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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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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.