Supporting Information for
DNA language models are powerful predictors of genome-wide variant effects

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Supplementary Tables

Table S1: Genome assemblies used for training

Assembly Accession	Assembly Name	Organism Name
GCF_000001735.4	TAIR10.1	*Arabidopsis thaliana*
GCF_000309985.2	CAAS_Brap_v3.01	*Brassica rapa*
GCF_000633955.1	Cs	*Camelina sativa*
GCF_000375325.1	Caprub1_0	*Capsella rubella*
GCF_000150535.2	Papaya1.0	*Carica papaya*
GCF_000478725.1	Eutsalg1_0	*Eutrema salsugineum*
GCF_000801105.1	Rs1.0	*Raphanus sativus*
GCF_000463585.1	ASM46358v1	*Tarenaya hassleriana*

Table S2: Test perplexity. Perplexity, defined as the exponentiation of the cross-entropy loss, is equivalent to 1 over the probability given to the correct nucleotide. *Arabidopsis thaliana* chromosomes 4 and 5 were used for validation and testing, respectively. Note that reducing the repeat weight leads to improved test perplexity in non-repetitive regions, which are often of greater interest. Compared to full down-weighting, moderate down-weighting results in a similar improvement in perplexity for non-repetitive regions without sacrificing genome-wide perplexity as much.

Model	Chromosome-wide	Non-repeat regions
Repeat weight 1	2.88	2.99
Repeat weight 0.1	2.90	2.92
Repeat weight 0	3.03	2.92

Table S3: Training hyperparameters

Window size (L)	512
Repeat weight	0.1
Embedding dimension (D)	512
Convolutional blocks	25
Convolutional kernel size	9
Convolutional dilation schedule	$1, 2, 4, 8, 16, 32, 1, 2, 4, 8, 16, 32, \ldots$
Optimizer	AdamW
Weight decay	0.01
Batch size	2048
Learning rate	10^{-3} for 120 K steps + decaying (cosine) for 30 K steps
Learning rate warmup	1 K steps

Supplementary Figures

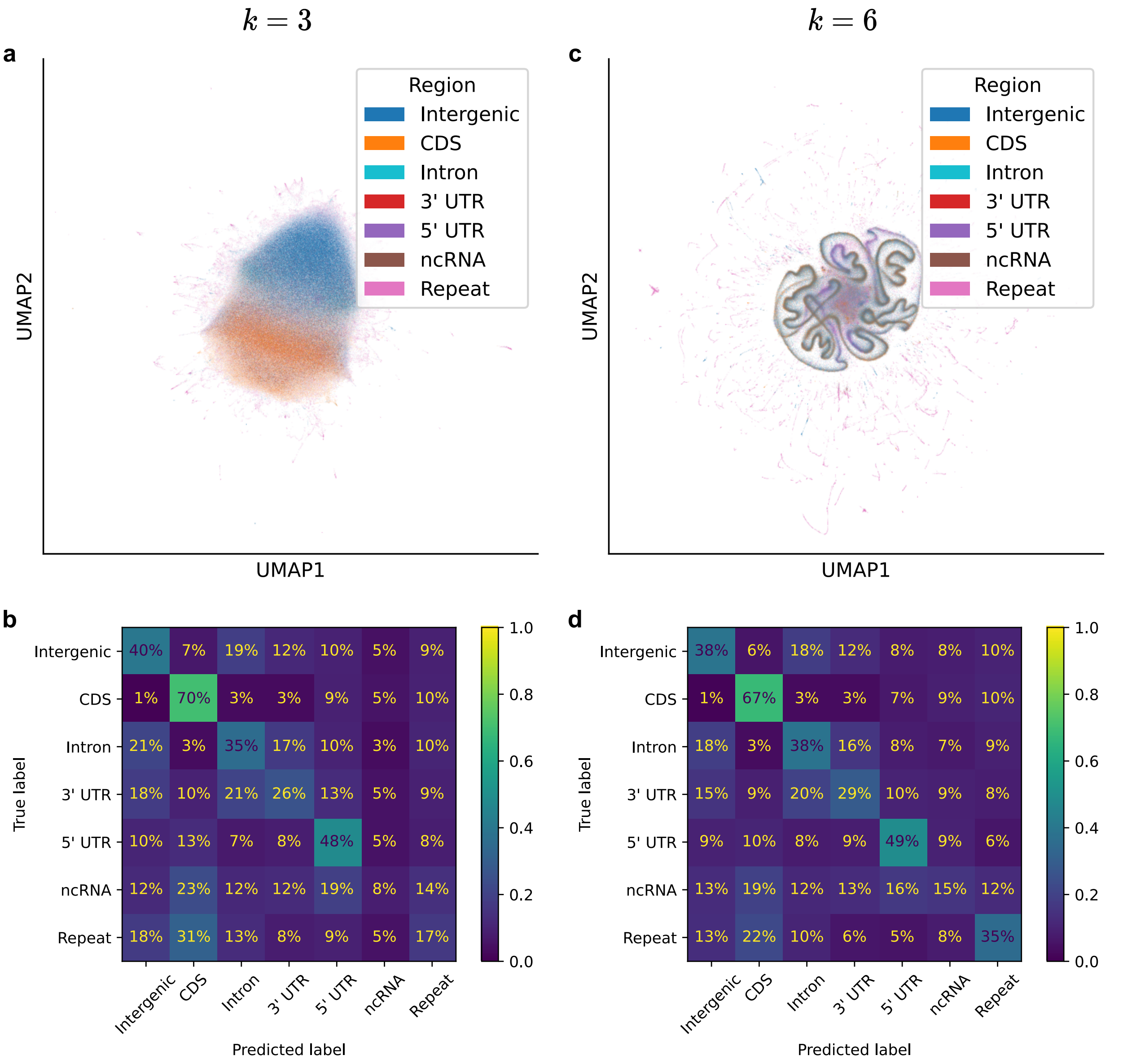

Figure S1: UMAP visualization of k-mer spectrum of different windows, as in Fig. 2, annotated with gene region. (a,b) $k = 3$. (c,d) $k = 6$.

