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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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Foreword

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

A list of all parts in the ISO 20397 series can be found on the ISO website.

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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.