Technical Specification

ISO/TS 20738

Genomics informatics — Requirements of data analysis for direct-to-consumer testing

Informatique génomique — Exigences d'analyse des données pour les tests en libre accès

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Contents

Page

Foreword

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This document was prepared by Technical Committee ISO/TC 215, *Health informatics*, Subcommittee SC 1, *Genomics Informatics*.

Any feedback or questions on this document should be directed to the user's national standards body. A complete listing of these bodies can be found at www.iso.org/members.html.

Introduction

With increasing people's awareness of their right to know their own body and of the need for disease prevention, prediction, participation and personalized treatment, and with the rapid development of sequencing technology, genetic testing has expanded from clinical application to general consumer application. Direct-to-customer (DTC) testing refers to genetic testing that individuals can order without needing a clinician or a health care provider. These tests typically analyze DNA from a sample – often saliva – to provide insights into various genetic traits.

DTC tests cover a wide range of genetic analyses, including ancestry and heritage (understanding ethnic background and lineage), health and disease risk (identifying genetic predispositions to conditions such as cancer or heart disease), traits and lifestyle (examining genetic influences on taste preferences, hair loss, or lactose digestion), pharmacogenomics (assessing how genetic variations affect drug metabolism). DTC testing improves the awareness and attention to certain diseases, and it allows to adjust existing precaution under the guidance of professionals. It provides the necessary basis for the formation of personalized disease prevention programs. As an increasing prevalent commonality that connects clinical care and lifestyle, DTC testing has grown enormously both in practical and expected use, becoming more and more indispensable in the genetic testing ecosystem.

This document is based on current DTC industry data, combined with the needs of upstream and downstream industry users. It puts forward general requirements and suggestions on the data and technical content of genotype imputation technology, analysis and interpretation of results, as well as specific requirements in the development of a supporting evaluation model and database. With this document's specifications as the basis of data analysis in the development of DTC testing products and services, consumers can have greater confidence in the conclusions drawn from the data, thereby facilitating greater confidence in DTC testing.

Genomics informatics — Requirements of data analysis for direct-to-consumer testing

1 Scope

This document specifies the requirements for genetic data analysis relating to direct-to-consumer (DTC) testing, including preprocessing, detection site, evaluation models, the use of databases and the elements of assessment reports.

This document applies to the analysis of genetic data from DTC testing without the involvement of a health care provider.

2 Normative references

There are no normative references in this document.

3 Terms and definitions

For the purposes of this document, the following terms and definitions apply.

ISO and IEC maintain terminology databases for use in standardization at the following addresses:

— ISO Online browsing platform: available at https://www.iso.org/obp

— IEC Electropedia: available at https://www.electropedia.org/

3.1
coverage
coverage depth
number of times that a given base position is read in a sequencing run

Note 1 to entry: The number of reads that cover a particular position.

[SOURCE: ISO 20397-2:2021 3.6]

3.2
DNA chip
DNA microarray
solid substrate where a collection of probe DNA arranged in a specific design is attached in a high-density fashion, directly or indirectly, that assays large amounts of biological material using high-throughput screening methods

[SOURCE: ISO 16577:2022 3.4.13]

3.3
direct-to-customer
DTC
retail business model which eliminates any intermediaries and sells direct to consumer

Note 1 to entry: Also referred to as business to consumer (B2C).

Note 2 to entry: The sample, blood, saliva, cheek swab (cells from buccal cavity), fecal matter, nail clipping, are provided by the consumer in assumed accordance with the collection protocol provided by the business.