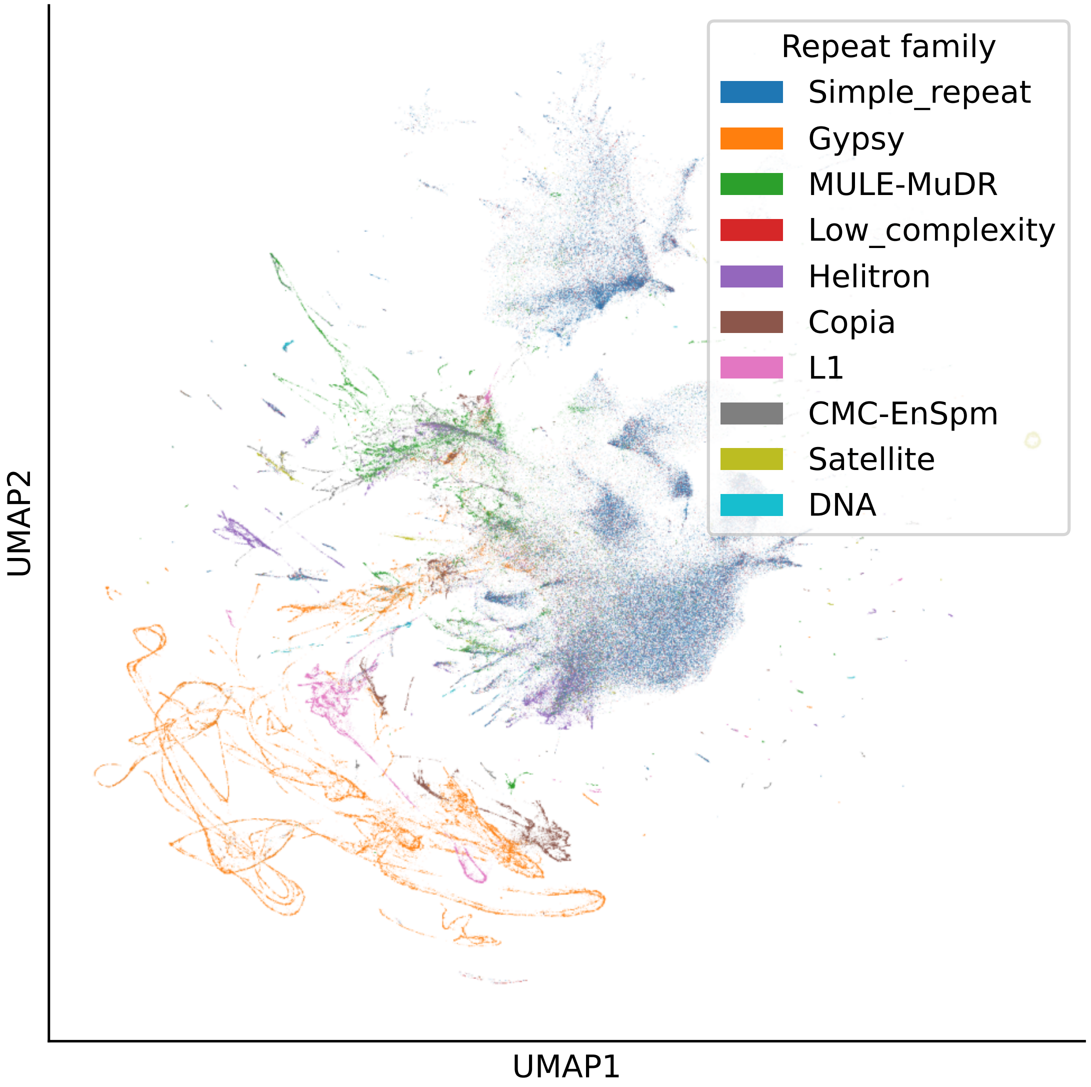

Figure S2: UMAP visualization of GPN embeddings, as in Fig. 2, annotated by repeat family.

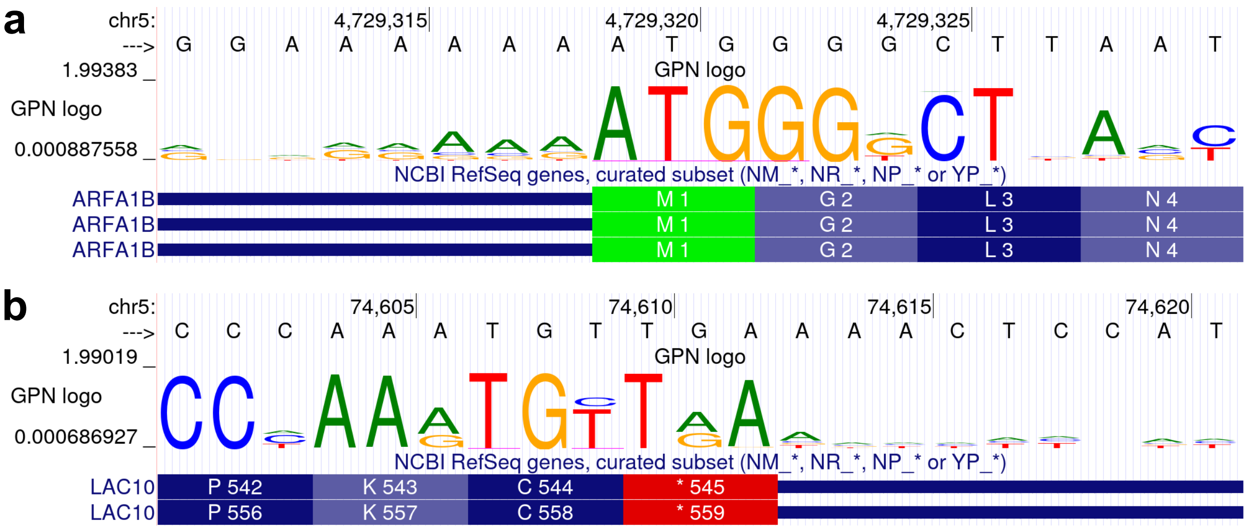

Figure S3: Additional GPN sequence logos. (a) Start codon. (b) Stop codon.

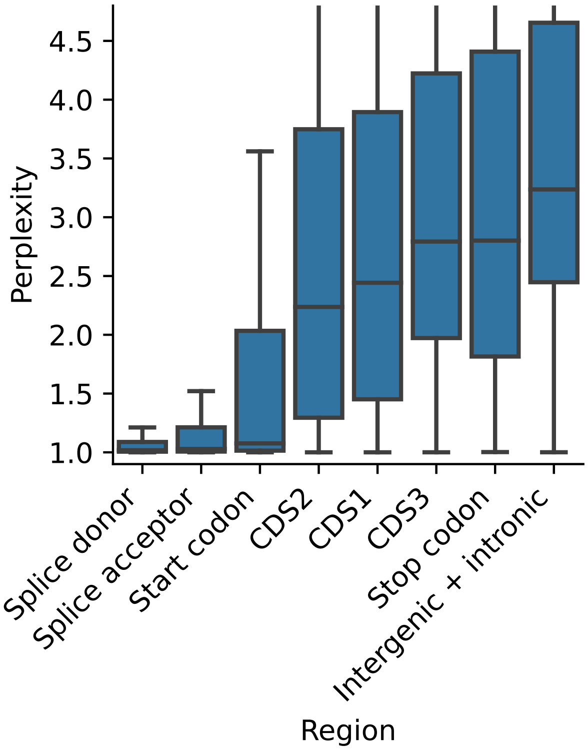

Figure S4: Perplexity on select positions from the 1 Mb region `Chr5:3,500,000-4,500,000` (test chromosome). CDS1-3: frame within the coding sequence.

