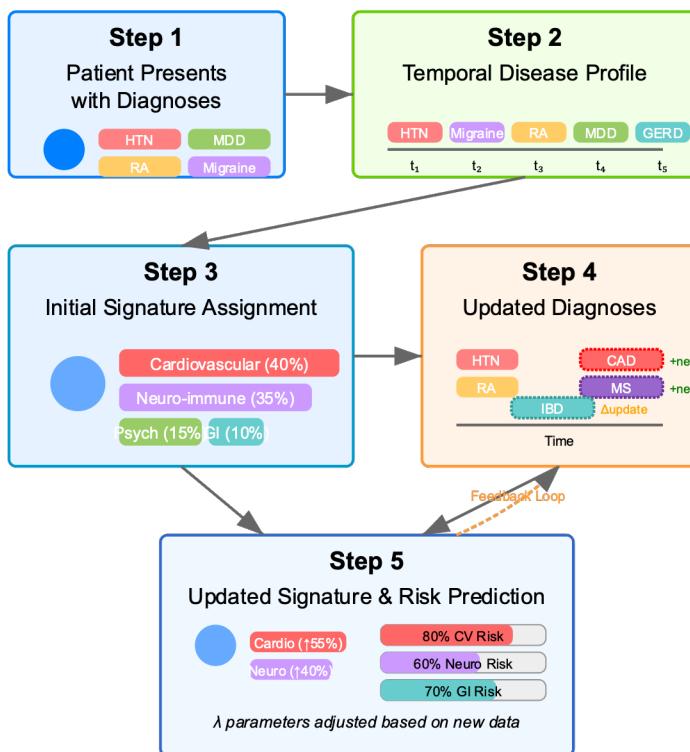


Patient Journey: From Diagnoses to Signature Assignment

Panel 1: Clinical Flow & Model Evolution



Panel 2: Mathematical Framework

AladynNouli Model Core

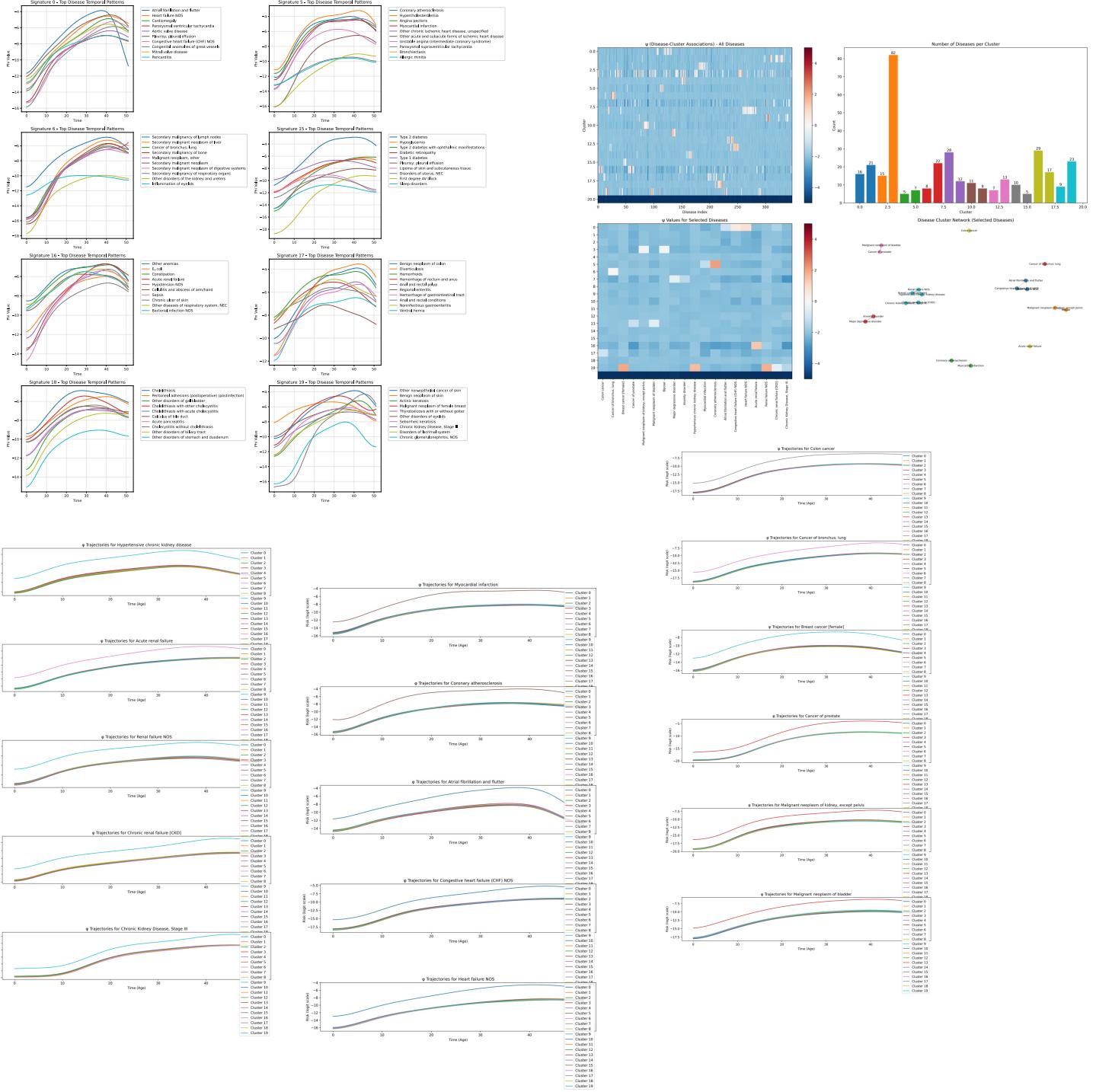
Input Variables:

