



**International  
Standard**

**ISO/IEC 23092-3**

**Information technology — Genomic  
information representation —**

**Part 3:  
Metadata and application  
programming interfaces (APIs)**

*Technologie de l'information — Représentation des informations  
génomiques —*

*Partie 3: Métadonnées et interfaces de programmation  
d'application (API)*

ISO/IEC 23092-3:2025

<https://standards.iteh.ai/catalog/standards/iso/c0471a33-f16a-4342-8899-c82386dbf996/iso-iec-23092-3-2025>

**Third edition  
2025-05**

iTeh Standards  
(<https://standards.iteh.ai>)  
Document Preview

ISO/IEC 23092-3:2025

<https://standards.iteh.ai/catalog/standards/iso/c0471a33-f16a-4342-8899-c82386dbf996/iso-iec-23092-3-2025>



**COPYRIGHT PROTECTED DOCUMENT**

© ISO/IEC 2025

All rights reserved. Unless otherwise specified, or required in the context of its implementation, no part of this publication may be reproduced or utilized otherwise in any form or by any means, electronic or mechanical, including photocopying, or posting on the internet or an intranet, without prior written permission. Permission can be requested from either ISO at the address below or ISO's member body in the country of the requester.

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

Published in Switzerland

# Contents

Page

<b>Foreword</b>	<b>v</b>
<b>Introduction</b>	<b>vi</b>
<b>1 Scope</b>	<b>1</b>
<b>2 Normative references</b>	<b>1</b>
<b>3 Terms and definitions</b>	<b>2</b>
<b>4 Abbreviated terms</b>	<b>2</b>
<b>5 Conventions</b>	<b>2</b>
5.1 Character encoding	2
5.2 Bit Ordering	2
5.3 Syntax functions and data types	3
5.4 Graphic notations	3
<b>6 Information metadata</b>	<b>4</b>
6.1 General	4
6.2 Dataset group metadata	4
6.3 Reference metadata	5
6.4 Dataset metadata	5
6.5 Annotation table metadata	7
6.5.1 General	7
6.5.2 Annotation table general metadata	7
6.5.3 Annotation table analytics metadata	10
6.5.4 Annotation table linkages metadata	13
6.5.5 Annotation table history metadata	14
6.6 Metadata protection	15
6.7 Mechanism for extensions of the metadata set	16
6.7.1 General	16
6.7.2 Example for dataset metadata extensions	16
6.7.3 Example for obfuscating labels	16
6.7.4 Example for obfuscating sequences	17
6.8 Metadata profiles	17
6.8.1 General	17
6.8.2 Example of metadata profile — Run	17
6.8.3 Example of metadata profile — Genomic data commons	18
<b>7 Metrics metadata</b>	<b>18</b>
7.1 General	18
7.2 Syntax	18
7.3 Semantics	19
<b>8 Clinical data linkage metadata</b>	<b>20</b>
8.1 General	20
8.2 CDL Metadata protection	22
<b>9 Protection metadata</b>	<b>22</b>
9.1 General	22
9.2 Encryption of <b>gen_info</b> elements and blocks	23
9.2.1 General	23
9.2.2 EncryptionParameters carried in dataset group protection	23
9.2.3 EncryptionParameters carried in dataset protection	24
9.2.4 EncryptionParameters carried in annotation table protection	28
9.2.5 Key retrieval	33
9.2.6 Decryption	34
9.3 Privacy rules for the use of the genomic information	36
9.3.1 General	36
9.3.2 Example of use of privacy rules	37