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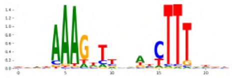
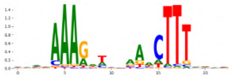
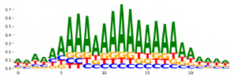
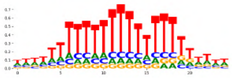
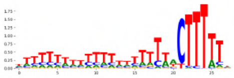
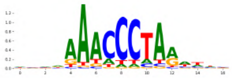
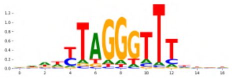
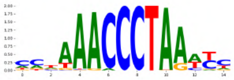
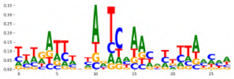
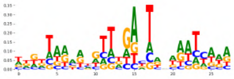
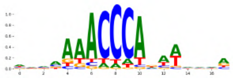
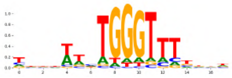
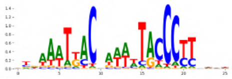
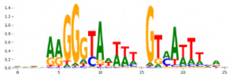
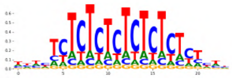
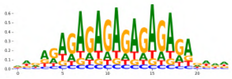
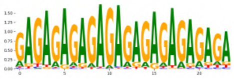
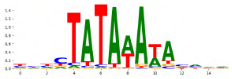
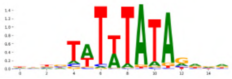
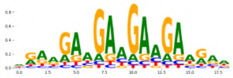
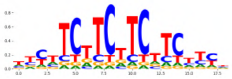
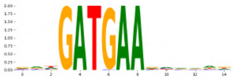
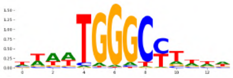
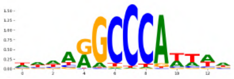
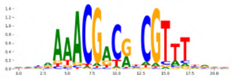
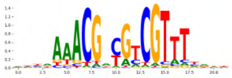
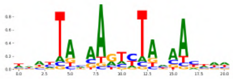
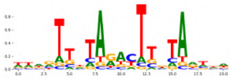
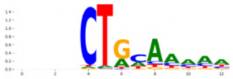
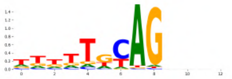
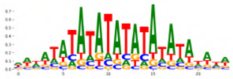
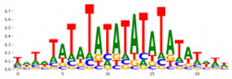
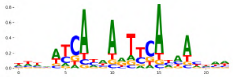
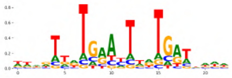
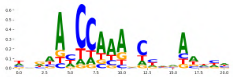
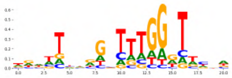
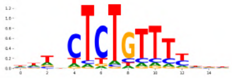
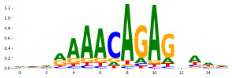
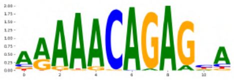
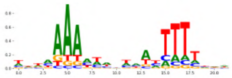
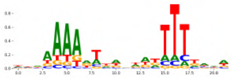
pos_patterns.pattern_0	5028					
pos_patterns.pattern_1	4509			AT4G38000	0.0	
pos_patterns.pattern_2	3386			AT1G72740	0.000117	
pos_patterns.pattern_3	1658					
pos_patterns.pattern_4	1611					
pos_patterns.pattern_5	1556					
pos_patterns.pattern_6	1490			AT2G01930	0.0	
pos_patterns.pattern_7	1424					
pos_patterns.pattern_8	1391			AT5G18090	0.044918	
pos_patterns.pattern_9	1385					
pos_patterns.pattern_10	1057					
pos_patterns.pattern_11	1052					
pos_patterns.pattern_12	928					
pos_patterns.pattern_13	921					
pos_patterns.pattern_14	844					
pos_patterns.pattern_15	837					
pos_patterns.pattern_16	836			AT3G48430	0.000584	
pos_patterns.pattern_17	828					

Figure S5: Promoter motifs predicted by GPN and matching motifs in PlantTFDB.

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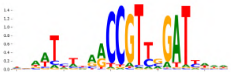
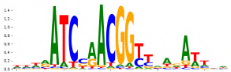
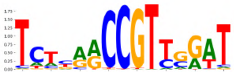
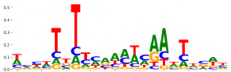
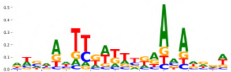
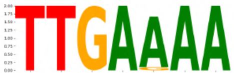
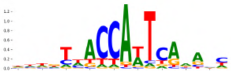
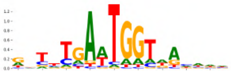
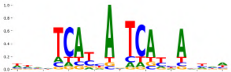
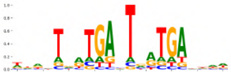
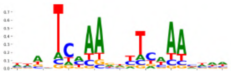
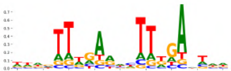
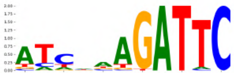
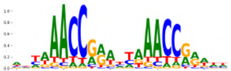
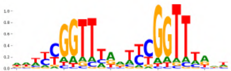
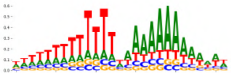
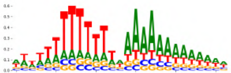
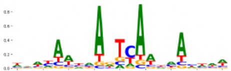
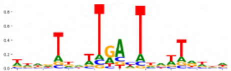
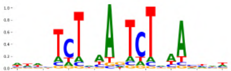
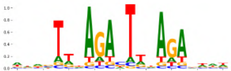
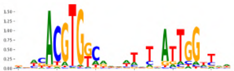
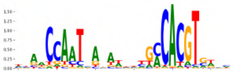
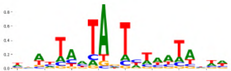
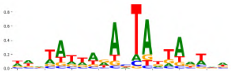
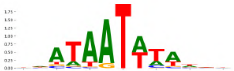
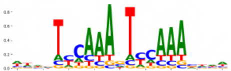
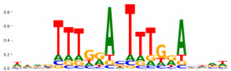
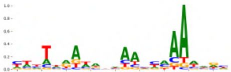
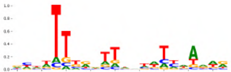
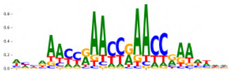
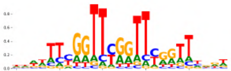
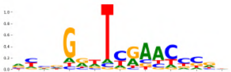
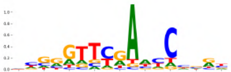
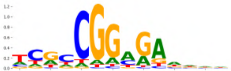
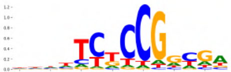
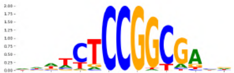
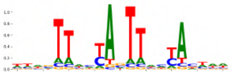
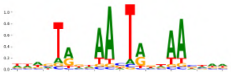
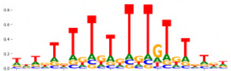
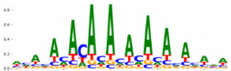
pos_patterns.pattern_18	780			AT4G24470	0.0	
pos_patterns.pattern_19	775			AT2G41835	0.016114	
pos_patterns.pattern_20	756					
pos_patterns.pattern_21	743					
pos_patterns.pattern_22	729			AT1G49560	0.029145	
pos_patterns.pattern_23	708					
pos_patterns.pattern_24	702					
pos_patterns.pattern_25	696					
pos_patterns.pattern_26	688					
pos_patterns.pattern_27	674					
pos_patterns.pattern_28	643			AT2G36610	0.026016	
pos_patterns.pattern_29	640					
pos_patterns.pattern_30	631					
pos_patterns.pattern_31	631					
pos_patterns.pattern_32	630					
pos_patterns.pattern_33	604			AT3G58630	0.029229	
pos_patterns.pattern_34	595					
pos_patterns.pattern_35	595					