Model Parameters:

λ Parameter Evolution

Mathematical Flow

Model optimizes $\lambda, \varphi, v, \psi$ to maximize likelihood of observed diagnoses



Calculated Risk of "Coronary atherosclerosis" for Individual 100
(Composition from Signatures)

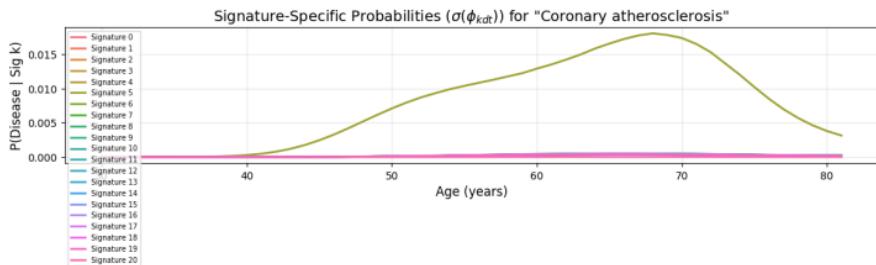
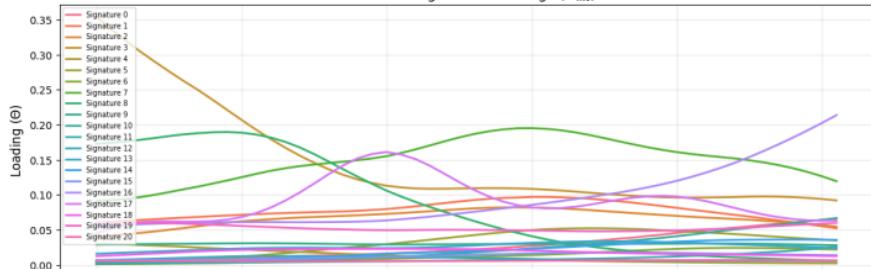
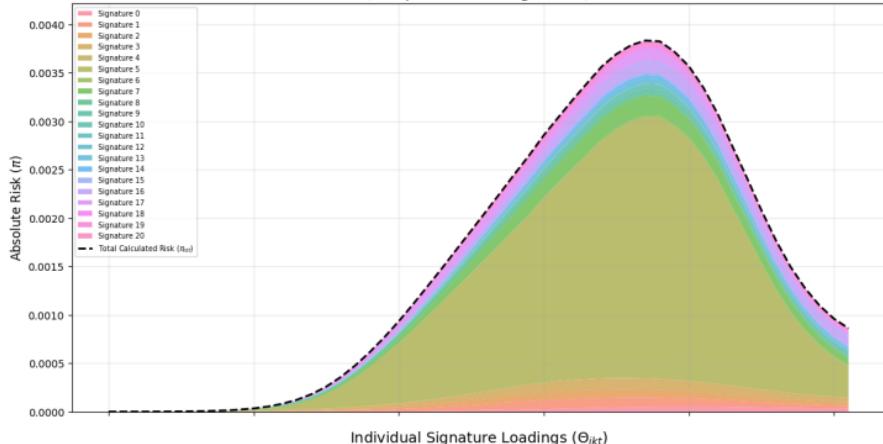
