ISO/TS 4424:2023-12 (E)

Genomics informatics - Data elements and their metadata for describing the tumor mutation burden (TMB) information of clinical massive parallel DNA sequencing

Contents

Page

Foreword ..... **v**

Introduction ..... **vi**

1 Scope **1**

2 Normative references **1**

3 Terms and definitions **1**

4 Abbreviated terms **4**

5 Tumor mutation burden (TMB) **5**

6 Composition of elements for describing TMB on the clinical DNA NGS report **6**
6.1 General 6
6.2 Summary part 6
6.3 Detail part 6

7 Fields and their nomenclature of required data **6**
7.1 General 6
7.2 Clinical sequencing order 7
7.2.1 General 7
7.2.2 Clinical sequencing order code 7
7.2.3 Date and time 7
7.3 Information on subject of care 8
7.3.1 General 8
7.3.2 Subject of care identifier 8
7.3.3 Subject of care name 8
7.3.4 Subject of care birth date 8
7.3.5 Subject of care sex 8
7.3.6 Referring diagnosis 9
7.4 Information on legally authorized person ordering clinical sequencing 9
7.4.1 General 9
7.5 Performing laboratory 9
7.5.1 General 9
7.5.2 Basic information on performing laboratory 9
7.5.3 Information on report generator 9
7.5.4 Information of legally confirmed person on sequencing report 9
7.6 Biospecimen information 9
7.6.1 General 9
7.6.2 Type of specimen 9
7.7 TMB status result information 10
7.7.1 General 10
7.7.2 TMB value 10
7.7.3 TMB status 10
7.8 Recommended treatment 10
7.8.1 General 10
7.8.2 Medication 10
7.8.3 Clinical trial information 10
7.8.4 Other recommendation 10
7.8.5 Supporting information 11

8 Fields and their nomenclature of optional data **11**
8.1 General 11

8.2 Reference genome version 12
8.3 TMB information 12
8.3.1 General 12
8.3.2 Criteria of TMB status 12
8.3.3 Approach of filtering germline variants 12
8.3.4 Variant types used for calculating TMB value 12
8.3.5 TMB region covered 12
8.3.6 Calibrated TMB value 12
8.4 Sequencing information 13
8.4.1 Clinical sequencing date 13
8.4.2 Sequencing type 13
8.4.3 Quality control metrics 13
8.4.4 Sequencing platform information 13
8.4.5 Analysis platform information 14
Annex A (informative) **Example structure of TMB report** **16**
Bibliography **20**