

ISO/TS 20428:2017-05 (E)

Health informatics - Data elements and their metadata for describing structured clinical genomic sequence information in electronic health records

Contents		Page
Foreword		v
Introduction		vi
1	Scope	1
2	Normative references	1
3	Terms and definitions	1
4	Abbreviated terms	5
5	Use case scenario	6
6	Composition of a clinical sequencing report	7
6.1	General	7
6.2	Overall interpretation in summary	8
6.3	Detailed contents	8
7	Fields and their nomenclature of required data	9
7.1	General	9
7.2	Clinical sequencing orders	10
7.2.1	General	10
7.2.2	Clinical sequencing order code	10
7.2.3	Date and time	10
7.2.4	Specimen information	11
7.3	Information on subject of care	11
7.3.1	General	11
7.3.2	Subject of care identifiers	11
7.3.3	Subject of care name	11
7.3.4	Subject of care birth date	11
7.3.5	Subject of care sex	11
7.3.6	Subject of care ethnicity	11
7.4	Information on legally authorized person ordering clinical sequencing	11
7.4.1	General	11
7.5	Performing laboratory	12
7.5.1	General	12
7.5.2	Basic information on performing laboratory	12
7.5.3	Information on report generator	12
7.5.4	Information of legally confirmed person on sequencing report	12
7.6	Associated diseases and phenotypes	12
7.7	Biomaterial information	12
7.7.1	General	12
7.7.2	Types of sample	12
7.7.3	Genomic source class in biomaterial	12
7.7.4	Conditions of specimen that may limit adequacy of testing	12
7.8	Genetic variations	13
7.8.1	General	13
7.8.2	Gene symbols and names	13
7.8.3	Sequence variation information	13
7.9	Classification of variants	14

7.9.1	General	14
7.9.2	Classification of variants based on the pathogeny	14
7.9.3	Classification of variants based on clinical relevance	15
7.10	Recommended treatment	15
7.10.1	General	15
7.10.2	Classification of variants based on clinical relevance	15
7.10.3	Clinical trial information	15
7.10.4	Known protocols related to a variant	16
7.10.5	Other recommendation	16
7.11	Addendum	16
8	Fields and their nomenclature of optional data	16
8.1	General	16
8.2	Medical history	17
8.3	Family history/Pedigree information	17
8.4	Reference genome version	17
8.5	Racial genomic information	18
8.6	Genetic variation	18
8.7	Detailed sequencing information	18
8.7.1	Clinical sequencing date	18
8.7.2	Quality control metrics	18
8.7.3	Base calling information	18
8.7.4	Sequencing platform information	19
8.7.5	Analysis platform information	20
8.8	References	20
Annex A (informative)	Example structure of clinical sequencing report	21
Annex B (informative)	Example layout of clinical sequencing report	28
Bibliography	32