
**Biotechnology — Massively parallel
sequencing —**

**Part 2:
Quality evaluation of sequencing data**

Biotechnologie — Séquençage massivement parallèle —

Partie 2: Évaluation de la qualité des données de séquençage



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Contents

Page

Foreword	iv
Introduction	v
1 Scope	1
2 Normative references	1
3 Terms and definitions	1
4 Raw data	6
4.1 General	6
4.2 Raw data file	6
4.3 Quality assessment of raw data	6
4.3.1 General	6
4.3.2 Basic statistics	7
4.3.3 Quality metrics	7
4.4 Raw data pre-processing	8
5 Sequence alignment and mapping	8
5.1 General	8
5.2 Alignment and mapping file format	9
5.3 Quality control of sequencing alignment and mapping	9
5.3.1 Basic alignment statistics	9
5.3.2 Quality indicators	10
5.3.3 Methods for alignment and mapping quality assessment	11
5.4 Alignment post-processing	11
6 Variant calling	11
6.1 General	11
6.2 Data file for variant calling	11
6.3 Quality metrics in the variant calling	12
6.4 Processing of false positive variants	12
6.5 Sequence annotation	12
7 Validation	12
7.1 General	12
7.2 Validation of quality metrics	13
8 Documentation	14
Annex A (informative) Quality metrics for specific example MPS platforms	15
Annex B (informative) Coverage and read recommendations by applications	16
Annex C (informative) Software for sequence alignment and mapping	18
Bibliography	19

Foreword

ISO (the International Organization for Standardization) is a worldwide federation of national standards bodies (ISO member bodies). The work of preparing International Standards is normally carried out through ISO technical committees. Each member body interested in a subject for which a technical committee has been established has the right to be represented on that committee. International organizations, governmental and non-governmental, in liaison with ISO, also take part in the work. ISO collaborates closely with the International Electrotechnical Commission (IEC) on all matters of electrotechnical standardization.

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Introduction

Massively parallel sequencing (MPS) is a high-throughput analytical approach to nucleic acid sequencing utilizing massively parallel processing, that allows whole genomes, transcriptomes and specific nucleic acid targets from different organisms to be investigated in a relatively short time.

MPS is used in many life science disciplines permitting determination and high throughput analysis of millions and thousands of millions of nucleotide bases. The biological variability of deoxyribonucleic and ribonucleic acid polymers from living organisms results in challenges in accurately determining their sequences. The quality of sequence determination by MPS depends on many factors including but not limited to sample quality, library preparation, platform selection, and sequencing data quality.

The analysis of sequencing data poses significant bioinformatics challenges in various areas such as data storage, computation time and variant detection accuracy. One of the major challenges associated with sequencing data that is sometimes easily overlooked is monitoring quality control metrics over all stages of the data processing pipeline. Knowledge of data quality is essential for downstream analysis of sequences. Quality control for nucleic acid sequencing data handling and analysis can be separated into three stages: raw data, alignment and variant calling. This document provides a list of considerations for quality evaluation of MPS sequencing data, and the specific recommendations for different MPS platforms.

Biotechnology — Massively parallel sequencing —

Part 2:

Quality evaluation of sequencing data

1 Scope

This document specifies general requirements and recommendations for quality assessments and control of massively parallel sequencing (MPS) data. It covers post raw data generation procedures, sequencing alignments, and variant calling.

This document also gives general guidelines for validation and documentation of MPS data.

This document does not apply to any processes related to de novo assembly.

2 Normative references

There are no normative references in this document.

3 Terms and definitions

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

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

- ISO Online browsing platform: available at <https://www.iso.org/obp>
- IEC Electropedia: available at <http://www.electropedia.org/>

3.1

adapter sequence

adapter

artificial oligonucleotide of a known sequence that can be added to the 3' or 5' ends of a nucleic acid fragment

Note 1 to entry: It provides the primer site as well as other necessary sequences for sequencing the insert.

3.2

algorithm

completely determined finite sequence of instructions by which the values of the output variables may be calculated from the values of the input variables

[SOURCE: IEC 60050-351:2013, 351-42-27, modified — The notes were deleted.]

3.3

base calling

computational process in massively parallel sequencing of translating raw electrical signals to nucleotide sequence

Note 1 to entry: Base calling application and algorithm performance is characteristically defined by read and consensus accuracy.