ISO/TS 22692:2020 (E)

Genomics informatics— Quality control metrics for DNA sequencing

Contents

Foreword

Introduction

1 Scope

2 Normative references

3 Terms and definitions

4 Abbreviated terms

5 Quality control metrics for sample preparation

- 5.1 General
- 5.2 Sample sequencing type
- 5.2.1 Sequencing type
- 5.2.2 Target gene
- 5.3 Sample information
- 5.3.1 Specimen type
- 5.3.2 Sampling date
- 5.3.3 Specimen origin
- 5.4 Summary of sample preparation related metrics

6 Quality control metrics for library preparation

- 6.1 General
- 6.2 DNA extraction method
- 6.2.1 DNA extraction kit
- 6.3 DNA quality
- 6.3.1 General
- 6.3.2 DNA purity
- 6.3.3 DNA integrity
- 6.4 Library construction
- 6.4.1 Library input amount
- 6.4.2 Library insert size
- 6.4.3 Library construction kit
- 6.5 Summary of library preparation related metrics

7 Quality control metrics for sequencing

- 7.1 General
- 7.2 Sequencing information
- 7.2.1 Sequencing instrument
- 7.2.2 Read length
- 7.2.3 Sequencing direction
- 7.2.4 Running mode
- 7.3 Running quality information
- 7.3.1 Error rate
- 7.3.2 Percent data quality >Q30
- 7.4 Summary of sequencing related metrics

8 Quality control metrics for data processing

- 8.1 General
- 8.2 Data quality measurement
- 8.2.1 Total reads
- 8.2.2 Mean coverage

8.2.3 Uniformity
8.2.4 Duplication rate
8.2.5 On-target rate
8.2.6 Q30 rate
8.3 Sequencing alignment
8.3.1 Mapping algorithm
8.3.2 Local realignment software and version
8.4 Variant calling
8.4.1 Variant calling software and version
8.4.2 Variant call quality score
8.4.3 Allelic read percentage & ratio
8.5 Variant filtering and annotation
8.5.1 General
8.5.2 Germline filter criteria
8.5.3 Mutation and annotation database
8.6 Summary of data processing related metrics

Annex A (informative) Example layout of quality control metrics

Page count: 15