9.4	Digital signature of <b>gen_info</b> elements and blocks .....	38
9.4.1	General .....	38
9.4.2	Signatures carried in dataset group protection .....	38
9.4.3	Signatures carried in dataset protection .....	38
9.4.4	Signatures carried in annotation table protection .....	40
9.4.5	Signatures carried in descriptor stream protection .....	42
<b>10</b>	<b>Access unit information .....</b>	<b>42</b>
10.1	General .....	42
10.2	genAuxRecord .....	43
10.3	genAux .....	44
10.4	genTag .....	44
<b>11</b>	<b>Decoding process for metadata .....</b>	<b>45</b>
11.1	General .....	45
11.2	Initialization of parameters .....	47
11.2.1	General .....	47
11.2.2	Properties .....	47
11.2.3	Parameters .....	48
11.2.4	Constants .....	48
11.2.5	Process .....	49
11.3	Macros .....	51
11.4	Decoding process .....	53
<b>12</b>	<b>Application programming interfaces (APIs) .....</b>	<b>60</b>
12.1	General .....	60
12.2	Structure of the API .....	60
12.3	Detailed specification of the API .....	61
12.3.1	Data types .....	61
12.3.2	Return codes .....	61
12.3.3	Metadata fields .....	62
12.3.4	Output structures .....	62
12.3.5	Filters .....	71
12.3.6	Genomic information .....	79
12.3.7	Metadata .....	84
12.3.8	Protection .....	87
12.3.9	Reference .....	91
12.3.10	Statistics .....	91
<b>Annex A</b> (normative)	<b>XML schemas corresponding to metadata information and protection elements .....</b>	<b>96</b>
<b>Annex B</b> (informative)	<b>Example use cases of annotation table linkages metadata .....</b>	<b>98</b>
<b>Annex C</b> (informative)	<b>XML schemas and XML-based data .....</b>	<b>100</b>
<b>Annex D</b> (informative)	<b>Example of key transport .....</b>	<b>109</b>
<b>Annex E</b> (informative)	<b>SAM interoperability .....</b>	<b>113</b>
<b>Bibliography</b> .....	<b>120</b>	

## Foreword

ISO (the International Organization for Standardization) and IEC (the International Electrotechnical Commission) form the specialized system for worldwide standardization. National bodies that are members of ISO or IEC participate in the development of International Standards through technical committees established by the respective organization to deal with particular fields of technical activity. ISO and IEC technical committees collaborate in fields of mutual interest. Other international organizations, governmental and non-governmental, in liaison with ISO and IEC, also take part in the work.

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 document 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](http://www.iso.org/directives) or [www.iec.ch/members\\_experts/refdocs](http://www.iec.ch/members_experts/refdocs)).

ISO and IEC draw attention to the possibility that the implementation of this document may involve the use of (a) patent(s). ISO and IEC take no position concerning the evidence, validity or applicability of any claimed patent rights in respect thereof. As of the date of publication of this document, ISO and IEC had received notice of (a) patent(s) which may be required to implement this document. However, implementers are cautioned that this may not represent the latest information, which may be obtained from the patent database available at [www.iso.org/patents](http://www.iso.org/patents) and <https://patents.iec.ch>. ISO and IEC shall not be held responsible for identifying any or all such patent rights.

Any trade name used in this document is information given for the convenience of users and does not constitute an endorsement.

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](http://www.iso.org/iso/foreword.html). In the IEC, see [www.iec.ch/understanding-standards](http://www.iec.ch/understanding-standards).

This document was prepared by Joint Technical Committee ISO/IEC JTC 1, *Information technology*, Subcommittee SC 29, *Coding of audio, picture, multimedia and hypermedia information*.

This third edition cancels and replaces the second edition (ISO/IEC 23092-3:2022), which has been technically revised.

The main changes are as follows:

- addition of annotation table metadata ([subclause 6.5](#)) that contains general, analytics, linkages and access history information associated with an annotation table;
- addition of metrics metadata ([Clause 7](#)) that contains pre-computed sequencing data metrics associated with a dataset or an access unit;
- addition of clinical data linkage metadata ([Clause 8](#)) that contains linkage information for enabling clinical data interchange (CDI) with external data sources;
- addition of annotation table protection metadata, including encryption parameters ([subclause 9.2.4](#)) and digital signatures ([subclause 9.4.4](#)), and updates to the decryption process ([subclause 9.2.6](#)) and privacy rules ([subclause 9.3](#)) for enabling the selective protection of annotation data;
- extension of the APIs ([Clause 12](#)) for supporting the random access and query of annotation data, the retrieval of pre-computed sequencing data statistics, and the return of only the number of matching records without the actual data.

