ISO/TS 4425:2023-04 (E)

Genomics informatics - Data elements and their metadata for describing the microsatellite instability (MSI) information of clinical massive parallel DNA sequencing

Contents

Page

Foreword v

Introduction vi

1 Scope 1

2 Normative references 1

3 Termsanddefinitions 1

4 Abbreviated terms 5

5 Microsatellite instability (MSI) 5

6 Composition of elements for describing MSI status on clinical DNA NGS report 6
6.1 General 6
6.2 Summary part 7
6.3 Detail part 7

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

7.8.3 Clinical trial information 11
7.8.4 Other recommendations 11
7.8.5 Supporting information 11

8 Fields and their nomenclature of optional data 12
8.1 General 12
8.2 Reference genome information 12
8.3 MSI information 13
8.3.1 General 13
8.3.2 Criteria of MSI status 13
8.3.3 Genomic position for determining MSI status 13

8.3.4 Genomic position against markers of alternative method 13
8.3.5 Clinical implication of MSI status 13
8.4 Sequencing information 13
8.4.1 Clinical sequencing date 13
8.4.2 Sequencing type 13
8.4.3 Quality control metrics 14
8.4.4 Sequencing platform information 14
8.4.5 Analysis platform information 14

Annex A (informative) Example structure of MSI status report 16

Bibliography 19