

Autism

Table 1: The 40 highest-scoring gene sets ranked by TEMPO score for the ASD data set.

Rank	Gene Set	Control MSE	Score	MSE p-value	Score p-value	Score BH
1	peptidyl-tyrosine phosphorylation	6.573	2.972	0.012	0.004	0.099
2	peptidyl-tyrosine modification	6.573	2.972	0.014	0.004	0.099
3	phosphatidylcholine metabolic process	5.472	2.851	0.002	0.002	0.099
4	transcription elongation from RNA polymerase II promoter	6.823	2.729	0.010	0.008	0.099
5	ammonium ion metabolic process	7.238	2.649	0.042	0.008	0.099
6	double-strand break repair via nonhomologous end joining	6.351	2.647	0.004	0.004	0.099
7	ethanolamine-containing compound metabolic process	6.095	2.622	0.002	0.010	0.099
8	positive regulation of apoptotic signaling pathway	5.965	2.601	0.012	0.008	0.099
9	regulation of apoptotic signaling pathway	5.516	2.595	0.002	0.016	0.099
10	regulation of myeloid cell differentiation	7.081	2.586	0.016	0.008	0.099
11	non-recombinational repair	6.352	2.561	0.002	0.004	0.099
12	protein monoubiquitination	6.548	2.554	0.002	0.006	0.099
13	leukocyte cell-cell adhesion	6.913	2.537	0.022	0.010	0.099
14	positive regulation of transporter activity	7.933	2.528	0.042	0.012	0.099
15	alcohol metabolic process	6.294	2.516	0.006	0.036	0.101
16	cell cycle arrest	5.903	2.483	0.002	0.022	0.099
17	intrinsic apoptotic signaling pathway	5.805	2.478	0.004	0.026	0.099
18	stress-activated protein kinase signaling cascade	6.854	2.455	0.020	0.026	0.099
19	stress-activated MAPK cascade	6.854	2.455	0.020	0.022	0.099
20	glycerophospholipid metabolic process	6.617	2.447	0.020	0.040	0.106
21	regulation of leukocyte differentiation	7.183	2.439	0.036	0.026	0.099
22	phosphatidylserine acyl-chain remodeling	8.574	2.420	0.026	0.002	0.099
23	negative regulation of cell proliferation	6.280	2.420	0.004	0.044	0.110
24	positive regulation of mitochondrion organization	6.485	2.380	0.010	0.036	0.101
25	regulation of intrinsic apoptotic signaling pathway	6.780	2.369	0.012	0.032	0.099
26	nuclear import	6.957	2.353	0.008	0.018	0.099
27	protein acetylation	7.439	2.320	0.032	0.032	0.099
28	leukocyte migration involved in inflammatory response	7.534	2.310	0.004	0.002	0.099
29	positive regulation of leukocyte differentiation	7.150	2.309	0.022	0.036	0.101
30	peptidyl-lysine acetylation	7.269	2.302	0.018	0.026	0.099
31	membrane budding	7.199	2.296	0.034	0.042	0.107
32	regulation of Ras protein signal transduction	7.520	2.294	0.026	0.022	0.099
33	protein import	7.461	2.288	0.032	0.046	0.111
34	regulation of organelle assembly	6.741	2.275	0.008	0.026	0.099
35	internal protein amino acid acetylation	7.278	2.260	0.012	0.022	0.099
36	negative regulation of viral genome replication	7.849	2.252	0.040	0.030	0.099
37	mitochondrial fusion	7.055	2.245	0.002	0.002	0.099
38	protein targeting to mitochondrion	7.454	2.244	0.016	0.026	0.099
39	regulation of myeloid leukocyte differentiation	6.999	2.237	0.014	0.026	0.099
40	positive regulation of lymphocyte migration	8.102	2.219	0.028	0.018	0.099

Table 2: The 40 highest-scoring upregulated gene sets returned by GSEA in ASD

Rank	Gene Set	NES	p-value	FDR
1	dorsal/ventral axis specification	-1.91	0.00	1.00
2	primary alcohol metabolic process	-1.89	0.00	1.00
3	regulation of chondrocyte differentiation	-1.89	0.00	0.78
4	positive regulation of chondrocyte differentiation	-1.88	0.00	0.67
5	positive regulation of camp biosynthetic process	-1.87	0.00	0.65
6	positive regulation of cyclic nucleotide biosynthetic process	-1.86	0.00	0.56
7	positive regulation of adenylate cyclase activity	-1.86	0.00	0.49
8	organ induction	-1.84	0.00	0.61
9	regulation of carbohydrate biosynthetic process	-1.84	0.00	0.54
10	positive regulation of smoothened signaling pathway	-1.81	0.00	0.67
11	positive regulation of lyase activity	-1.81	0.00	0.61
12	negative regulation of microtubule polymerization	-1.81	0.00	0.60
13	somatic motor neuron differentiation	-1.80	0.00	0.58
14	positive regulation of nucleotide biosynthetic process	-1.80	0.00	0.57
15	positive regulation of purine nucleotide biosynthetic process	-1.80	0.00	0.53
16	modulation by host of viral release from host cell	-1.79	0.00	0.51
17	positive regulation by host of viral release from host cell	-1.79	0.00	0.48
18	phototransduction	-1.79	0.00	0.47
19	negative regulation of t cell apoptotic process	-1.79	0.00	0.46
20	activation of adenylate cyclase activity	-1.79	0.00	0.44
21	negative regulation of platelet-derived growth factor receptor signaling pathway	-1.78	0.01	0.45
22	phototransduction, visible light	-1.77	0.00	0.47
23	notochord development	-1.77	0.01	0.46
24	apical junction assembly	-1.77	0.00	0.45
25	triglyceride catabolic process	-1.76	0.01	0.47
26	developmental induction	-1.76	0.00	0.48
27	regulation of cartilage development	-1.76	0.00	0.48
28	detection of light stimulus	-1.74	0.00	0.54
29	spinal cord development	-1.73	0.00	0.58
30	acid secretion	-1.73	0.00	0.56
31	digestion	-1.73	0.00	0.55
32	positive regulation of protein kinase a signaling	-1.73	0.00	0.55
33	lateral mesoderm development	-1.73	0.01	0.53
34	neutral lipid catabolic process	-1.73	0.01	0.53
35	acylglycerol catabolic process	-1.73	0.01	0.51
36	cyclic nucleotide metabolic process	-1.72	0.01	0.53
37	positive regulation of camp metabolic process	-1.71	0.00	0.58
38	detection of visible light	-1.71	0.00	0.57
39	negative regulation of vascular permeability	-1.71	0.01	0.57
40	aspartate transport	-1.71	0.01	0.56

