommendations on provenance information management

4.1 Provenance information generation

4.1.1 Output of machine-readable finalized provenance information shall occur only upon completion of the respective activity or process.

NOTE 1 Creating finalized provenance information only upon completion of a process is reasonable to avoid inconsistencies resulting from constant changes during recording of the data. Otherwise, requirements in terms of non-repudiation, trust and verifiability can be affected. In general, finalized provenance information is not meant to record the process conditions and parameters continuously, thus substituting for logging functionality, nor to detail any minor variation in the process conditions (e.g~~,~~. temperature fluctuations during storage).

Finalized provenance information should represent an aggregate of the data available, indicating adherence or deviation from predefined boundary conditions as necessary to assess fitness for purpose (e.g~~,~~. exceeding the temperature range for safe storage for a certain period, frequency~~,~~ and duration of deviations). If, however, detailed information is regarded vital or necessary to comply with particular regulations, embedding of comprehensive data is also possible.

If required, additional, specific finalized provenance information can be generated on request. Any additional finalized provenance information shall only be provided by the responsible subject.

NOTE 2 Such a request can be addressed by providing more detailed provenance information from recorded information. If only a subset of the finalized provenance information was made generally available (e.g~~,~~. for privacy reasons or because of confidentiality), additional finalized provenance information can be generated for a particular user.

To ascertain consistency and validity any additional finalized provenance information should be compared to the original provenance information for plausibility.

4.1.2 Machine-readable provenance information generated by the devices involved in processing, handling, transporting~~,~~ or storing biological material, or involved in data generation, shall be used, if available.

The compilation and processing of information relevant for the generation of finalized provenance information shall be documented and should be automated as much as possible, to minimize errors and optimize the amount and quality of information. This includes also any transformation applied to the information gathered. For any algorithmic transformation, the underlying algorithms should be either archived or referenced, so that any modification and processing of the information used to generate finalized provenance information is reproducible and documented.

Devices qualified for providing finalized provenance information as specified in this document shall be set to provide the provenance information accordingly and the information shall be used unmodified.

If a device is able to provide provenance information in a documented, machine-readable format which does not ~~comply with~~conform to this document, the device shall be set to produce machine-readable provenance information. The provenance information obtained shall be transformed subsequently to ~~comply with~~conform to the standard format. The implementation of the algorithm used for transformation shall be documented and referenced in the finalized provenance information.