Figure S5: (Continued)

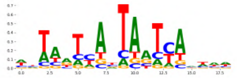
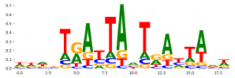
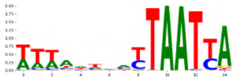
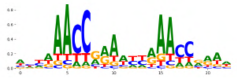
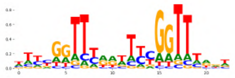
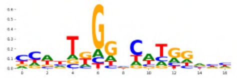
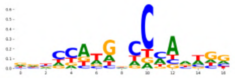
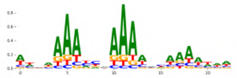
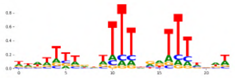
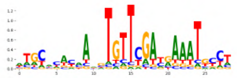
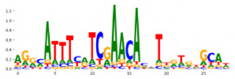
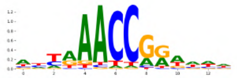
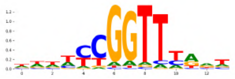
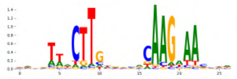
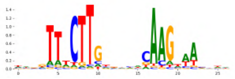
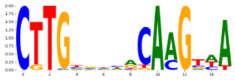
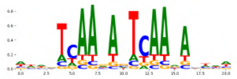
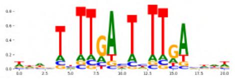
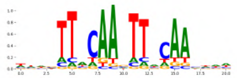
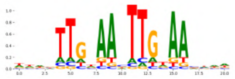
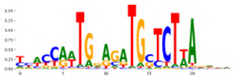
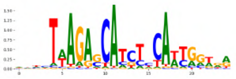
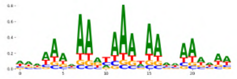
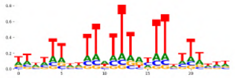
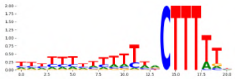
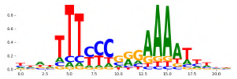
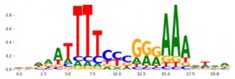
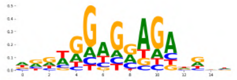
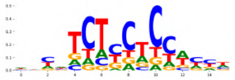
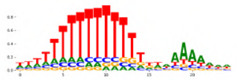
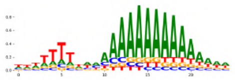
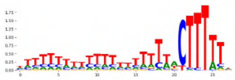
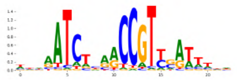
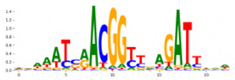
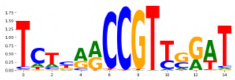
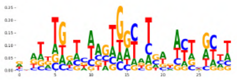
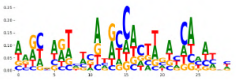
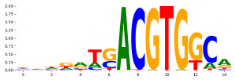
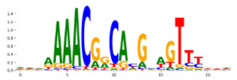
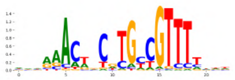
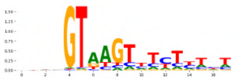
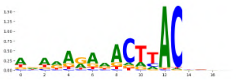
pattern	num_seqlets	modisco_cwm_fwd	modisco_cwm_rev	match0	qval0	match0_logo
pos_patterns.pattern_36	590			AT3G28920	0.000035	
pos_patterns.pattern_37	580					
pos_patterns.pattern_38	566					
pos_patterns.pattern_39	558					
pos_patterns.pattern_40	544					
pos_patterns.pattern_41	523					
pos_patterns.pattern_42	512			AT3G10480	0.000001	
pos_patterns.pattern_43	511					
pos_patterns.pattern_44	508					
pos_patterns.pattern_45	506					
pos_patterns.pattern_46	492			AT2G28810	0.000187	
pos_patterns.pattern_47	480					
pos_patterns.pattern_48	480					
pos_patterns.pattern_49	477			AT4G38000	0.002972	
pos_patterns.pattern_50	454			AT4G24470	0.000033	
pos_patterns.pattern_51	451			AT3G10800	0.025896	
pos_patterns.pattern_52	433					
pos_patterns.pattern_53	426					

Figure S5: (Continued)