Table 3: The 40 highest-scoring gene sets returned by maSigPro+GSEA preranked in ASD

Rank	Gene Set	NES	p-value	FDR
1	glutaminyl-trnagln biosynthesis via transamidation	1.48	0.00	1.00
2	vitamin b6 metabolic process	1.47	0.00	1.00
3	regulation of cortisol secretion	1.47	0.00	0.83
4	positive regulation of cortisol secretion	1.47	0.00	0.64
5	positive regulation of glucocorticoid secretion	1.46	0.00	0.60
6	pyridoxal phosphate metabolic process	1.46	0.01	0.54
7	cell-cell signaling involved in cell fate commitment	1.45	0.01	0.67
8	pyridoxal phosphate biosynthetic process	1.44	0.01	0.73
9	negative regulation of cortisol biosynthetic process	1.42	0.01	1.00
10	histone h2b conserved c-terminal lysine ubiquitination	1.42	0.01	1.00
11	negative regulation of tooth mineralization	1.42	0.01	1.00
12	negative regulation of aldosterone metabolic process	1.42	0.01	0.96
13	negative regulation of glucocorticoid biosynthetic process	1.42	0.00	0.89
14	sequestering of triglyceride	1.42	0.00	0.84
15	negative regulation of aldosterone biosynthetic process	1.41	0.01	0.80
16	negative regulation of steroid hormone biosynthetic process	1.41	0.00	0.81
17	aldehyde biosynthetic process	1.41	0.00	0.80
18	histone h3-k4 demethylation	1.40	0.01	0.91
19	negative regulation of glucocorticoid metabolic process	1.40	0.01	0.87
20	oxygen transport	1.40	0.00	0.97
21	cytoplasmic pattern recognition receptor signaling pathway in response to virus	1.39	0.01	0.99
22	regulation of toll-like receptor 9 signaling pathway	1.39	0.01	1.00
23	regulation of glucocorticoid metabolic process	1.39	0.00	1.00
24	udp-galactose transport	1.38	0.00	1.00
25	nephric duct morphogenesis	1.38	0.00	1.00
26	fatty acid beta-oxidation using acyl-coa dehydrogenase	1.38	0.01	1.00
27	negative regulation of endothelial cell differentiation	1.37	0.02	1.00
28	regulation of low-density lipoprotein particle clearance	1.37	0.00	1.00
29	phospholipase c-activating g-protein coupled acetylcholine receptor signaling pathway	1.37	0.00	1.00
30	regulation of glucocorticoid biosynthetic process	1.37	0.01	1.00
31	regulation of cortisol biosynthetic process	1.37	0.00	1.00
32	positive regulation of chaperone-mediated protein complex assembly	1.37	0.02	1.00
33	negative regulation of epithelial cell differentiation involved in kidney development	1.37	0.00	1.00
34	negative regulation of histone h3-k36 methylation	1.36	0.01	1.00
35	negative regulation of nephron tubule epithelial cell differentiation	1.36	0.00	1.00
36	negative regulation of interleukin-2 secretion	1.36	0.01	1.00
37	negative regulation of hormone biosynthetic process	1.36	0.00	1.00
38	regulation of chaperone-mediated protein complex assembly	1.36	0.02	1.00
39	regulation of type iii interferon production	1.35	0.03	1.00
40	urinary tract smooth muscle contraction	1.35	0.02	1.00

Alzheimer's Disease

Table 4: The 40 highest-scoring gene sets ranked by TEMPO score for the Alzheimer's data set.

