

# Bioinformatics metadata

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- Elements
  - Immutable reference files
    - \* Possible PHI
    - \* Provenance
  - Mutable "sample sheet"
    - \* xlsx or series of tsvs
    - \* No PHI
    - \* Source of truth
    - \* Validation input
- No PHI
- With Libreoffice calc
  - Use Excel A1 formula syntax (Tools -> Options -> LibreOffice Calc -> Formula -> Formula Options -> Formula syntax)

# Logical data representation, Common Data Model schema

## Documentation

Good Best Practices for Bioinformatics Metadata Schema with Frictionless

- Components
  - Excel as the user interface: non-programmers enter data, schema validates it.  
Hierarchical data model: organize by biological and technical relationships
  - schema yaml  
Self-documenting: schemas generate data dictionaries programmatically
- Schema Structure
  - Version control by repo-level versioning
  - Define Shared Elements Once, Use YAML anchors for repeated definitions:
  - Basic Resource Structure
  - Field Definition Principles
    - \* Every field should include:
      - name: Machine-readable identifier
      - title: Human-readable label
      - description: Clear explanation including format requirements and units
      - example: Representative value
      - type: Data type
      - constraints: Validation rules

- Example:
- Primary Key Conventions
  - \* Use descriptive prefix plus zero-padded four-digit numbers:
    - Enables lexicographic sorting
    - Format: `^[prefix]4$`
    - Examples: `subj0001`, `samp0001`, `lib0001`, `run0001`
    - Always include pattern constraint to catch typos.
- Foreign Key Conventions
  - \* Foreign key fields should be minimal:
  - \* Include pattern constraint to validate format before checking referential integrity.
  - \* Detailed metadata lives in the parent table only - avoid redundancy.
- Data Type Decision Principles
  - \* Prefer Enums Over Booleans. Use enums for domain-specific concepts that require explanation:
  - \* Booleans are acceptable only when semantics are universally obvious and require no explanation.
  - \* Document Enum Values
    - Use custom `enumDescriptions` property for programmatic data dictionary generation:
  - \* Standardize Units in Field Names: Include units in field name and description:

- Table Design Principles

- Hierarchical Relationships

- \* Organize tables by biological and technical hierarchy:

- `subjects` → `samples` → `libraries` → `fastqs`

- \* Use foreign keys to enforce referential integrity.

- When to Split Tables

- \* Split into separate tables when entities have:

- Many divergent fields (>5 unique per subtype)
      - Fundamentally different semantics
      - Example: human participants vs mouse subjects

- Keep entities in one table when:

- \* Few optional fields (2-3)
    - \* Structural similarity between subtypes
    - \* Add discriminator field for subtype and leave subtype-specific fields optional:

- Schema Evolution Principles

- Never use "other" catch-all categories or allow arbitrary values - this defeats validation.

- Constraint Defaults

- \* Frictionless defaults are:
      - `required: false` (field is optional)

- unique: false (duplicates allowed)
- No enum restrictions
- Only declare constraints when restricting behavior.
- Field Ordering
  - Primary key first
  - Foreign keys second
  - Required fields
  - Optional fields
  - Group related fields together

**Preamble**

**Subjects**

**Participants**

**Mice**

**Samples**

**Libraries**

**Sequencing runs**

**FASTQs**

`#+end_src`