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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)

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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 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~~www.iso.org/directives or www.iec.ch/members_experts/refdocs).

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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:

- ~~The~~ addition of annotation table metadata ~~(subclause 6.5)~~(subclause 6.5) that contains general, analytics, linkages and access history information associated with an annotation table;
- ~~The~~ addition of metrics metadata ~~(Clause 7)~~(Clause 7) that contains pre-computed sequencing data metrics associated with a dataset or an access unit;
- ~~The~~ addition of clinical data linkage metadata ~~(Clause 8)~~(Clause 8) that contains linkage information for enabling clinical data interchange (CDI) with external data sources;
- ~~The~~ addition of annotation table protection metadata, including encryption parameters ~~(subclause 9.2.4)~~(subclause 9.2.4) and digital signatures ~~(subclause 9.4.4)~~,(subclause 9.4.4), and updates to the decryption process ~~(subclause 9.2.6)~~(subclause 9.2.6) and privacy rules ~~(subclause 9.3)~~(subclause 9.3) for enabling the selective protection of annotation data;

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— The extension of the APIs (Clause 12)(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.htmlwww.iso.org/members.html and www.iec.ch/national-committees.

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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

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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 ~~compliant with~~confirming 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~~Figure 1 shows how access units, blocks and genomic records relate to each other in the ISO/IEC 23092 series data structure.

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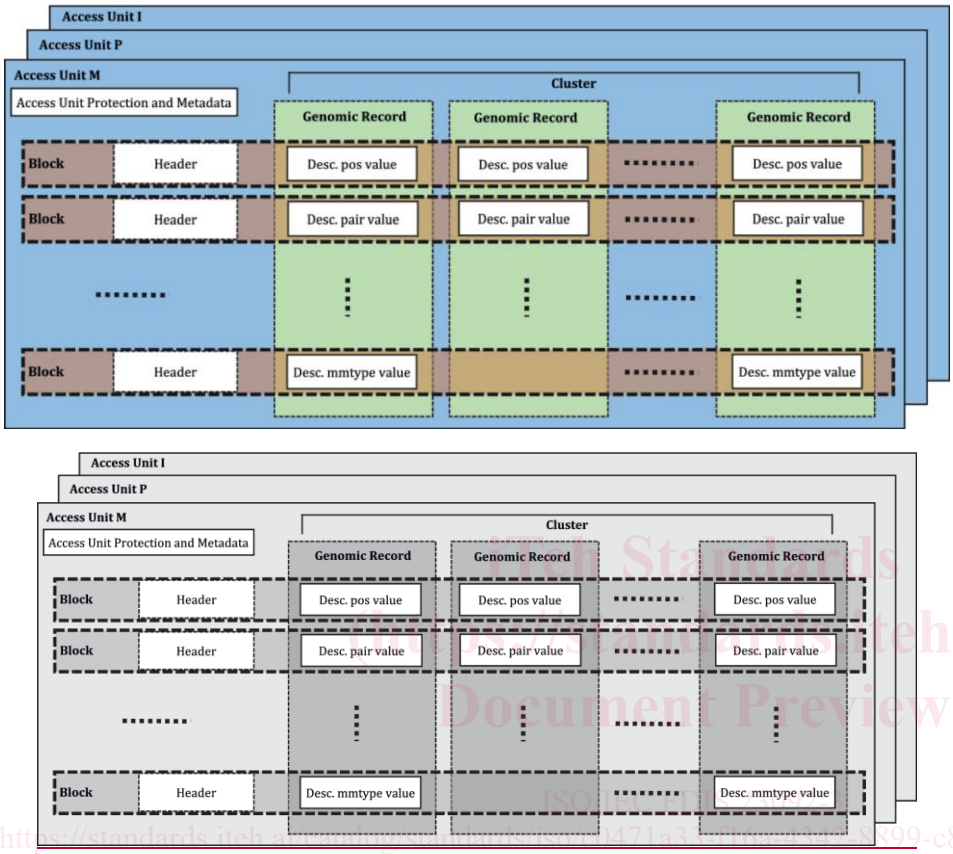