Rank	Gene Set	Control MSE	Score	MSE p-value	Score p-value	Score BH
1	peptidyl-tyrosine phosphorylation	6.573	2.972	0.012	0.004	0.099
2	peptidyl-tyrosine modification	6.573	2.972	0.014	0.004	0.099
3	phosphatidylcholine metabolic process	5.472	2.851	0.002	0.002	0.099
4	transcription elongation from RNA polymerase II promoter	6.823	2.729	0.010	0.008	0.099
5	ammonium ion metabolic process	7.238	2.649	0.042	0.008	0.099
6	double-strand break repair via nonhomologous end joining	6.351	2.647	0.004	0.004	0.099
7	ethanolamine-containing compound metabolic process	6.095	2.622	0.002	0.010	0.099
8	positive regulation of apoptotic signaling pathway	5.965	2.601	0.012	0.008	0.099
9	regulation of apoptotic signaling pathway	5.516	2.595	0.002	0.016	0.099
10	regulation of myeloid cell differentiation	7.081	2.586	0.016	0.008	0.099
11	non-recombinational repair	6.352	2.561	0.002	0.004	0.099
12	protein monoubiquitination	6.548	2.554	0.002	0.006	0.099
13	leukocyte cell-cell adhesion	6.913	2.537	0.022	0.010	0.099
14	positive regulation of transporter activity	7.933	2.528	0.042	0.012	0.099
15	alcohol metabolic process	6.294	2.516	0.006	0.036	0.101
16	cell cycle arrest	5.903	2.483	0.002	0.022	0.099
17	intrinsic apoptotic signaling pathway	5.805	2.478	0.004	0.026	0.099
18	stress-activated protein kinase signaling cascade	6.854	2.455	0.020	0.026	0.099
19	stress-activated MAPK cascade	6.854	2.455	0.020	0.022	0.099
20	glycerophospholipid metabolic process	6.617	2.447	0.020	0.040	0.106
21	regulation of leukocyte differentiation	7.183	2.439	0.036	0.026	0.099
22	phosphatidylserine acyl-chain remodeling	8.574	2.420	0.026	0.002	0.099
23	negative regulation of cell proliferation	6.280	2.420	0.004	0.044	0.110
24	positive regulation of mitochondrion organization	6.485	2.380	0.010	0.036	0.101
25	regulation of intrinsic apoptotic signaling pathway	6.780	2.369	0.012	0.032	0.099
26	nuclear import	6.957	2.353	0.008	0.018	0.099
27	protein acetylation	7.439	2.320	0.032	0.032	0.099
28	leukocyte migration involved in inflammatory response	7.534	2.310	0.004	0.002	0.099
29	positive regulation of leukocyte differentiation	7.150	2.309	0.022	0.036	0.101
30	peptidyl-lysine acetylation	7.269	2.302	0.018	0.026	0.099
31	membrane budding	7.199	2.296	0.034	0.042	0.107
32	regulation of Ras protein signal transduction	7.520	2.294	0.026	0.022	0.099
33	protein import	7.461	2.288	0.032	0.046	0.111
34	regulation of organelle assembly	6.741	2.275	0.008	0.026	0.099
35	internal protein amino acid acetylation	7.278	2.260	0.012	0.022	0.099
36	negative regulation of viral genome replication	7.849	2.252	0.040	0.030	0.099
37	mitochondrial fusion	7.055	2.245	0.002	0.002	0.099
38	protein targeting to mitochondrion	7.454	2.244	0.016	0.026	0.099
39	regulation of myeloid leukocyte differentiation	6.999	2.237	0.014	0.026	0.099
40	positive regulation of lymphocyte migration	8.102	2.219	0.028	0.018	0.099

Table 5: The 40 highest-scoring gene sets returned by GSEA in Alzheimer's

Rank	Gene Set	NES	p-value	FDR
1	transepithelial transport	-2.064	0.000	0.025
2	neuron cell-cell adhesion	-1.929	0.000	0.135
3	inorganic anion transport	-1.921	0.000	0.106
4	cardiac ventricle morphogenesis	-1.916	0.000	0.087
5	neuron migration	-1.915	0.000	0.071
6	regulation of smad protein import into nucleus	-1.893	0.002	0.078
7	positive regulation of glycogen metabolic process	-1.885	0.000	0.076
8	chondroitin sulfate proteoglycan metabolic process	-1.881	0.000	0.071
9	cardiac muscle tissue morphogenesis	-1.869	0.000	0.077
10	mesenchyme morphogenesis	-1.864	0.000	0.075
11	chloride transport	-1.861	0.000	0.071
12	chondroitin sulfate metabolic process	-1.853	0.000	0.075
13	cyclic nucleotide metabolic process	-1.851	0.002	0.071
14	endocardial cushion formation	-1.851	0.000	0.066
15	mucopolysaccharide metabolic process	-1.847	0.000	0.065
16	positive regulation of glucose metabolic process	-1.843	0.000	0.065
17	muscle organ morphogenesis	-1.837	0.000	0.066
18	muscle tissue morphogenesis	-1.837	0.000	0.062
19	regulation of cholesterol storage	-1.833	0.000	0.062
20	regulation of glycogen metabolic process	-1.833	0.000	0.059
21	sodium-independent organic anion transport	-1.829	0.000	0.059
22	glycosaminoglycan metabolic process	-1.827	0.000	0.059
23	regulation of synapse assembly	-1.822	0.004	0.060
24	aminoglycan metabolic process	-1.817	0.000	0.063
25	positive regulation of glycogen biosynthetic process	-1.814	0.000	0.061
26	pulmonary valve development	-1.810	0.000	0.063
27	pulmonary valve morphogenesis	-1.810	0.000	0.061
28	chloride transmembrane transport	-1.808	0.000	0.060
29	cardioblast differentiation	-1.807	0.002	0.058
30	negative regulation of peptide hormone secretion	-1.799	0.000	0.061
31	inorganic anion transmembrane transport	-1.799	0.000	0.060
32	positive regulation of cardiac muscle tissue development	-1.797	0.000	0.059
33	glycosaminoglycan biosynthetic process	-1.795	0.000	0.059
34	positive regulation of lipid transport	-1.793	0.000	0.060
35	ventricular cardiac muscle tissue development	-1.785	0.000	0.065
36	blood vessel endothelial cell differentiation	-1.785	0.000	0.063
37	camp metabolic process	-1.784	0.006	0.062
38	excretion	-1.784	0.002	0.060
39	polyol transport	-1.783	0.000	0.060
40	aminoglycan biosynthetic process	-1.781	0.000	0.060

Table 6: The 40 highest-scoring gene sets returned by maSigPro+GSEA preranked in Alzheimer's