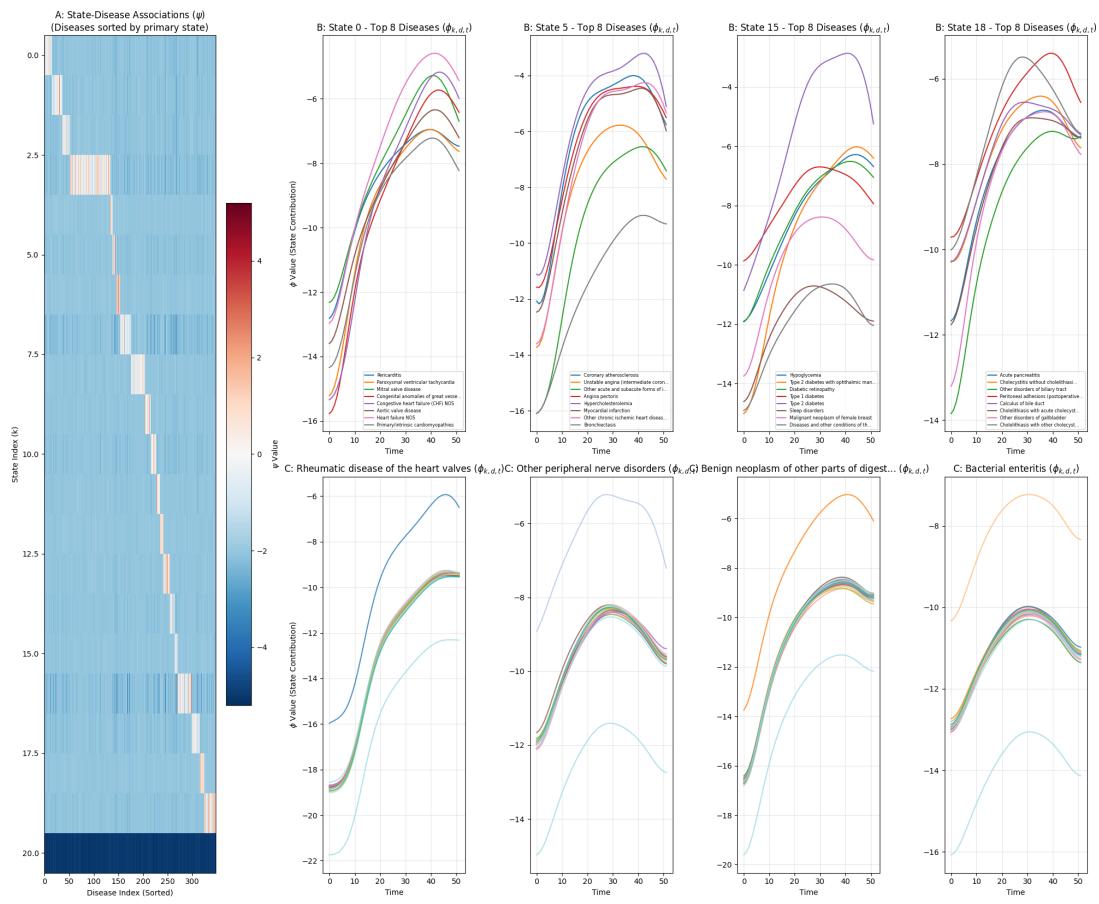
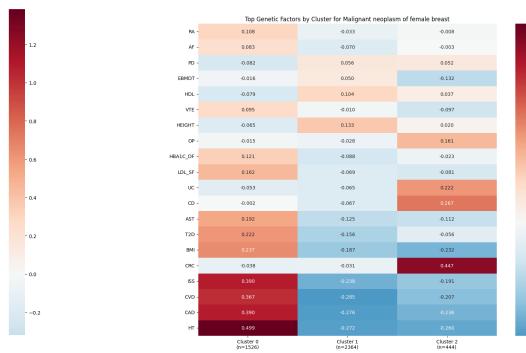
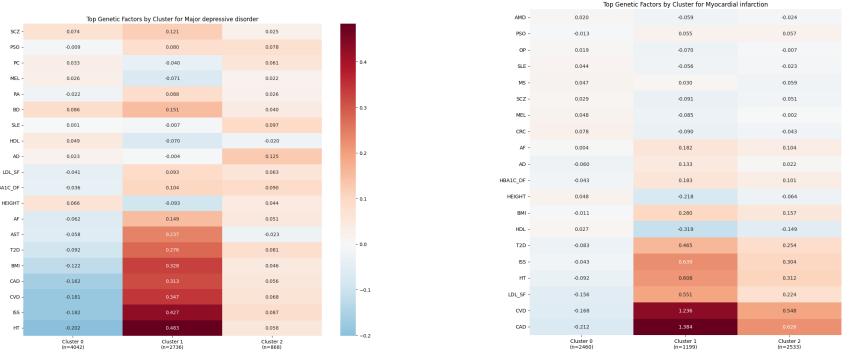
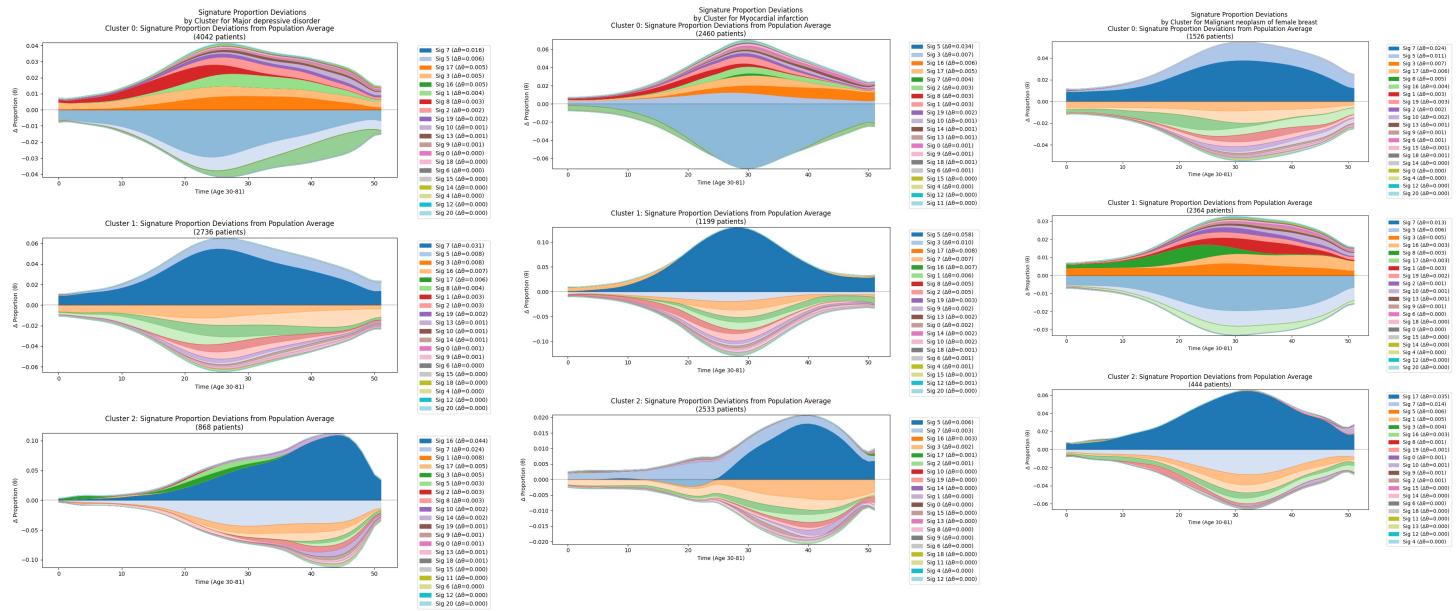
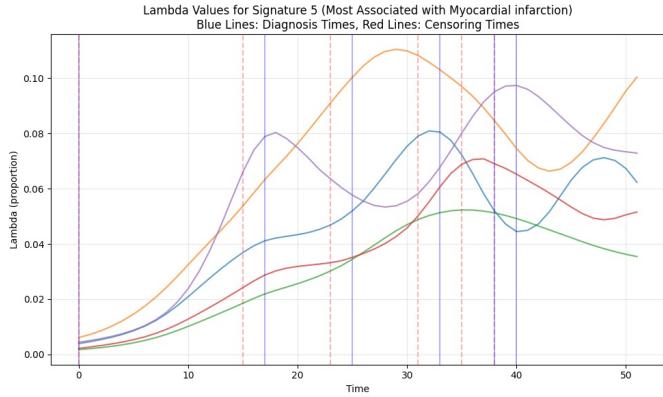


