ICH E18: Guideline for Genomic Sampling and Management of Genomic Data

Prepared by the ICH E18 Expert Working Group

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Legal Notice

Outline

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Background

- Awareness of, and interest in, genomic data obtained from clinical studies are growing.
- Regulatory agencies in the ICH regions have independently published guidelines encouraging genomic sample collection.
- Lack of a harmonised ICH Guideline impedes collecting genomic samples and conducting genomic research in a consistent manner in global clinical studies.

Background (continued)

- Genomic samples may be used for a variety of analyses, including single genes, sets of genes and the whole genome, which may or may not be pre-specified in the clinical study objectives at the time of collection.
- Storage of genomic samples and data in clinical studies may be subject to national laws and regulations.

Guideline Objectives

- To provide harmonised principles of genomic sampling and management of genomic data in clinical studies.
- To facilitate the implementation of genomic studies by enabling a common understanding of critical parameters.
- To increase awareness and provide considerations regarding subjects' privacy, data protection, informed consent, and transparency and communication of findings.
- To foster interactions amongst stakeholders, including drug developers, investigators and regulators.

Scope

- Technical aspects and principles of genomic sampling and management of genomic data from interventional and non-interventional clinical studies.
  - Irrespective of the timing of analyses
  - Both pre-specified and non-pre-specified use
- Principles may apply to any genomic research using human-derived materials.

General Principles

- Genomic sample acquisition is encouraged, ideally, in all subjects from all phases and studies of clinical development.
- Maintain genomic sample integrity to ensure scientific utility.
- Standardised practice for handling and processing of genomic samples to foster integration of data from different platforms.
- Genomic data should be treated with the same high standards of confidentiality as other clinical data.
- Secure storage and controlled access to genomic samples and data.

Overview of Guideline Content

- Genomic Sampling
  - Genomic research objectives will determine the specimen, analyte(s) and methods to be used.
  - Sample quantity and quality influence the reliability of the genomic data.
  - Standardised sample collection and handling procedures improve sample and data quality.
  - Samples should not be exposed to conditions that may affect stability of nucleic acid targets during transport and storage.

Overview of Guideline Content (continued)

- Genomic sampling (continued)
  - Consider pre-analytical variables when developing a strategy for sample collection, processing, transport, and storage
    - Specimen type
    - Timing of Specimen Collection
    - Specimen Preservation Conditions
    - Sample Stability and Degradation
    - Specimen Volume and Composition
    - Parameters Influencing Genomic Sample Quality and Quantity
    - Sources of Interference
    - Transport of Samples
    - Storage of Samples
    - Curation of Sample Inventory

Overview of Guideline Content (continued)

- Genomic Data
  - The type of genomic data generated depends on the analytes and the applied technology platforms.
  - The appropriate platform and method (research grade versus validated) should be chosen in light of the intended purpose of the genomic data.
  - The processing and analytical workflow for all stages of analysis should be documented, and appropriate quality control (QC) procedures and metric thresholds should be set for all stages.
  - A robust quality management system should be in place.

Overview of Guideline Content (continued)

- Genomic Data (continued)
  - The use of publicly available annotation resources is highly recommended to enable cross-platform comparisons and integration of genomic and non-genomic (e.g., proteomic) results from different studies.
  - Data files that maintain the complete features of the primary data should be retained.
  - Destruction of data contradicts the principles of scientific integrity.
  - It is recommended to retain data once generated and used in a study.
  - Where applicable, procedures may have to be developed to enable desired disposition of genomic data at request of the subject.

Overview of Guideline Content (continued)

- Privacy and Confidentiality
  - Processing and handling of genomic samples and data should be conducted in a manner that protects the subjects' privacy.
  - Single coding is recommended for genomic samples and data.
  - Anonymisation is not recommended.
  - Strict control of access rights to genomic samples and data should be established (similar to other clinical data).

Overview of Guideline Content (continued)

- Informed Consent
  - Informed consent should be consistent with ICH E6, and comply with local legislation.
  - Consent for genomic research can be included in the consent for the clinical study or obtained separately.
  - Identification of common and essential elements for a globally acceptable informed consent would facilitate genomic sampling.
  - Informed consent should permit broad sample analysis regardless of timing (e.g., sets of genes, transcriptome analysis).
  - Consent should allow for broad sample use (e.g., assay development, disease research).

Overview of Guideline Content (continued)

- Communication of Findings
  - Sponsors should adopt a position regarding return of findings to subjects.
  - When communicating results to subjects, the pertinence of genetic counseling should be evaluated.
  - The subjects' desire and consent to receive genomic information or not should be respected.
  - Local and regional guidances and legislation may apply.

Considerations

- No detailed guidance is included in biobanking regulations, ethical aspects, or issues related to privacy/data protection as these are governed by the principles of the Declaration of Helsinki and national rules and regulations.

Implementation

- The guideline intends to facilitate the implementation of genomic studies by enabling a common understanding of critical parameters for the unbiased collection, storage, and optimal use of genomic samples and data.
- The recommendations in this guideline are principles and they should be interpreted in accordance with legislations, regulations as well as policies in each jurisdiction where genomic research is undertaken.

THANK YOU !