Rank	Gene Set	NES	p-value	FDR
1	equilibrioception	1.510	0.000	0.686
2	aortic valve morphogenesis	1.478	0.000	1.000
3	aortic valve development	1.469	0.001	0.976
4	alkaloid metabolic process	1.429	0.012	1.000
5	intracellular transport of viral protein in host cell	1.427	0.006	1.000
6	intracellular protein transport in other organism involved in symbiotic interaction	1.424	0.008	1.000
7	heterochromatin organization	1.421	0.001	1.000
8	symbiont intracellular protein transport in host	1.420	0.002	1.000
9	positive regulation of organic acid transport	1.419	0.005	1.000
10	atp synthesis coupled proton transport	1.418	0.000	1.000
11	energy coupled proton transport, down electrochemical gradient	1.417	0.000	1.000
12	mitochondrial atp synthesis coupled proton transport	1.415	0.000	0.986
13	wound healing, spreading of epidermal cells	1.409	0.008	1.000
14	regulation of axon extension involved in axon guidance	1.408	0.006	0.993
15	electron transport chain	1.407	0.000	0.951
16	oxygen homeostasis	1.406	0.007	0.911
17	positive regulation of icosanoid secretion	1.404	0.011	0.877
18	positive regulation of axon guidance	1.402	0.008	0.877
19	respiratory electron transport chain	1.402	0.000	0.833
20	camp-mediated signaling	1.401	0.000	0.793
21	spermatid nucleus differentiation	1.400	0.005	0.773
22	positive regulation of t cell differentiation in thymus	1.399	0.005	0.752
23	sperm chromatin condensation	1.398	0.012	0.732
24	negative regulation of oxidative stress-induced neuron intrinsic apoptotic signaling pathway	1.398	0.009	0.704
25	myelin assembly	1.397	0.009	0.694
26	positive regulation of transcription from rna polymerase ii promoter involved in heart development	1.395	0.015	0.696
27	cardiac chamber formation	1.395	0.010	0.672
28	heterochromatin assembly	1.394	0.005	0.658
29	positive regulation of axon extension involved in axon guidance	1.394	0.007	0.639
30	negative regulation of jun kinase activity	1.392	0.006	0.634
31	positive regulation of fatty acid transport	1.391	0.006	0.627
32	positive regulation of fibroblast apoptotic process	1.389	0.011	0.632
33	positive regulation of thymocyte aggregation	1.384	0.006	0.673
34	nls-bearing protein import into nucleus	1.383	0.000	0.664
35	camera-type eye photoreceptor cell differentiation	1.379	0.013	0.702
36	negative regulation of hydrogen peroxide-mediated programmed cell death	1.379	0.017	0.684
37	tissue regeneration	1.377	0.007	0.685
38	riboflavin metabolic process	1.366	0.015	0.806
39	translational elongation	1.366	0.000	0.786
40	regulation of rna export from nucleus	1.366	0.006	0.774

Huntington's Disease

Table 7: The 40 highest-scoring gene sets ranked by TEMPO score for the Huntington's Disease data set.

Rank	Gene Set	Control MSE	Score	MSE p-value	Score p-value	Score BH
1	negative regulation of DNA recombination	55.689	3.830	0.002	0.002	0.050
2	telomere maintenance via recombination	55.109	3.215	0.002	0.002	0.050
3	pos. regulation of sodium ion transmembrane transport	65.303	2.982	0.004	0.004	0.050
4	phototransduction, visible light	52.178	2.957	0.006	0.006	0.058
5	phototransduction	55.145	2.835	0.002	0.002	0.050
6	regulation of anion transport	88.395	2.813	0.040	0.004	0.050
7	regulation of EGFR signaling pathway	65.323	2.739	0.012	0.004	0.050
8	negative regulation of transcription from RNA polymerase II promoter in response to stress	66.673	2.660	0.004	0.002	0.050
9	regulation of ERBB signaling pathway	70.726	2.557	0.012	0.008	0.063
10	detection of visible light	65.855	2.519	0.008	0.008	0.063
11	tumor necrosis factor-mediated signaling pathway	60.396	2.486	0.006	0.004	0.050
12	ammonium ion metabolic process	54.216	2.473	0.008	0.008	0.063
13	detection of light stimulus	69.491	2.427	0.012	0.012	0.069
14	negative regulation of cation channel activity	63.369	2.421	0.004	0.008	0.063
15	intestinal absorption	60.407	2.339	0.002	0.004	0.050
16	negative regulation of protein acetylation	80.507	2.330	0.014	0.004	0.050
17	reg.n of sodium ion transmembrane transporter activity	84.293	2.327	0.020	0.008	0.063
18	regulation of peptidyl-lysine acetylation	88.258	2.310	0.050	0.008	0.063
19	regulation of cholesterol metabolic process	72.227	2.304	0.006	0.002	0.050
20	mitotic recombination	80.405	2.302	0.026	0.020	0.073
21	digestion	60.423	2.248	0.002	0.006	0.058
22	cellular response to biotic stimulus	72.764	2.242	0.026	0.012	0.069
23	negative reg. of protein exit from endoplasmic reticulum	65.601	2.223	0.004	0.004	0.050
24	neg. reg. of retrograde protein transport, ER to cytosol	65.601	2.223	0.006	0.004	0.050
25	negative regulation of peptidyl-lysine acetylation	82.606	2.204	0.012	0.004	0.050
26	cellular response to molecule of bacterial origin	84.347	2.192	0.048	0.018	0.073
27	regulation of leukocyte degranulation	61.048	2.186	0.006	0.020	0.073
28	bile acid and bile salt transport	62.671	2.176	0.004	0.004	0.050
29	organophosphate catabolic process	79.399	2.174	0.040	0.030	0.080
30	reg. of transcription from RNA polymerase I promoter	87.380	2.171	0.022	0.012	0.069
31	negative regulation of ERAD pathway	73.598	2.166	0.004	0.004	0.050
32	digestive system process	68.070	2.154	0.004	0.020	0.073
33	peroxisomal membrane transport	66.423	2.138	0.004	0.006	0.058
34	protein import into peroxisome membrane	66.423	2.138	0.002	0.004	0.050
35	cell communication involved in cardiac conduction	76.003	2.109	0.012	0.012	0.069
36	regulation of nitric oxide biosynthetic process	81.990	2.104	0.034	0.018	0.073
37	positive reg. of ion transmembrane transporter activity	70.092	2.096	0.012	0.026	0.079
38	CDP-choline pathway	72.253	2.085	0.006	0.002	0.050
39	intracellular protein transmembrane import	70.021	2.075	0.004	0.022	0.077
40	positive regulation of transporter activity	74.556	2.074	0.012	0.022	0.077

Table 8: The 40 highest-scoring gene sets ranked by TEMPO score for the Huntington’s Disease data set, with symptomatic patients only.