A list of all parts in the ISO/IEC 23092 series can be found on the ISO and IEC websites.

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](http://www.iso.org/members.html) and [www.iec.ch/national-committees](http://www.iec.ch/national-committees).

## Introduction

The advent of high-throughput sequencing (HTS) technologies has the potential to boost the adoption of genomic information in everyday practice, ranging from biological research to personalized genomic medicine in the clinic. As a consequence, the volume of generated data has increased dramatically during the last few years, and an even more pronounced growth is expected in the near future.

At the moment, genomic information is mostly exchanged through a variety of data formats, such as FASTA/FASTQ for unaligned sequencing reads and SAM/BAM/CRAM for aligned reads. With respect to such formats, the ISO/IEC 23092 series provides a new solution for the representation and compression of genome sequencing information by:

- specifying an abstract representation of the sequencing data rather than a specific format with its direct implementation;
- being designed at a time point when technologies and use cases are more mature. This permits the addressing of one limitation of the textual SAM format, for which incremental ad-hoc addition of features followed along the years, resulting in an overall redundant and suboptimal format which at the same time results not general and unnecessarily complicated;
- normatively separating free-field user-defined information with no clear semantics from the normative genomic data representation. This allows a fully interoperable and automatic exchange of information between different data producers;
- allowing multiplexing of relevant metadata information with the data since data and metadata are partitioned at different conceptual levels;
- following a strict and supervised development process which has proven successful in the last 30 years in the domain of digital media for the transport format, the file format, the compressed representation and the application program interfaces.

This document provides the enabling technology that will allow the community to create an ecosystem of novel, interoperable solutions in the field of genomic information processing. In particular, it offers:

- consistent, general and properly designed format definitions and data structures to store sequencing and alignment information. A robust framework which can be used as a foundation to implement different compression algorithms;
- speed and flexibility in the selective access to coded data, by means of newly designed data clustering and optimized storage methodologies;
- low latency in data transmission and consequent fast availability at remote locations, based on transmission protocols inspired by real-time application domains;
- built-in privacy and protection of sensitive information, thanks to a flexible framework which allows customizable secured access at all layers of the data hierarchy;
- reliability of the technology and interoperability among tools and systems, owing to the provision of a normative procedure to assess conformance to the standard on an exhaustive dataset;
- support to the implementation of a complete ecosystem of compliant devices and applications, through the availability of a normative reference implementation covering the totality of the specification.

The fundamental structure of the ISO/IEC 23092 series data representation is the genomic record. The genomic record is a data structure consisting of either a single sequence read, or a paired sequence read, and its associated sequencing and alignment information; it may contain detailed mapping and alignment data, a single or paired read identifier (read name) and quality values.

Without breaking traditional approaches, the genomic record introduced in the ISO/IEC 23092 series provides a more compact, simpler and manageable data structure grouping all the information related to a single DNA template, from simple sequencing data to sophisticated alignment information.

The genomic record, although it is an appropriate logic data structure for interaction and manipulation of coded information, is not a suitable atomic data structure for compression. To achieve high compression ratios, it is necessary to group genomic records into clusters and to transform the information of the same type into sets of descriptors structured into homogeneous blocks. Furthermore, when dealing with selective data access, the genomic record is a too small unit to allow effective and fast information retrieval.

For these reasons, this document introduces the concept of access unit, which is the fundamental structure for coding and access to information in the compressed domain.

The access unit is the smallest data structure that can be decoded by a decoder conforming to ISO/IEC 23092-2. An access unit is composed of one block for each descriptor used to represent the information of its genomic records; therefore, a block payload is the coded representation of all the data of the same type (i.e. a descriptor) in a cluster.