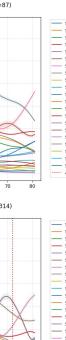
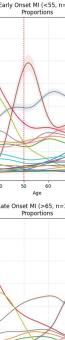
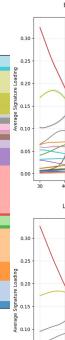
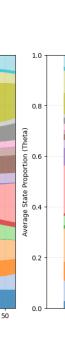
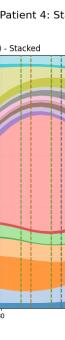
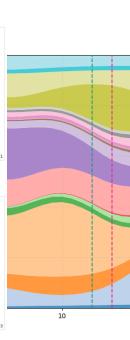
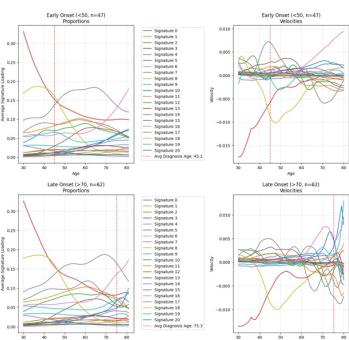
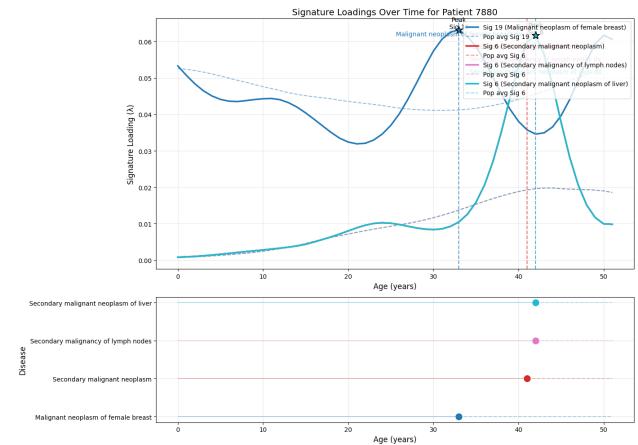
Figure 2: Population-Level State Definitions and Temporal Patterns