Rank	Gene Set	Control MSE	Score	MSE p-value	Score p-value	Score BH
1	negative regulation of DNA recombination	55.689	4.273	0.002	0.002	0.043
2	positive regulation of sodium ion transmembrane transport	65.303	3.452	0.002	0.002	0.043
3	telomere maintenance via recombination	55.109	3.422	0.002	0.004	0.047
4	phototransduction, visible light	52.178	3.375	0.002	0.002	0.043
5	phototransduction	55.145	3.215	0.002	0.002	0.043
6	negative regulation of transcription from RNA polymerase II promoter in response to stress	66.673	3.070	0.004	0.002	0.043
7	regulation of EGFR signaling pathway	65.323	3.039	0.012	0.002	0.043
8	tumor necrosis factor-mediated signaling pathway	60.396	2.894	0.012	0.002	0.043
9	detection of visible light	65.855	2.886	0.014	0.006	0.059
10	regulation of ERBB signaling pathway	70.726	2.843	0.008	0.004	0.047
11	ammonium ion metabolic process	54.216	2.782	0.002	0.004	0.047
12	detection of light stimulus	69.491	2.767	0.014	0.012	0.073
13	regulation of sodium ion transmembrane transporter activity	84.293	2.719	0.030	0.004	0.047
14	intestinal absorption	60.407	2.665	0.002	0.004	0.047
15	negative regulation of cation channel activity	63.369	2.638	0.002	0.002	0.043
16	digestion	60.423	2.595	0.004	0.012	0.073
17	negative regulation of protein acetylation	80.507	2.552	0.016	0.006	0.059
18	regulation of peptidyl-lysine acetylation	88.258	2.538	0.048	0.010	0.070
19	bile acid and bile salt transport	62.671	2.528	0.002	0.006	0.059
20	regulation of cholesterol metabolic process	72.227	2.517	0.004	0.002	0.043
21	digestive system process	68.070	2.504	0.006	0.008	0.064
22	reg. of transcription from RNAP II promoter in response to stress	86.002	2.465	0.036	0.008	0.064
23	cellular response to biotic stimulus	72.764	2.453	0.008	0.008	0.064
24	mitotic recombination	80.405	2.447	0.016	0.022	0.077
25	negative regulation of peptidyl-lysine acetylation	82.606	2.437	0.012	0.008	0.064
26	positive regulation of transporter activity	74.556	2.417	0.014	0.014	0.074
27	organophosphate catabolic process	79.399	2.408	0.038	0.022	0.077
28	positive regulation of ion transmembrane transporter activity	70.092	2.405	0.006	0.018	0.077
29	regulation of transcription from RNA polymerase I promoter	87.380	2.403	0.026	0.014	0.074
30	locomotor rhythm	64.605	2.382	0.002	0.002	0.043
31	peroxisomal membrane transport	66.423	2.381	0.004	0.004	0.047
32	protein import into peroxisome membrane	66.423	2.381	0.002	0.002	0.043
33	neg. regulation of protein exit from endoplasmic reticulum	65.601	2.356	0.006	0.016	0.077
34	neg. regulation of retrograde protein transport, ER to cytosol	65.601	2.356	0.002	0.002	0.043
35	positive regulation of cation transmembrane transport	68.454	2.348	0.010	0.018	0.077
36	synapse organization	91.797	2.342	0.048	0.018	0.077
37	cell communication involved in cardiac conduction	76.003	2.337	0.012	0.014	0.074
38	blood coagulation, intrinsic pathway	77.824	2.324	0.014	0.004	0.047
39	cellular aldehyde metabolic process	79.671	2.285	0.024	0.020	0.077
40	negative regulation of ERAD pathway	73.598	2.279	0.010	0.012	0.073

Table 9: The 40 highest-scoring gene sets ranked by TEMPO score for the Huntington’s Disease data set, with pre-symptomatic patients only.

