

ISO/TS 22693:2021 (E)

Genomics informatics — Structured clinical gene fusion report in electronic health records

Contents

	Foreword
	Introduction
1	Scope
2	Normative references
3	Terms and definitions
4	Abbreviated terms
5	Gene fusion
6	Composition of clinical gene fusion report
6.1	General
6.2	Summary section
6.3	Detailed section
7	Fields and their nomenclature of required data
7.1	General
7.2	Clinical sequencing order
7.2.1	General
7.2.2	Clinical sequencing order code
7.2.2.1	Order code
7.2.2.2	Information on sequencing order
7.2.3	Date and time
7.2.3.1	General
7.2.3.2	Order date
7.2.3.3	Order received date
7.2.3.4	Specimen collection date
7.2.3.5	Report date
7.2.3.6	Addendum creation date
7.3	Information on subject of care
7.3.1	General
7.3.2	Subject of care identifier
7.3.3	Subject of care name
7.3.4	Subject of care birth date
7.3.5	Subject of care sex
7.3.6	Subject of care ancestry
7.3.7	Referring diagnosis
7.4	Information on legally authorized person ordering clinical sequencing
7.4.1	General
7.5	Performing laboratory
7.5.1	General
7.5.2	Basic information on performing laboratory
7.5.3	Information on report generator
7.5.4	Information of legally confirmed person on sequencing report
7.6	Biospecimen information
7.6.1	General
7.6.2	Type of specimen
7.7	Fusion gene information
7.7.1	General

- 7.7.2 Gene and its partner gene information
 - 7.7.3 Chromosome information
 - 7.7.4 Breakpoints information
 - 7.8 Classification of variants
 - 7.9 Recommend treatment
 - 7.9.1 General
 - 7.9.2 Medication
 - 7.9.3 Clinical trial information
 - 7.9.4 Other recommendation
 - 7.9.5 Supporting information
 - 8 Fields and their nomenclature of optional data
 - 8.1 General
 - 8.2 Associated disease and phenotypes
 - 8.3 Reference genome version
 - 8.4 Genomic information related to race
 - 8.5 Fusion gene information
 - 8.5.1 Fusion gene ID
 - 8.5.2 HGVS version
 - 8.5.3 Fusion transcript image
 - 8.5.4 Read counts for evidence reads (or supporting reads)
 - 8.5.5 Count of total fusion events
 - 8.5.6 Type of fusion
 - 8.5.7 Validation using standard method (RT-PCR)
 - 8.6 Detailed sequencing information
 - 8.6.1 Clinical sequencing date
 - 8.6.2 Quality control metrics
 - 8.6.3 Sequencing platform information
 - 8.6.3.1 General
 - 8.6.3.2 Type of sequencers
 - 8.6.3.3 Library preparation methods
 - 8.6.3.4 Target capture methods
 - 8.6.3.5 Read type
 - 8.6.3.6 Read length
 - 8.6.4 Analysis platform information
 - 8.6.4.1 General
 - 8.6.4.2 Alignment tools
 - 8.6.4.3 Fusion gene caller
 - 8.6.4.4 Annotation tools and databases
 - 8.7 References
- Annex A (informative) Example structure of clinical sequencing report**

Page count: 21