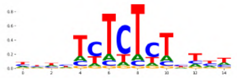
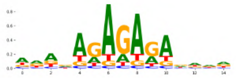
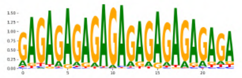
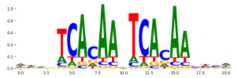
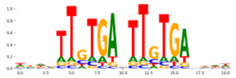
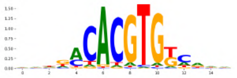
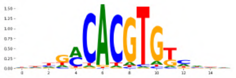
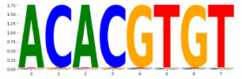
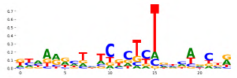
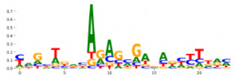
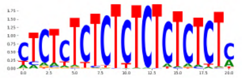
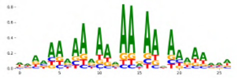
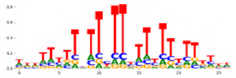
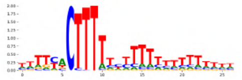
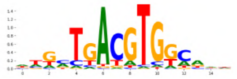
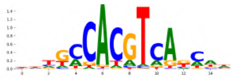
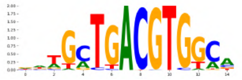
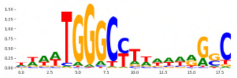
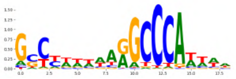
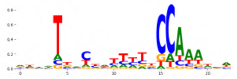
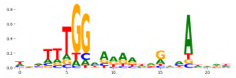
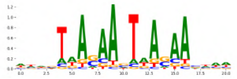
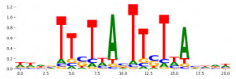
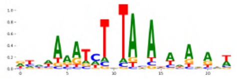
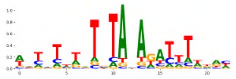
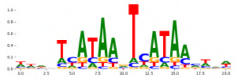
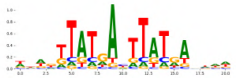
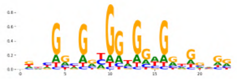
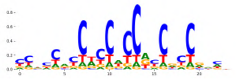
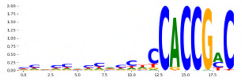
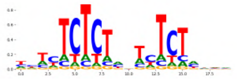
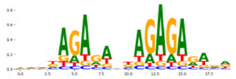
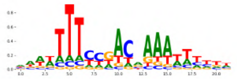
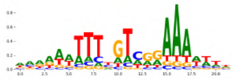
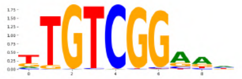
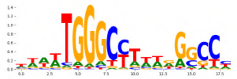
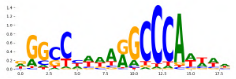
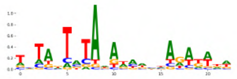
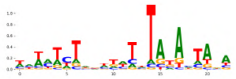
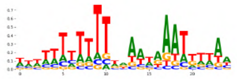
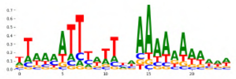
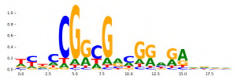
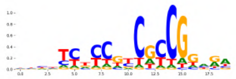
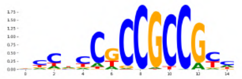
pattern	num_seqlets	modisco_cwm_fwd	modisco_cwm_rev	match0	qval0	match0_logo
pos_patterns.pattern_54	407			AT2G01930	0.002768	
pos_patterns.pattern_55	404					
pos_patterns.pattern_56	387			AT4G34000	0.000455	
pos_patterns.pattern_57	357			AT5G42520	0.037446	
pos_patterns.pattern_58	354			AT1G69570	0.0	
pos_patterns.pattern_59	352			AT3G62420	0.000053	
pos_patterns.pattern_60	322					
pos_patterns.pattern_61	319					
pos_patterns.pattern_62	314					
pos_patterns.pattern_63	311					
pos_patterns.pattern_64	309					
pos_patterns.pattern_65	302			AT1G21910	0.0	
pos_patterns.pattern_66	293					
pos_patterns.pattern_67	291			AT2G33860	0.005094	
pos_patterns.pattern_68	285					
pos_patterns.pattern_69	282					
pos_patterns.pattern_70	280					
pos_patterns.pattern_71	280			AT1G53170	0.001163	

Figure S5: (Continued)

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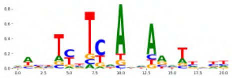
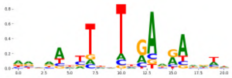
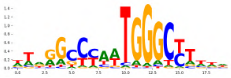
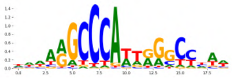
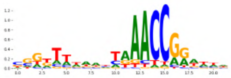
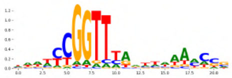
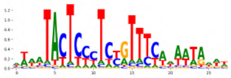
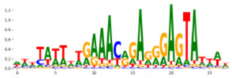
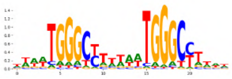
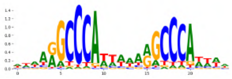
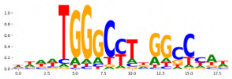
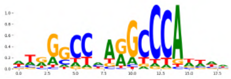
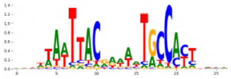
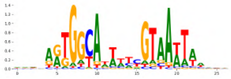
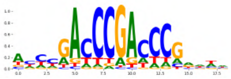
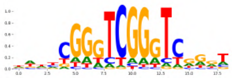
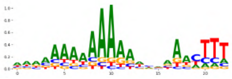
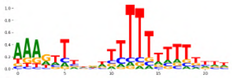
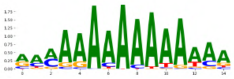
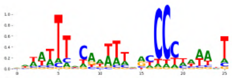
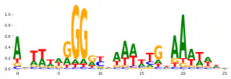
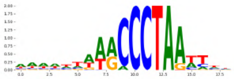
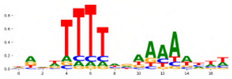
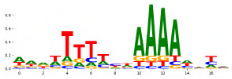
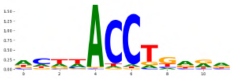
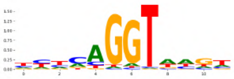
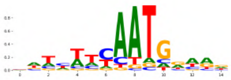
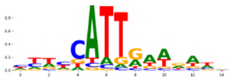
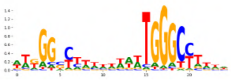
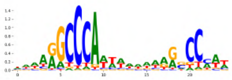
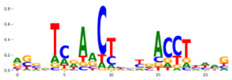
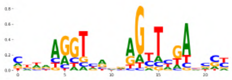
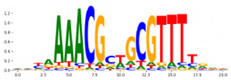
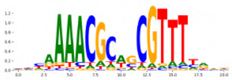
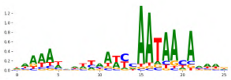
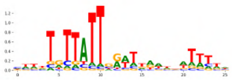
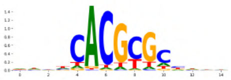
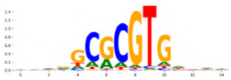
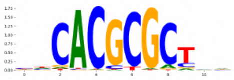
pos_patterns.pattern_72	275					
pos_patterns.pattern_73	265					
pos_patterns.pattern_74	263					
pos_patterns.pattern_75	262					
pos_patterns.pattern_76	259					
pos_patterns.pattern_77	253					
pos_patterns.pattern_78	249					
pos_patterns.pattern_79	248					
pos_patterns.pattern_80	242			AT1G49480	0.000331	
pos_patterns.pattern_81	228			AT5G67580	0.016528	
pos_patterns.pattern_82	225					
pos_patterns.pattern_83	221					
pos_patterns.pattern_84	215					
pos_patterns.pattern_85	209					
pos_patterns.pattern_86	206					
pos_patterns.pattern_87	199					
pos_patterns.pattern_88	198					
pos_patterns.pattern_89	194			AT3G22170	0.002435	