Rank	Gene Set	Control MSE	Score	MSE p-value	Score p-value	Score BH
1	negative regulation of DNA recombination	55.689	2.778	0.002	0.002	0.051
2	telomere maintenance via recombination	55.109	2.725	0.002	0.002	0.051
3	regulation of anion transport	88.395	2.418	0.042	0.006	0.068
4	neg. reg. of reactive oxygen species biosynthetic process	84.943	2.104	0.004	0.002	0.051
5	regulation of nitric oxide biosynthetic process	81.990	2.091	0.018	0.006	0.068
6	regulation of EGFR signaling pathway	65.323	2.027	0.010	0.004	0.068
7	intracellular protein transmembrane import	70.021	2.000	0.004	0.010	0.075
8	regulation of leukocyte degranulation	61.048	1.970	0.004	0.012	0.075
9	phototransduction, visible light	52.178	1.967	0.002	0.012	0.075
10	mitotic recombination	80.405	1.960	0.024	0.010	0.075
11	intracellular protein transmembrane transport	78.604	1.937	0.014	0.008	0.070
12	phototransduction	55.145	1.934	0.002	0.006	0.068
13	negative regulation of telomere capping	85.164	1.915	0.008	0.002	0.051
14	negative regulation of cation channel activity	63.369	1.906	0.010	0.010	0.075
15	neg. regulation of protein exit from endoplasmic reticulum	65.601	1.906	0.002	0.002	0.051
16	neg. regulation of retrograde protein transport, ER to cytosol	65.601	1.906	0.002	0.002	0.051
17	negative regulation of ERAD pathway	73.598	1.898	0.010	0.006	0.068
18	regulation of ERBB signaling pathway	70.726	1.880	0.020	0.014	0.075
19	positive regulation of sodium ion transmembrane transport	65.303	1.869	0.002	0.004	0.068
20	cellular glucose homeostasis	83.696	1.852	0.020	0.008	0.070
21	negative regulation of protein acetylation	80.507	1.803	0.012	0.006	0.068
22	regulation of cholesterol metabolic process	72.227	1.799	0.008	0.006	0.068
23	mitotic G2/M transition checkpoint	86.450	1.795	0.024	0.016	0.077
24	cellular response to biotic stimulus	72.764	1.741	0.022	0.020	0.079
25	ammonium ion metabolic process	54.216	1.739	0.004	0.014	0.075
26	DNA strand elongation	89.665	1.703	0.048	0.026	0.082
27	negative regulation of histone H3-K9 methylation	86.229	1.695	0.014	0.002	0.051
28	negative regulation of transcription from RNA polymerase II promoter in response to stress	66.673	1.687	0.002	0.008	0.070
29	phosphatidylethanolamine acyl-chain remodeling	84.213	1.665	0.016	0.008	0.070
30	protein heterooligomerization	92.321	1.662	0.034	0.018	0.077
31	negative regulation of peptidyl-lysine acetylation	82.606	1.650	0.012	0.020	0.079
32	detection of visible light	65.855	1.649	0.016	0.028	0.082
33	pos. regulation of potassium ion transmembrane transport	80.479	1.637	0.016	0.014	0.075
34	developmental process involved in reproduction	81.601	1.633	0.026	0.008	0.070
35	acute inflammatory response	75.302	1.633	0.010	0.024	0.082
36	CDP-choline pathway	72.253	1.632	0.002	0.002	0.051
37	negative regulation of nitric oxide biosynthetic process	96.945	1.629	0.048	0.002	0.051
38	negative regulation of nitric oxide metabolic process	96.945	1.629	0.044	0.002	0.051
39	protein targeting to peroxisome	68.215	1.629	0.012	0.024	0.082
40	peroxisomal transport	68.215	1.629	0.008	0.026	0.082

Table 10: The 40 highest-scoring gene sets returned by GSEA in Huntingtons

Rank	Gene Set	NES	p-value	FDR
1	regulation of symbiosis, encompassing mutualism through parasitism	-1.87	0.00	1.00
2	carnitine shuttle	-1.84	0.00	1.00
3	negative regulation of insulin receptor signaling pathway	-1.83	0.01	1.00
4	regulation of insulin receptor signaling pathway	-1.81	0.00	1.00
5	negative regulation of cellular response to insulin stimulus	-1.76	0.01	1.00
6	amino-acid betaine transport	-1.75	0.01	1.00
7	carnitine transport	-1.75	0.01	1.00
8	bone morphogenesis	-1.73	0.01	1.00
9	sarcomere organization	-1.73	0.01	1.00
10	endochondral ossification	-1.73	0.00	1.00
11	replacement ossification	-1.73	0.00	1.00
12	regulation of wound healing	-1.71	0.00	1.00
13	positive regulation of leukocyte chemotaxis	-1.71	0.02	1.00
14	actin-mediated cell contraction	-1.70	0.01	1.00
15	fatty acid transmembrane transport	-1.69	0.01	1.00
16	positive regulation of cell-substrate adhesion	-1.68	0.04	1.00
17	anterior/posterior axis specification	-1.68	0.01	1.00
18	regulation of blood pressure	-1.67	0.00	1.00
19	positive regulation of cell-matrix adhesion	-1.66	0.03	1.00
20	activation of mapkk activity	-1.65	0.02	1.00
21	positive regulation of endothelial cell proliferation	-1.65	0.01	1.00
22	phosphatidylglycerol biosynthetic process	-1.65	0.03	1.00
23	nucleoside bisphosphate metabolic process	-1.64	0.01	1.00
24	ribonucleoside bisphosphate metabolic process	-1.64	0.01	1.00
25	purine nucleoside bisphosphate metabolic process	-1.64	0.01	1.00
26	regulation of establishment or maintenance of cell polarity	-1.64	0.02	1.00
27	purine ribonucleoside bisphosphate metabolic process	-1.64	0.01	1.00
28	3'-phosphoadenosine 5'-phosphosulfate metabolic process	-1.64	0.01	1.00
29	copper ion transport	-1.64	0.03	1.00
30	bone development	-1.64	0.01	1.00
31	regulation of coagulation	-1.63	0.01	1.00
32	regulation of blood coagulation	-1.63	0.02	1.00
33	regulation of hemostasis	-1.63	0.02	1.00
34	n-acetylneuraminate metabolic process	-1.62	0.01	1.00
35	inflammatory response	-1.62	0.02	1.00
36	neutrophil activation	-1.62	0.04	1.00
37	endochondral bone morphogenesis	-1.61	0.02	1.00
38	regulated secretory pathway	-1.61	0.03	1.00
39	post-embryonic hemopoiesis	-1.61	0.00	1.00
40	trabecula formation	-1.61	0.02	1.00

Table 11: The 40 highest-scoring gene sets returned by maSigPro+GSEA preranked in Huntingtons