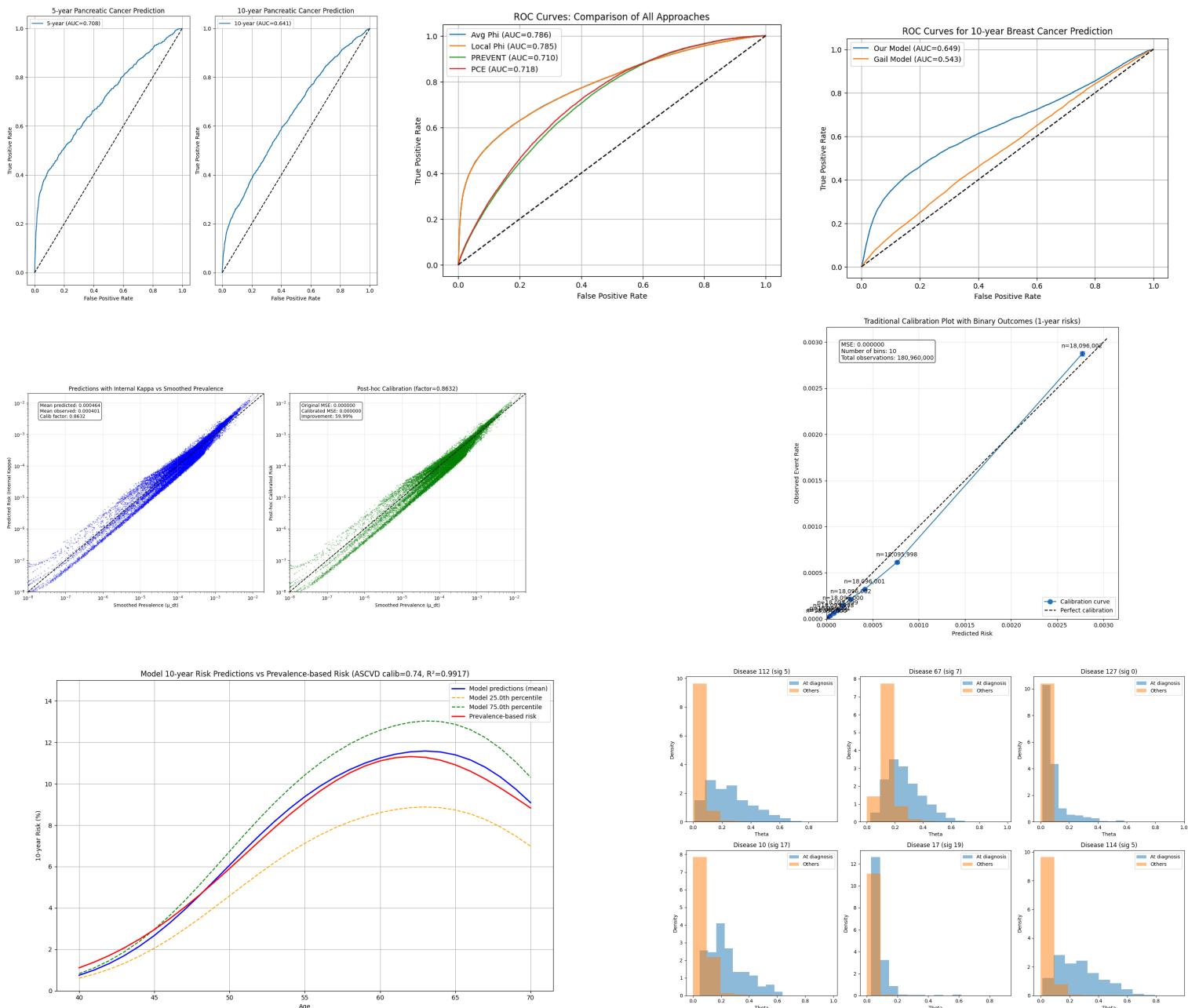






Patient 9384
Patient 4175
Patient 5120
Patient 7971
Patient 7613
Diagnosis Time
Censoring Time





MAGMA Gene-Set Analysis

MAGMA gene-set analysis is performed for curated gene sets and GO terms obtained from MsigDB (see [here](#) for details).

The table displays either significant gene sets with $P_{\text{bon}} < 0.05$ or top 10 gene sets when there are less than 10 significant gene sets. Full results are downloadable from "C" tab.

Note that MAGMA gene-set analyses uses the full distribution of SNP p-values and is different from a pathway enrichment test as implemented in GENE2FUNC that only tests enrichment of prioritized genes.

Show entries

Search:

Gene Set	N genes	Beta
GOBP_CARDIAC_MUSCLE_CELL_CONTRACTION	67	0.50695
GOBP_CELL_COMMUNICATION_INVOLVED_IN_CARDIAC_CONDUCTION	58	0.52455
GOBP_REGULATION_OF_CELL_COMMUNICATION_BY_ELECTRICAL_COUPLING_INVOLVED_IN_CARDIAC_CONDUCTION	12	1.1819
GOBP_CARDIAC_CONDUCTION	91	0.41314
GOBP_REGULATION_OF_ACTIN_FILAMENT_BASED_MOVEMENT	37	0.59892
GOBP_CARDIAC_MUSCLE_CELL_ACTION_POTENTIAL	69	0.44909
GOBP_REGULATION_OF_ACTION_POTENTIAL	53	0.49904
GOBP_ACTIN_MEDIATED_CELL_CONTRACTION	95	0.35262
GOBP_BUNDLE_OF_HIS_CELL_TO_PURKINJE_MYOCYTE_COMMUNICATION	14	0.97912
