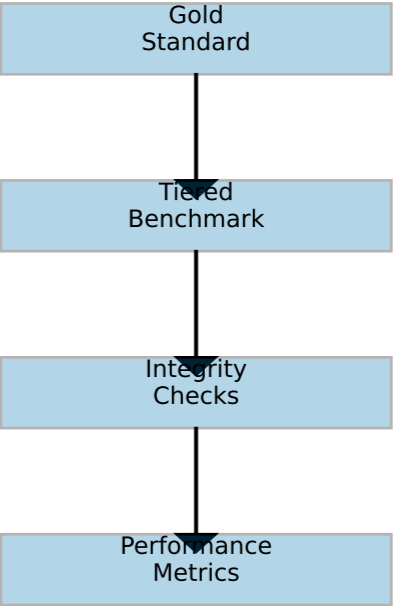


Figure 1: Task Taxonomy and Benchmark Integrity Framework

A. Task Taxonomy

- Task A: GWAS Credible Set → Causal Gene
Input: LD-expanded SNP set from GWAS locus
Output: Ranked list of candidate genes
Methods: Distance, ABC, PoPS, eQTL, Hi-C
- Task B: Enhancer → Gene
Input: Known regulatory element (CRISPRi-validated)
Output: Target gene(s)
Methods: ABC, Hi-C, correlation
- Task C: Variant → Regulatory Element
Input: Individual variant
Output: Regulatory activity prediction
Methods: ChromHMM, DeepSEA, enformer

B. Validation Pipeline



C. Benchmark Integrity Checklist

- ✓

Task-appropriate
- ✓

No training leakage
- ✗

Source diversity
- ✓

Temporal ordering
- ✓

No circular validation
- ✓

Size adequacy

D. Benchmark Statistics

Task A (GWAS → Gene):
Loci: 14,016
Genes: 4,892
Tier-0 (G2P): 1,248 pairs
Tier-1 (ClinVar): 2,764 pairs

Task B (Enhancer → Gene):
Enhancers: 19,825
Genes: 3,141
CRISPRi-validated: 15,692
MPRA-validated: 4,133

Evidence Sources:
G2P/OMIM: 38%
ClinVar: 29%
CRISPR: 21%
MPRA: 12%