Rank	Gene Set	NES	p-value	FDR
1	steroid biosynthetic process	2.31	0.00	0.71
2	dna metabolic process	2.12	0.00	1.00
3	transcription-coupled nucleotide-excision repair	2.11	0.01	1.00
4	negative regulation of rna metabolic process	2.11	0.00	0.96
5	cellular response to reactive oxygen species	2.07	0.00	0.97
6	dna strand elongation involved in dna replication	2.05	0.00	1.00
7	negative regulation of nitrogen compound metabolic process	2.04	0.00	0.87
8	response to oxidative stress	2.04	0.00	0.77
9	cholesterol biosynthetic process	2.03	0.00	0.73
10	peroxisome fission	2.02	0.00	0.72
11	deoxyribose phosphate metabolic process	2.01	0.00	0.69
12	dna repair	2.01	0.00	0.65
13	cgmp metabolic process	2.01	0.00	0.61
14	sterol biosynthetic process	2.00	0.00	0.58
15	positive regulation of gtpase activity	2.00	0.01	0.54
16	regulation of dna-dependent transcription in response to stress	2.00	0.00	0.52
17	monocyte differentiation	1.98	0.00	0.56
18	regulation of histone h3-k4 methylation	1.98	0.00	0.53
19	negative regulation of transcription, dna-dependent	1.98	0.00	0.51
20	regulation of cytokine production involved in immune response	1.97	0.00	0.50
21	metabolic process	1.97	0.01	0.48
22	alcohol biosynthetic process	1.97	0.01	0.47
23	deoxyribonucleotide catabolic process	1.96	0.01	0.46
24	cholesterol metabolic process	1.96	0.01	0.45
25	regulation of nucleoside metabolic process	1.95	0.01	0.46
26	nucleotide-excision repair	1.94	0.01	0.47
27	negative regulation of immune effector process	1.94	0.01	0.45
28	regulation of transcription from rna polymerase ii promoter in response to stress	1.94	0.01	0.44
29	negative regulation of nucleobase-containing compound metabolic process	1.94	0.00	0.43
30	response to reactive oxygen species	1.93	0.01	0.43
31	deoxyribose phosphate catabolic process	1.93	0.00	0.42
32	protein k48-linked ubiquitination	1.93	0.01	0.41
33	regulation of ras protein signal transduction	1.93	0.01	0.40
34	sterol metabolic process	1.92	0.00	0.40
35	regulation of receptor activity	1.92	0.01	0.40
36	negative regulation of cellular macromolecule biosynthetic process	1.92	0.00	0.39
37	2'-deoxyribonucleotide metabolic process	1.92	0.01	0.38
38	regulation of production of molecular mediator of immune response	1.91	0.00	0.39
39	positive regulation of histone h3-k4 methylation	1.90	0.00	0.40
40	steroid metabolic process	1.90	0.01	0.39

COPD

Table 12: The 40 highest-scoring gene sets ranked by TEMPO score for the COPD data set

Rank	Gene Set	Control		MSE	Score	Score
		MSE	Score	p-value	p-value	BH
1	alanine transport	0.430	305.978	0.002	0.002	0.076
2	positive regulation of interferon-gamma secretion	0.145	272.491	0.002	0.004	0.093
3	positive regulation of phospholipid biosynthetic process	2.419	203.185	0.004	0.002	0.076
4	T-helper cell lineage commitment	5.208	194.402	0.004	0.004	0.093
5	T-helper 17 cell lineage commitment	5.208	194.402	0.004	0.006	0.093
6	transcytosis	0.759	192.616	0.002	0.002	0.076
7	vascular smooth muscle cell development	2.600	177.217	0.004	0.002	0.076
8	nephric duct development	1.756	175.896	0.002	0.002	0.076
9	mRNA transcription from RNA polymerase II promoter	1.468	137.089	0.002	0.002	0.076
10	opioid receptor signaling pathway	7.566	128.480	0.028	0.008	0.095
11	regulation of extracellular matrix organization	6.449	122.182	0.030	0.002	0.076
12	epithelial tube branching involved in lung morphogenesis	1.066	109.133	0.002	0.008	0.095
13	RNA surveillance	4.160	106.883	0.014	0.006	0.093
14	purine nucleobase transport	3.093	98.951	0.004	0.010	0.095
15	monocyte chemotaxis	4.356	96.412	0.008	0.006	0.093
16	cell proliferation in forebrain	6.000	95.990	0.014	0.002	0.076
17	regulation of cell-cell adhesion mediated by cadherin	5.771	93.285	0.012	0.006	0.093
18	negative regulation of extracellular matrix organization	4.058	88.945	0.006	0.008	0.095
19	regulation of protein complex stability	5.739	87.307	0.010	0.008	0.095
20	regulation of oligodendrocyte differentiation	4.785	86.896	0.010	0.004	0.093
21	adrenal gland development	8.023	85.309	0.048	0.006	0.093
22	glutamine family amino acid metabolic process	3.983	83.608	0.004	0.002	0.076
23	sulfide oxidation	4.087	83.123	0.010	0.010	0.095
24	sulfide oxidation, using sulfide:quinone oxidoreductase	4.087	83.123	0.004	0.012	0.105
25	regulation of cardiac muscle cell membrane potential	5.520	81.049	0.012	0.010	0.095
26	pyrimidine-containing compound transmembrane transport	2.909	79.713	0.004	0.006	0.093
27	neg. reg. of mitochondrial outer membrane permeabilization involved in apoptotic signaling pathway	4.213	79.237	0.008	0.010	0.095
28	negative regulation of activin receptor signaling pathway	6.134	78.457	0.018	0.014	0.105
29	regulation of microvillus organization	5.683	77.186	0.014	0.014	0.105
30	regulation of microvillus assembly	5.683	77.186	0.014	0.012	0.105
31	glutamine transport	3.735	77.005	0.014	0.026	0.106
32	neuroblast proliferation	7.298	75.503	0.038	0.006	0.093
33	sequestering of metal ion	5.122	73.510	0.026	0.026	0.106
34	negative regulation of protein sumoylation	3.962	73.403	0.004	0.020	0.106
35	nucleobase-containing small molecule interconversion	1.875	71.447	0.002	0.006	0.093
36	bundle of His cell-Purkinje myocyte adhesion involved in cell communication	10.007	68.231	0.040	0.016	0.106
37	positive regulation of Rho protein signal transduction	5.440	62.829	0.018	0.010	0.095
38	response to acid chemical	2.122	58.860	0.002	0.002	0.076
39	reg. of cardiac muscle contraction by calcium ion signaling	3.821	58.222	0.004	0.004	0.093
40	bicellular tight junction assembly	7.333	57.457	0.044	0.006	0.093

Table 13: The 40 highest-scoring upregulated gene sets returned by GSEA in COPD.