In addition to clusters of genomic records compressed into access units, reads are further classified in six data classes: five classes are defined according to the result of their alignment against one or more reference sequences; the sixth class contains either reads that could not be mapped or raw sequencing data. The classification of sequence reads into classes enables to develop powerful selective data access. In fact, access units inherit a specific data characterization (e.g. perfect matches in Class P, substitutions in Class M, indels in Class I, half-mapped reads in Class HM) from the genomic records composing them, and thus constitute a data structure capable of providing powerful filtering capability for the efficient support of many different use cases.

Access units are the fundamental, finest grain data structure in terms of content protection and in terms of metadata association. In other words, each access unit can be protected individually and independently. [Figure 1](#) shows how access units, blocks and genomic records relate to each other in the ISO/IEC 23092 series data structure.

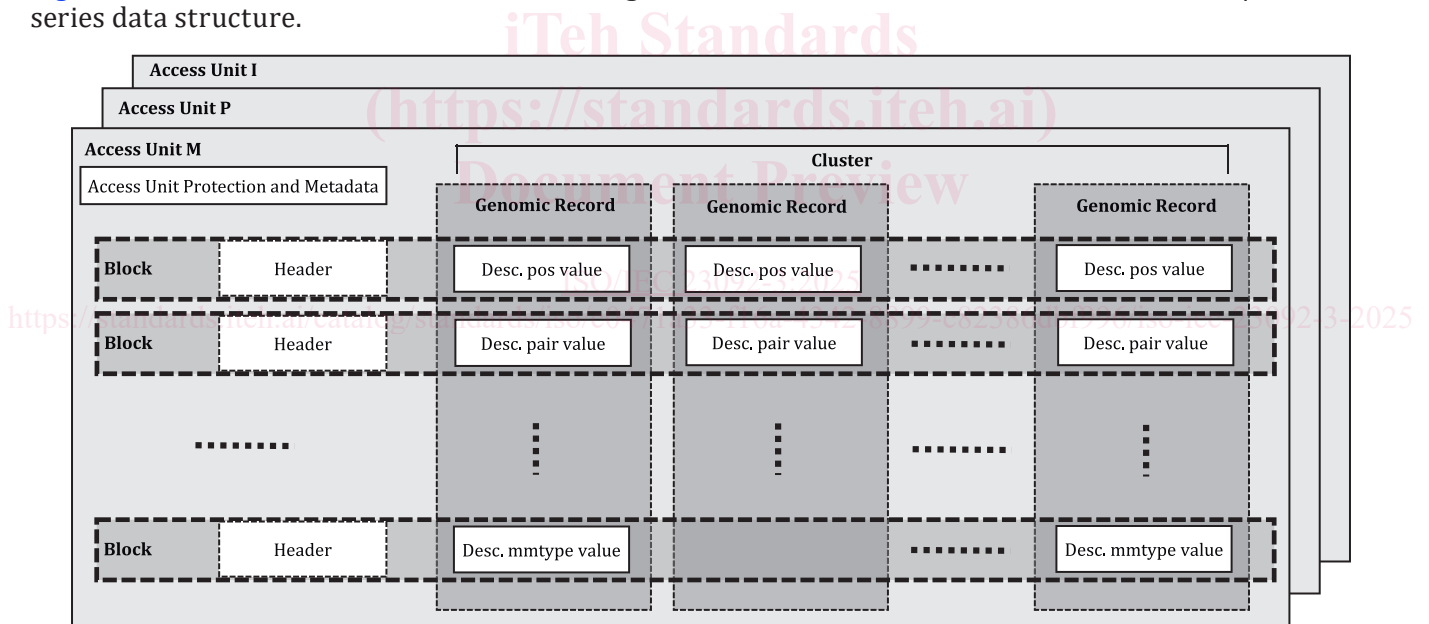


Figure 1 — Access units, blocks and genomic records