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Information technology — Genomic information representation —

Part 6: Coding of genomic annotations

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Foreword

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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*.

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

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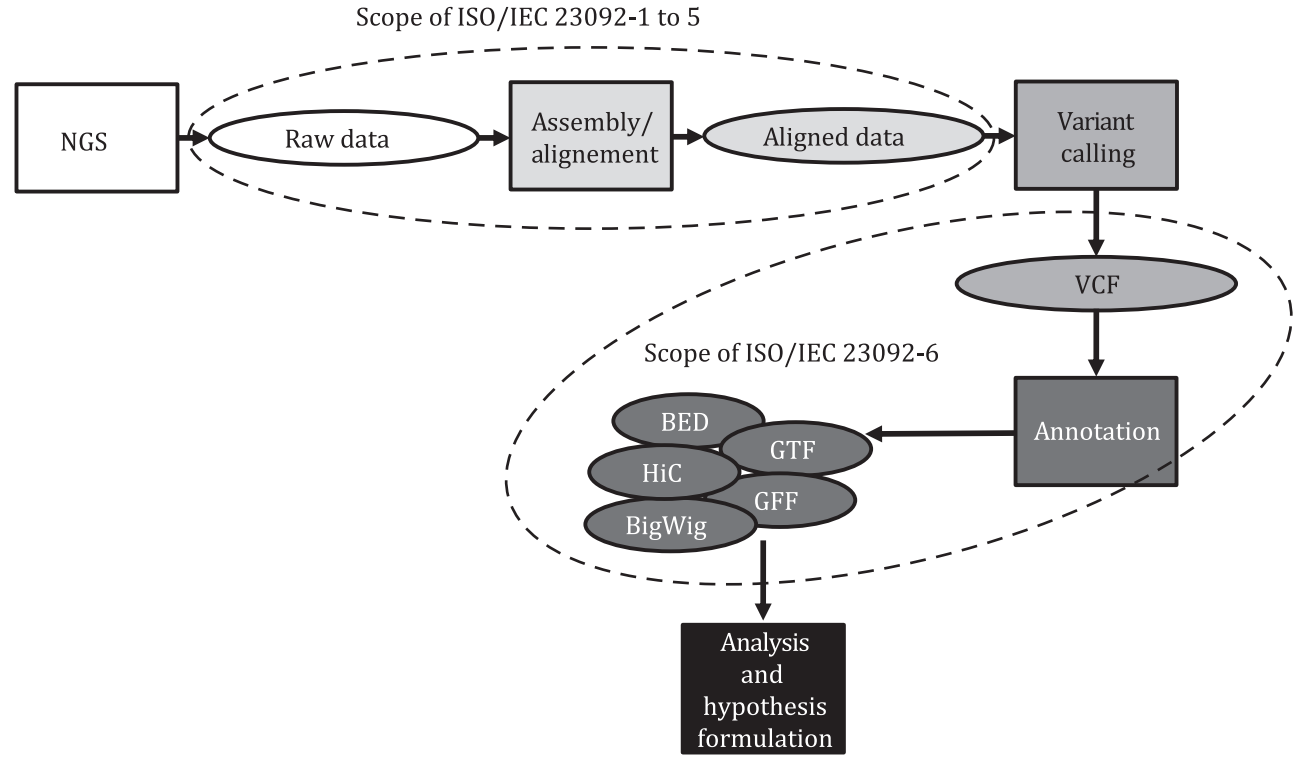
Introduction

While ISO/IEC 23092-1 to ISO/IEC 23092-5 (MPEG-G) deal with the representation of genomic information derived from the primary analysis of high-throughput sequencing (HTS) data – sequencing reads and qualities, and their alignment to a reference genome – which is only the first step in a long series. In particular, the results of primary analysis are usually processed further in order to obtain higher-level information. Such a process of aggregating information deduced from single reads and their alignments to the genome into more complex results is generally known as secondary analysis. In most HTS-based biological studies, the output of secondary analysis is usually represented as different types of annotations associated to one or more genomic intervals on the reference sequences.

Biological studies typically produce genomic annotation data such as mapping statistics, quantitative browser tracks, variants, genome functional annotations, gene expression data and Hi-C contact matrices. These diverse types of downstream genomic data are currently represented in different formats such as VCF, BED, WIG, etc., with loosely defined semantics, leading to issues with interoperability, the need for frequent conversions between formats, difficulty in the visualization of multi-modal data and complicated information exchange. Figure 1 depicts a typical pipeline for the primary and secondary analyses of HTS data, the file formats involved and the scopes of different parts of the ISO/IEC 23092 series.

Furthermore, the lack of a single format has stifled the work on compression algorithms and has led to the widespread use of general compression algorithms with suboptimum performance. These algorithms do not exploit the fact the annotation data typically comprises of multiple fields (attributes) with different statistical characteristics and instead compress them together. Therefore, while these algorithms support efficient random access with respect to genomic position, they do not allow extraction of specific fields without decompressing all the whole file.

In response to the aforementioned challenges, this document details a unified data format for the efficient representation and compression of diverse genomic annotation data for file storage or data transport. The benefits are manifold: reducing the cost of data storage, improving the speed of random data access and processing, providing support for data security and privacy in selective genomic regions, and creating linkages across different types of genomic annotation and sequencing data. The ultimate goal is to enable the secured and seamless sharing, processing and analysis of multi-modal genomic data in order to reduce the burden of data manipulation and management, so scientists can focus on biological interpretation and discovery.



Key

- sequencing generates raw reads
- read alignment
- variant calling
- variants annotations
- analysis

Figure 1 — Typical pipeline for the primary and secondary analyses of HTS data

Information technology — Genomic information representation —

Part 6: Coding of genomic annotations

1 Scope

This document provides specifications for the normative representation of the following types of genomic information:

— variants with genotyping information

— functional annotations

— tracks

— expression matrices

— contact matrices (from Hi-C experiments or similar).

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 specification. For dated references, only the edition cited applies. For undated references, the latest edition of the referenced document (including any amendments) applies.

ISO/IEC 10646, *Information technology — Universal coded character set (UCS)*

ISO/IEC 11544, *Information technology — Coded representation of picture and audio information — Progressive bi-level image compression*

ISO/IEC 23092-1, *Information technology — Genomic information representation — Part 1: Transport and storage of genomic information*

ISO/IEC 23092-2, *Information technology — Genomic Information Representation — Part 2: Coding of Genomic Information*