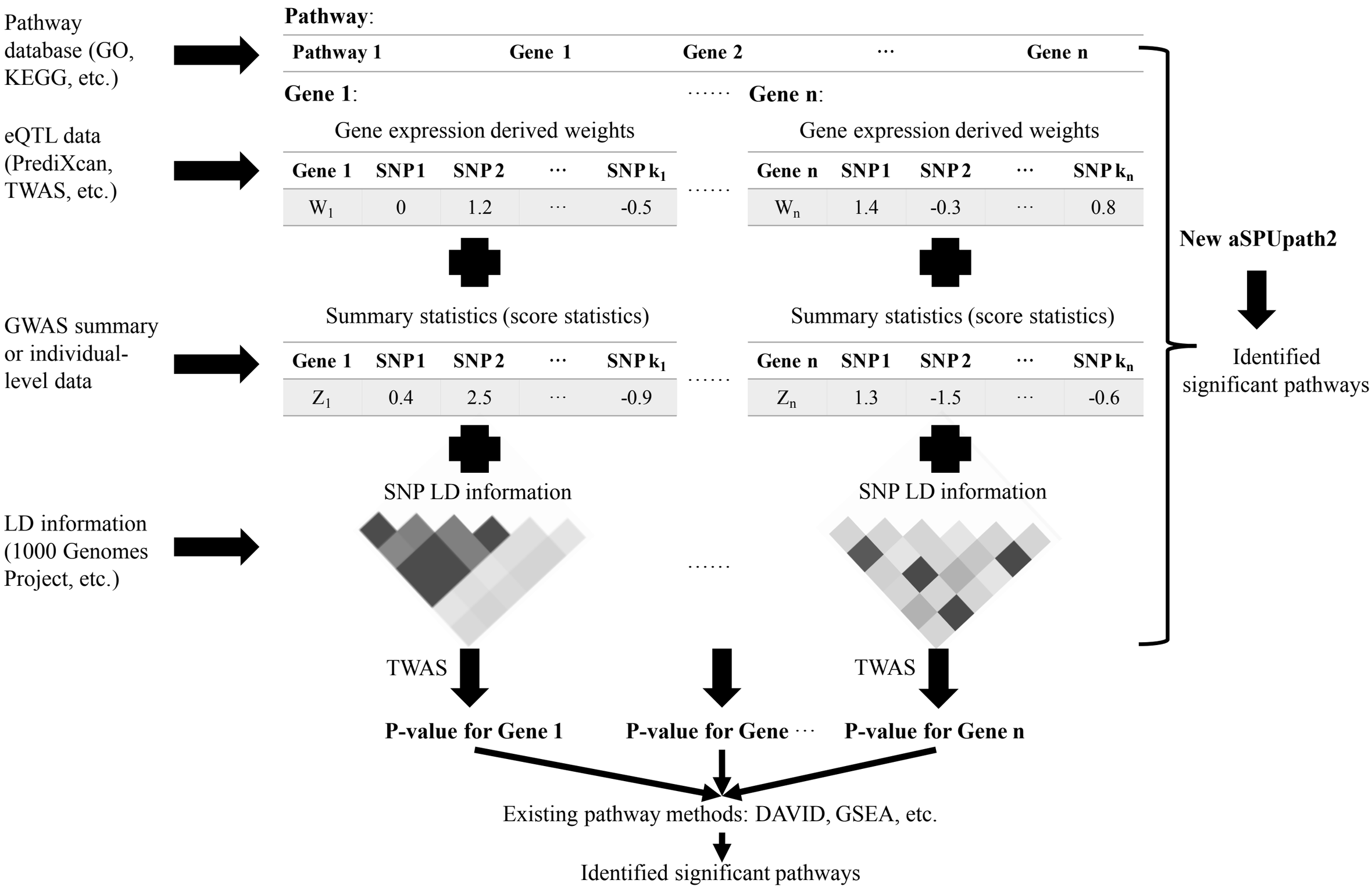
Pathway database (GO, KEGG, etc.)
eQTL data (PrediXcan, TWAS, etc.)
GWAS summary or individual-level data
LD information (1000 Genomes Project, etc.)
Pathway:
Pathway 1
Gene 1
Gene 2
···
Gene n
Gene 1:
Gene n:
Gene expression derived weights
Gene 1 SNP 1 SNP 2 ··· SNP k1
W1 0 1.2 ··· -0.5
Gene n SNP 1 SNP 2 ··· SNP kn
Wn 1.4 -0.3 ··· 0.8
Summary statistics (score statistics)
Gene 1 SNP 1 SNP 2 ··· SNP k1
Z1 0.4 2.5 ··· -0.9
Gene n SNP 1 SNP 2 ··· SNP kn
Zn 1.3 -1.5 ··· -0.6
SNP LD information
TWAS
P-value for Gene 1
P-value for Gene ···
P-value for Gene n
Existing pathway methods: DAVID, GSEA, etc.
Identified significant pathways
New aSPUpath2
Identified significant pathways