GOBP_NEGATIVE_REGULATION_OF BIOSYNTHETIC_PROCESS	1442	0.086634

Showing 1 to 10 of 10 entries

Previous

Sig 7

Genomic Locus	IndSigSNP	chr	bp	rsID	PMID	Trait	FirstAuth	Date	P-value
8	rs76752100	10	104871279	rs10883832	31835028	Anorexia nervosa, attention-deficit/hyperactivity disorder, autism spectrum disorder, bipolar disorder, major depression, obsessive-compulsive disorder, schizophrenia, or Tourette syndrome (pleiotropy)	Cross-Disorder Group of the Psychiatric Genomics Consortium	2019-12-01	3E-14
11	rs10139018	14	75186010	rs7147721	33859377	Depression	Thorp JG	2021-04-15	3E-18
11	rs10139018	14	75186010	rs7147721	35898629	Decaffeinated coffee consumption or major depression disorder	Yin B	2022-07-11	1E-11
11	rs10139018	14	75296831	rs10148293	29662059	Depression (broad)	Howard DM	2018-04-16	1E-7
11	rs10139018	14	75377555	rs2003490	31835028	Anorexia nervosa, attention-deficit/hyperactivity disorder, autism spectrum disorder, bipolar disorder, major depression, obsessive-compulsive disorder, schizophrenia, or Tourette syndrome (pleiotropy)	Cross-Disorder Group of the Psychiatric Genomics Consortium	2019-12-01	2E-8

Showing 1 to 5 of 5 entries (filtered from 1,761 total entries)

Previous 1 Next