Rank	Gene Set	NES	p-value	FDR
1	peptidyl-proline hydroxylation	-1.917	0.002	1.000
2	positive regulation of nuclease activity	-1.838	0.002	1.000
3	regulation of isotype switching	-1.834	0.000	1.000
4	peptidyl-serine autophosphorylation	-1.827	0.000	1.000
5	oocyte differentiation	-1.812	0.004	1.000
6	sulfide oxidation	-1.761	0.002	1.000
7	sulfide oxidation, using sulfide:quinone oxidoreductase	-1.761	0.002	1.000
8	regulation of mitophagy	-1.750	0.000	1.000
9	macromolecule depalmitoylation	-1.746	0.002	1.000
10	iron-sulfur cluster assembly	-1.745	0.000	1.000
11	metallo-sulfur cluster assembly	-1.745	0.000	1.000
12	protein hydroxylation	-1.732	0.000	1.000
13	regulation of torc1 signaling	-1.729	0.006	1.000
14	response to misfolded protein	-1.716	0.004	1.000
15	negative regulation of macroautophagy	-1.711	0.002	1.000
16	mitochondrial atp synthesis coupled proton transport	-1.704	0.046	1.000
17	positive regulation of ruffle assembly	-1.697	0.018	1.000
18	oocyte development	-1.696	0.010	1.000
19	positive regulation of isotype switching	-1.695	0.008	1.000
20	activation of signaling protein activity involved in unfolded protein response	-1.686	0.011	1.000
21	nucleosome disassembly	-1.685	0.002	1.000
22	chromatin disassembly	-1.685	0.002	1.000
23	protein-dna complex disassembly	-1.685	0.002	1.000
24	positive regulation of erad pathway	-1.680	0.000	1.000
25	energy coupled proton transport, down electrochemical gradient	-1.679	0.048	1.000
26	atp synthesis coupled proton transport	-1.679	0.048	1.000
27	base-excision repair, ap site formation	-1.677	0.011	1.000
28	peptidyl-diphthamide metabolic process	-1.676	0.011	1.000
29	peptidyl-diphthamide biosynthetic process from peptidyl-histidine	-1.676	0.011	1.000
30	positive regulation of neurotransmitter secretion	-1.672	0.004	1.000
31	mitochondrial respiratory chain complex assembly	-1.670	0.037	1.000
32	positive regulation of anoikis	-1.669	0.006	1.000
33	establishment of protein localization to mitochondrion	-1.668	0.034	1.000
34	regulation of autophagosome maturation	-1.663	0.004	1.000
35	protein localization to mitochondrion	-1.661	0.026	1.000
36	regulation of immunoglobulin production	-1.657	0.012	1.000
37	histone h4-k12 acetylation	-1.648	0.002	1.000
38	calcineurin-nfat signaling cascade	-1.643	0.011	1.000
39	regulation of nuclease activity	-1.643	0.006	1.000
40	heme biosynthetic process	-1.639	0.015	1.000

Table 14: The 40 highest-scoring upregulated gene sets returned by maSigPro+GSEA preranked in COPD.

Rank	Gene Set	NES	p-value	FDR
1	ectodermal placode development	2.104	0.000	0.091
2	ectodermal placode morphogenesis	2.102	0.000	0.047
3	ectodermal placode formation	2.077	0.000	0.049
4	response to amino acid	1.988	0.000	0.117
5	peptidyl-methionine modification	1.966	0.001	0.124
6	positive regulation of er-associated ubiquitin-dependent protein catabolic process	1.934	0.000	0.147
7	ubiquinone biosynthetic process	1.933	0.000	0.130
8	base-excision repair, ap site formation	1.930	0.000	0.117
9	positive regulation of intracellular estrogen receptor signaling pathway	1.922	0.001	0.112
10	cellular response to amino acid stimulus	1.917	0.000	0.109
11	spliceosomal tri-snrnp complex assembly	1.914	0.000	0.103
12	protein monoubiquitination	1.906	0.000	0.105
13	histone ubiquitination	1.903	0.000	0.099
14	quinone biosynthetic process	1.891	0.000	0.106
15	ubiquinone metabolic process	1.887	0.000	0.104
16	short-chain fatty acid catabolic process	1.883	0.001	0.101
17	jun phosphorylation	1.881	0.002	0.099
18	acetyl-coa biosynthetic process from pyruvate	1.871	0.000	0.105
19	regulation of establishment of protein localization to telomere	1.869	0.003	0.102
20	trna threonylcarbamoyladenine metabolic process	1.864	0.001	0.101
21	mitochondrial rna modification	1.862	0.002	0.100
22	intermediate filament organization	1.861	0.001	0.096
23	regulation of establishment of protein localization to chromosome	1.852	0.001	0.103
24	histone monoubiquitination	1.849	0.000	0.102
25	smad protein complex assembly	1.843	0.000	0.104
26	regulation of autophagosome maturation	1.840	0.001	0.104
27	spliceosomal snrnp assembly	1.839	0.000	0.101
28	mitochondrial rna processing	1.839	0.000	0.098
29	mitochondrial trna modification	1.836	0.002	0.097
30	depyrimidination	1.832	0.000	0.098
31	positive regulation of protein localization to cajal body	1.819	0.000	0.108
32	regulation of mrna 3'-end processing	1.816	0.000	0.108
33	regulation of low-density lipoprotein particle clearance	1.812	0.003	0.109
34	regulation of protein localization to cajal body	1.808	0.001	0.111
35	pyrimidine-containing compound transmembrane transport	1.797	0.001	0.121
36	mitochondrion morphogenesis	1.797	0.000	0.118
37	nucleobase-containing small molecule catabolic process	1.797	0.002	0.115
38	primary mirna processing	1.795	0.001	0.114
39	membrane depolarization during sa node cell action potential	1.792	0.009	0.115
40	viral release from host cell	1.791	0.000	0.114