

# Figure 1: Task Taxonomy and Benchmark Integrity Framework

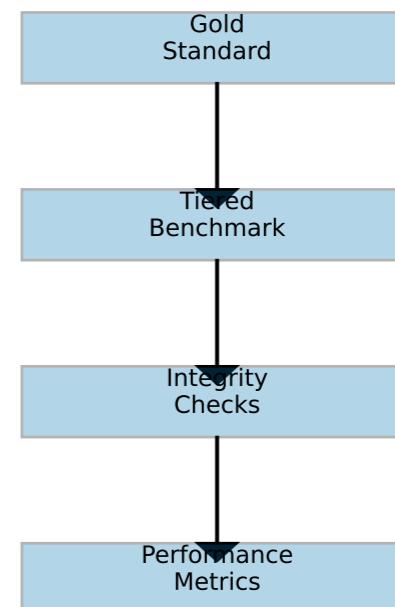
## A. Task Taxonomy

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%