Figure S5: (Continued)

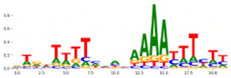
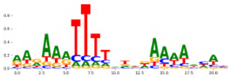
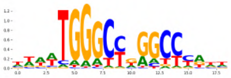
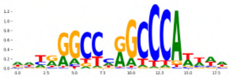
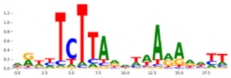
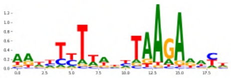
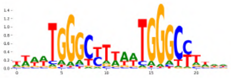
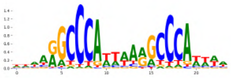
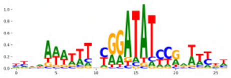
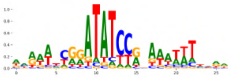
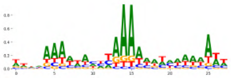
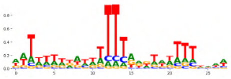
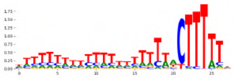
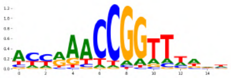
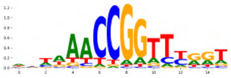
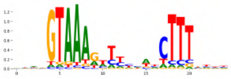
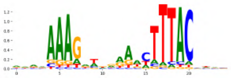
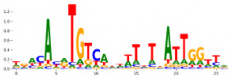
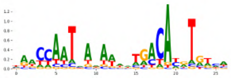
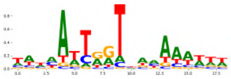
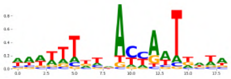
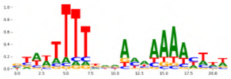
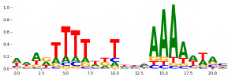
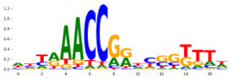
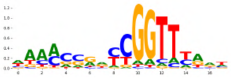
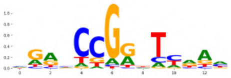
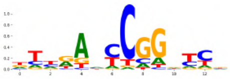
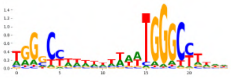
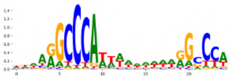
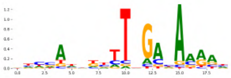
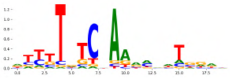
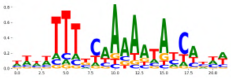
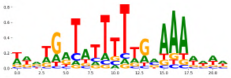
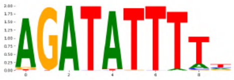
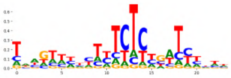
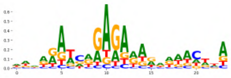
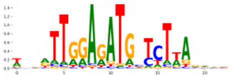
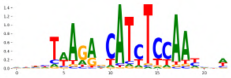
pattern	num_seqlets	modisco_cwm_fwd	modisco_cwm_rev	match0	qval0	match0_logo
pos_patterns.pattern_90	186					
pos_patterns.pattern_91	169					
pos_patterns.pattern_92	167					
pos_patterns.pattern_93	167					
pos_patterns.pattern_94	167					
pos_patterns.pattern_95	159			AT4G38000	0.005238	
pos_patterns.pattern_96	158					
pos_patterns.pattern_97	157					
pos_patterns.pattern_98	157					
pos_patterns.pattern_99	156					
pos_patterns.pattern_100	154					
pos_patterns.pattern_101	151					
pos_patterns.pattern_102	146					
pos_patterns.pattern_103	136					
pos_patterns.pattern_104	135					
pos_patterns.pattern_105	135			AT5G02840	0.041659	
pos_patterns.pattern_106	131					
pos_patterns.pattern_107	131					

Figure S5: (Continued)

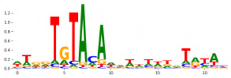
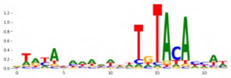
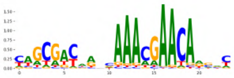
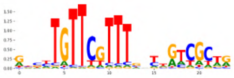
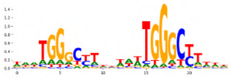
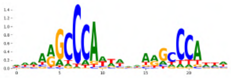
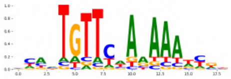
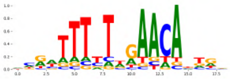
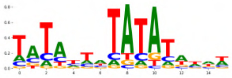
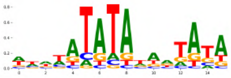
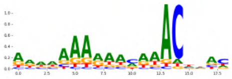
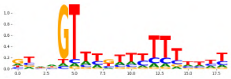
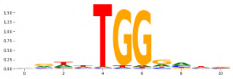
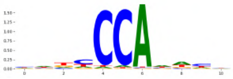
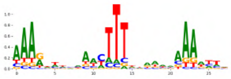
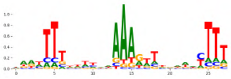
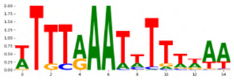
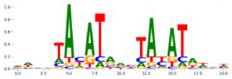
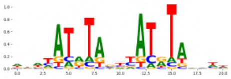
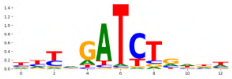
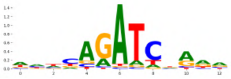
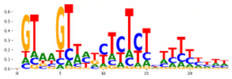
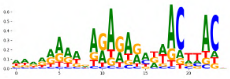
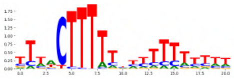
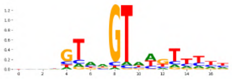
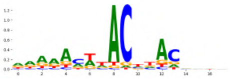
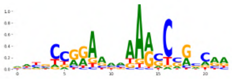
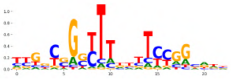
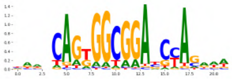
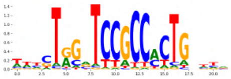
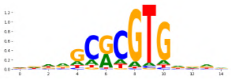
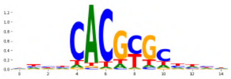
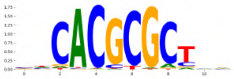
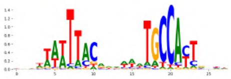
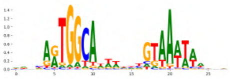
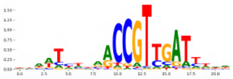
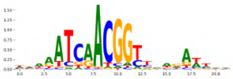
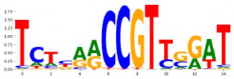
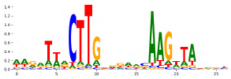
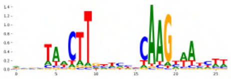
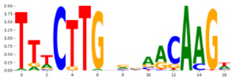
pattern	num_seqlets	modisco_cwm_fwd	modisco_cwm_rev	match0	qval0	match0_logo
pos_patterns.pattern_108	130					
pos_patterns.pattern_109	128					
pos_patterns.pattern_110	126					
pos_patterns.pattern_111	125					
pos_patterns.pattern_112	121					
pos_patterns.pattern_113	117					
pos_patterns.pattern_114	115					
pos_patterns.pattern_115	107			AT2G20110	0.049803	
pos_patterns.pattern_116	106					
pos_patterns.pattern_117	105					
pos_patterns.pattern_118	105			AT3G55370	0.025597	
pos_patterns.pattern_119	104					
pos_patterns.pattern_120	103					
pos_patterns.pattern_121	102					
pos_patterns.pattern_122	100			AT3G22170	0.007859	
pos_patterns.pattern_123	96					
pos_patterns.pattern_124	93			AT4G24470	0.000435	
pos_patterns.pattern_125	91			AT3G10500	0.000013	

