GENOMICS CANON

2026-03-18

Dexter Hadley, MD/PhD

Hadley Lab CANONIC

Abstract

Example

Contents

GenomicPython (2013) is the proto-CANONIC seed. Python abstracted away GENOMICS is the domain. CANONIC is closure. 12 years later, GenomicPython is resurrected as CANONIC.GENOMICS the proto-CANONIC work finds its completion in the framework.

0.1 2. Genetic Privacy

Genomic data MUST be protected under applicable genetic nondiscrimination and privacy laws.

Example: GINA (Genetic Information Nondiscrimination Act, 2008) prohibits discrimination in health insurance (Title I) and employment (Title II) based on genetic information. Does NOT cover life insurance, disability insurance, or long-term care insurance. State genetic privacy laws vary California Genetic Information Privacy Act (CalGIPA, 2022) requires opt-in consent for DTC genetic testing companies. 23andMe/Ancestry: FTC enforcement for unfair/deceptive practices regarding genetic data. HIPAA covers genetic information as PHI when held by covered entities. EU GDPR genetic data is special category (Art. 9) requiring explicit consent.

0.2 3. Research Governance

Human genomic research MUST comply with the Common Rule and institutional review requirements.

Example: Common Rule (45 CFR 46, revised 2018) informed consent requirements, broad consent option for future research use of biospecimens. IRB oversight expedited review for minimal risk genomic research, full board review for greater than minimal risk. Genomic Data Sharing Policy (NIH, 2014) large-scale genomic data from NIH-funded research must be shared through controlled-access repositories (dbGaP). Biobank governance: material transfer agreements (MTAs), community advisory boards, re-

turn of results policies. NAGPRA (Native American Graves Protection and Repatriation Act) special considerations for Indigenous genomic research.

0.3 4. Clinical Genomics

Clinical genomic testing MUST meet laboratory certification requirements and follow variant classification standards.

Example: CLIA (Clinical Laboratory Improvement Amendments, 42 CFR 493) all clinical labs must be CLIA-certified. CAP (College of American Pathologists) accreditation proficiency testing, laboratory inspections. ACMG/AMP variant classification (2015): Pathogenic, Likely Pathogenic, Variant of Uncertain Significance (VUS), Likely Benign, Benign five-tier system using 28 criteria. ClinVar (NCBI) public archive of variant-disease relationships. Pharmacogenomics: FDA Table of Pharmacogenomic Biomarkers in Drug Labeling (400+ entries), CPIC (Clinical Pharmacogenetics Implementation Consortium) guidelines. Germline vs somatic testing distinctions in clinical reporting.

0.4 5. Data Standards

Genomic data MUST conform to community-accepted standards for format, exchange, and interpretation.

Example: File formats: FASTQ (raw sequence reads), BAM/CRAM (aligned reads, CRAM is compressed), VCF (Variant Call Format variant calls), BED (genomic intervals). GA4GH (Global Alliance for Genomics and Health) frameworks: htsget (streaming genomic data), Beacon protocol (federated variant discovery), Phenopackets (structured phenotype data), DRS (Data Repository Service), TES (Task Execution Service). FHIR Genomics (HL7) integration of genomic data into clinical workflows via MolecularSequence resource. Reference genomes: GRCh38/hg38 (current), T2T-CHM13 (telomere-to-telomere assembly).

0.5 6. IP & Access

Genomic innovations MUST navigate the post-Myriad patent landscape balancing IP protection with access to genetic testing.

Example: AMP v. Myriad Genetics (SCOTUS 2013) naturally occurring DNA segments are products of nature and not patent-eligible (35 USC 101). cDNA (complementary DNA) remains patentable as synthetic creation. Post-Myriad: BRCA testing market opened from Myriad monopoly ($3,000+) to multiple providers ($250-$500). NIH data sharing policies: Genomic Data Sharing Policy (2014), Data Management and Sharing Policy (2023). Open-access databases: gnomAD (population variant frequencies), COSMIC (somatic mutations in cancer), dbSNP, ClinGen (clinical genome resource). Bayh-Dole Act (35 USC 200-212) federally-funded inventions can be patented by research institutions.

---

1. Constraints

```
MUST:     Cite specific regulation or genomic standard
MUST:     Distinguish between research-use and clinica
MUST NOT: Present genomic findings without variant cla
```

GENOMICS | CANON | VERTICALS