Figure 1 — Access units, blocks and genomic records

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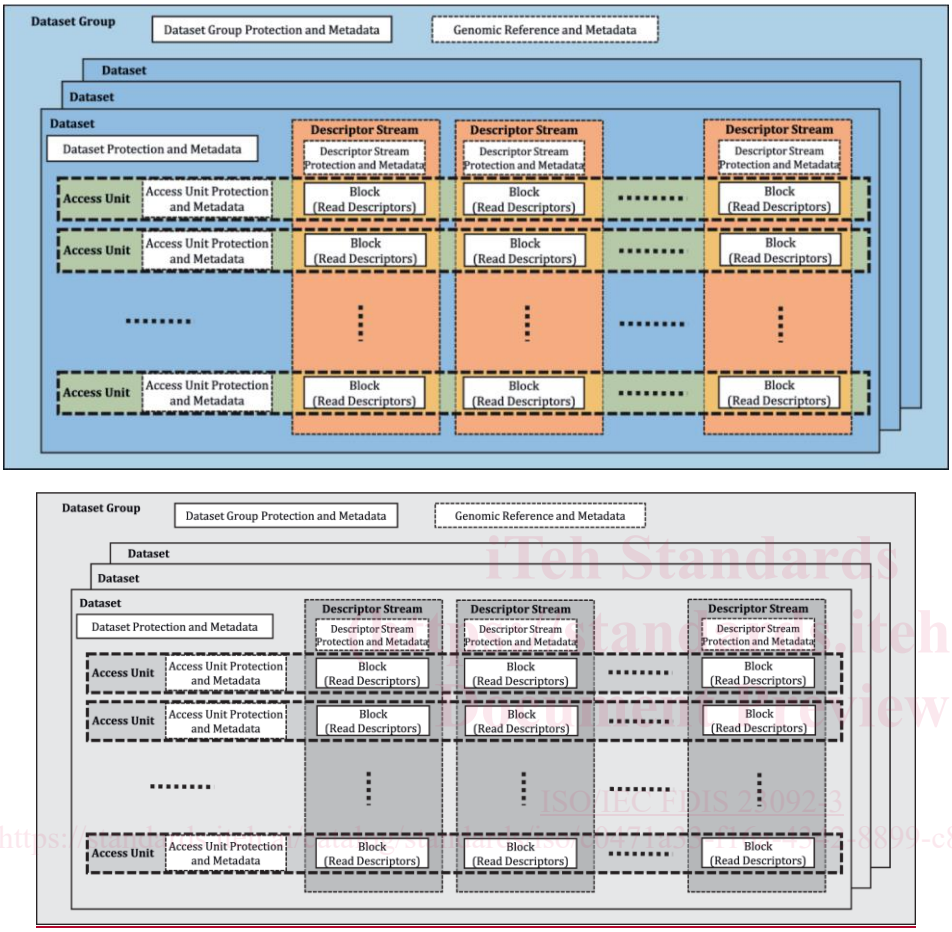

Figure 2. — High-level data structure: datasets and dataset group

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A dataset is a coded data structure containing headers and one or more access units. Typical datasets ~~could~~can, for example, contain the complete sequencing of an individual, or a portion of it. Other datasets ~~could~~can contain, for example, a reference genome or a subset of its chromosomes. Datasets are grouped in dataset groups, as shown in ~~Figure 2.~~Figure 2.

A simplified diagram of the dataset decoding process is shown in ~~Figure 3.~~Figure 3.

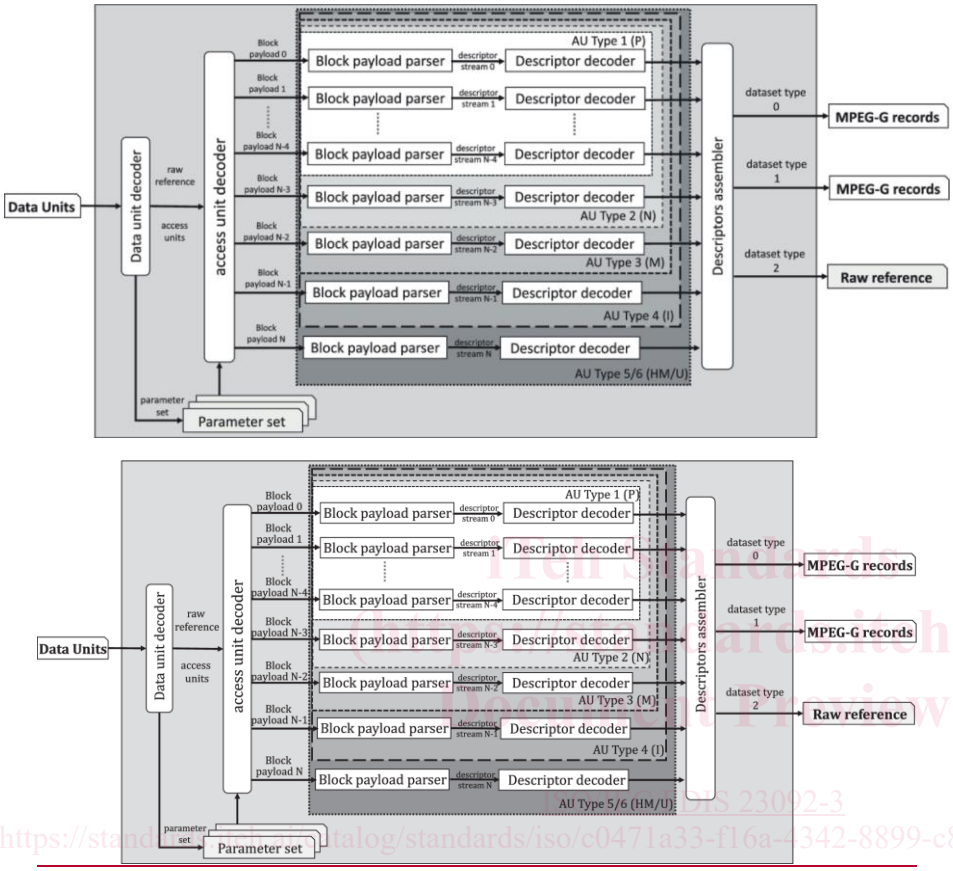

Figure 3 — Decoding process

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