Figure S5: (Continued)

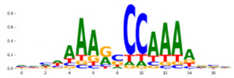
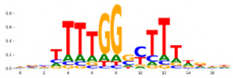
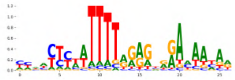
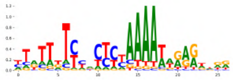
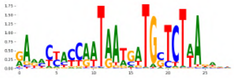
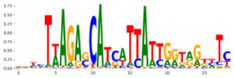
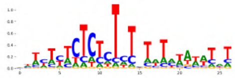
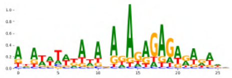
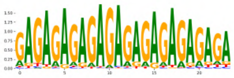
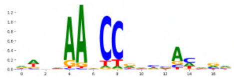
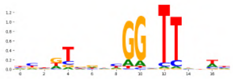
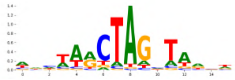
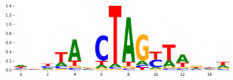
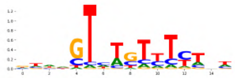
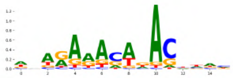
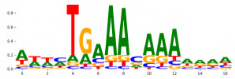
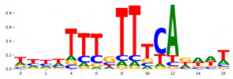
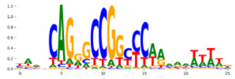
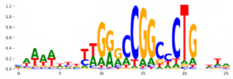
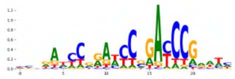
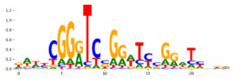
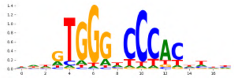
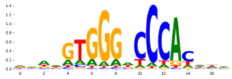
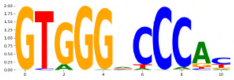
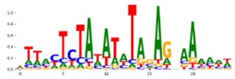
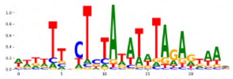
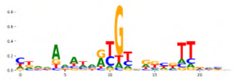
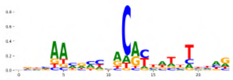
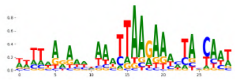
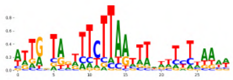
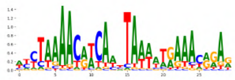
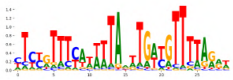
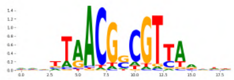
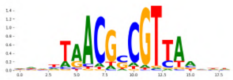
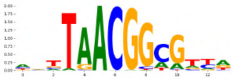
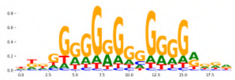
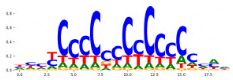
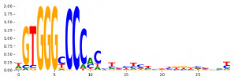
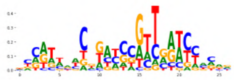
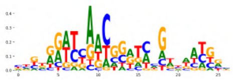
pattern	num_seqlets	modisco_cwm_fwd	modisco_cwm_rev	match0	qval0	match0_logo
pos_patterns.pattern_126	88					
pos_patterns.pattern_127	88					
pos_patterns.pattern_128	87					
pos_patterns.pattern_129	86			AT2G01930	0.007347	
pos_patterns.pattern_130	80					
pos_patterns.pattern_131	79					
pos_patterns.pattern_132	75					
pos_patterns.pattern_133	72					
pos_patterns.pattern_134	67					
pos_patterns.pattern_135	63					
pos_patterns.pattern_136	61			AT5G23280	0.000262	
pos_patterns.pattern_137	61					
pos_patterns.pattern_138	61					
pos_patterns.pattern_139	56					
pos_patterns.pattern_140	55					
pos_patterns.pattern_141	54			AT3G10030	0.000039	
pos_patterns.pattern_142	52			AT1G67260	0.031614	
pos_patterns.pattern_143	52					

Figure S5: (Continued)

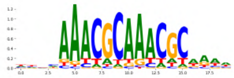
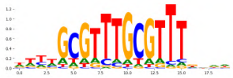
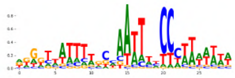
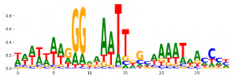
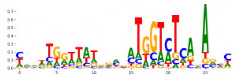
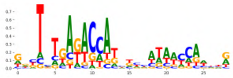
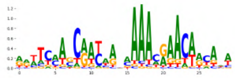
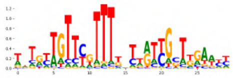
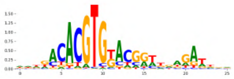
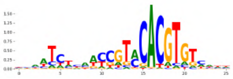
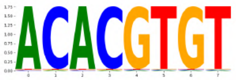
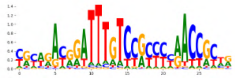
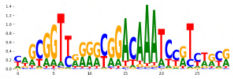
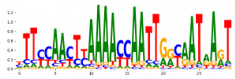
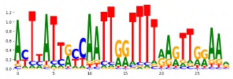
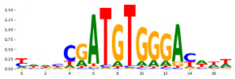
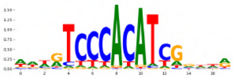
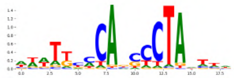
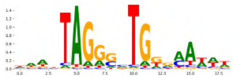
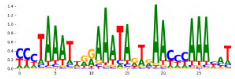
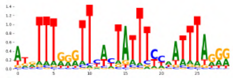
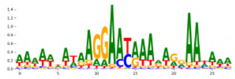
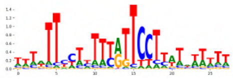
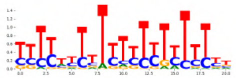
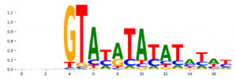
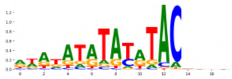
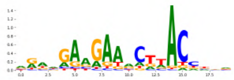
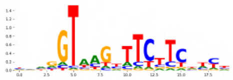
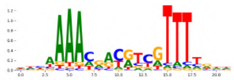
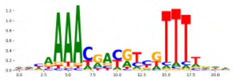
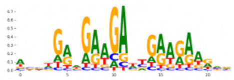
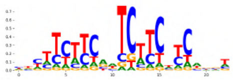
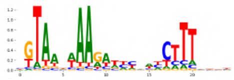
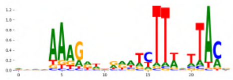
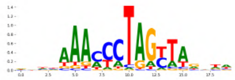
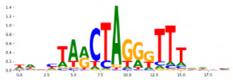
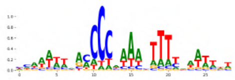
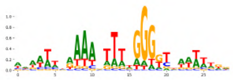
pattern	num_seqlets	modisco_cwm_fwd	modisco_cwm_rev	match0	qval0	match0_logo
pos_patterns.pattern_144	49					
pos_patterns.pattern_145	49					
pos_patterns.pattern_146	45					
pos_patterns.pattern_147	41					
pos_patterns.pattern_148	40			AT4G34000	0.002944	
pos_patterns.pattern_149	39					
pos_patterns.pattern_150	35					
pos_patterns.pattern_151	35					
pos_patterns.pattern_152	32					
pos_patterns.pattern_153	29					
pos_patterns.pattern_154	28			AT2G45660	0.02262	
pos_patterns.pattern_155	28					
pos_patterns.pattern_156	28					
pos_patterns.pattern_157	27					
pos_patterns.pattern_158	27					
pos_patterns.pattern_159	26					
pos_patterns.pattern_160	26					
pos_patterns.pattern_161	23					

Figure S5: (Continued)

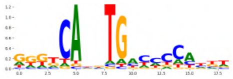
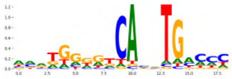
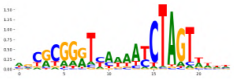
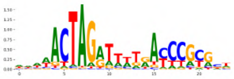
pattern	num_seqlets	modisco_cwm_fwd	modisco_cwm_rev	match0	qval0	match0_logo
pos_patterns.pattern_162	23					
pos_patterns.pattern_163	22					

Figure S5: (Continued)

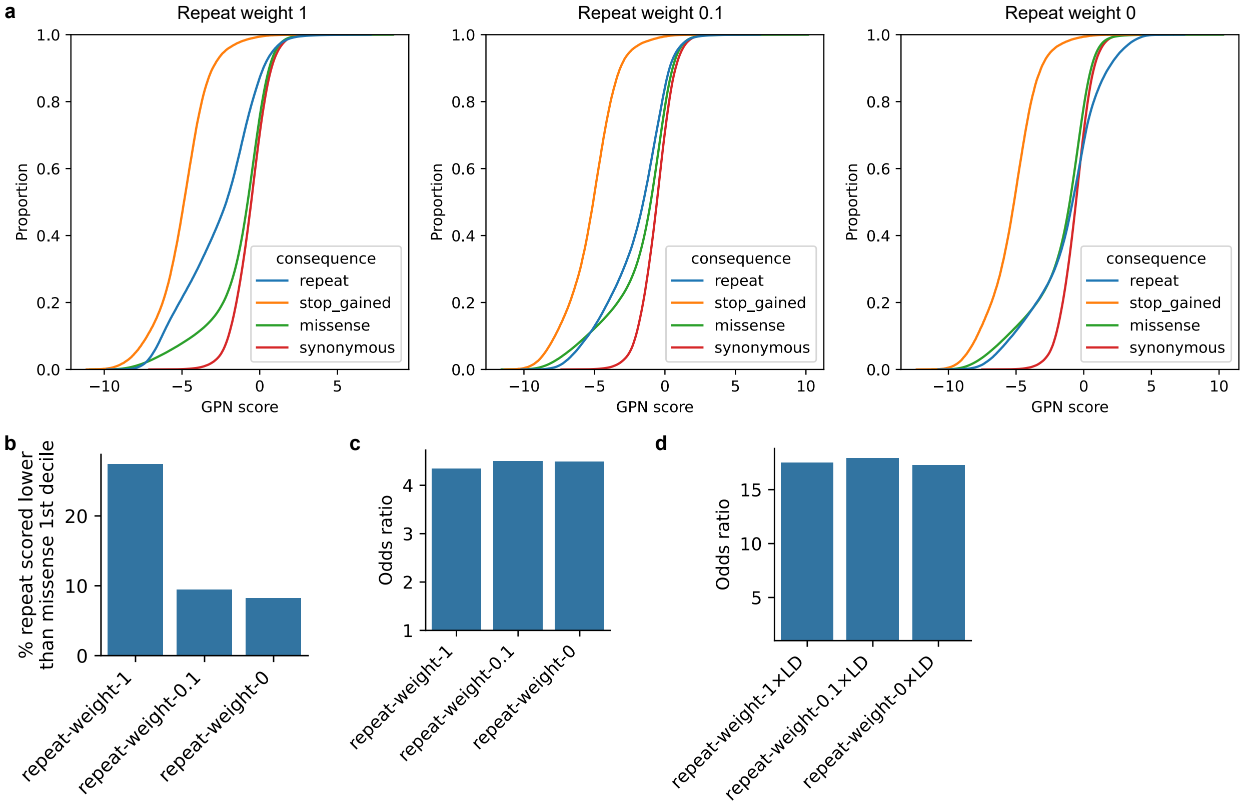


Figure S6: Comparison of GPN models trained with different loss weights on repeats. (a) Cumulative distribution function of GPN scores for simulated variants in specific categories, as described in Fig. 4. (b) Percentage of simulated repeat variants scored lower than the first decile of simulated missense variants. (c) Odds ratios for rare (AC = 1) vs. common (AF $\geq$ 5%) variants, as described in Fig. 5c. AC: allele count. AF: allele frequency. (d) Odds ratios for GWAS hits, as described in Fig. 6c.

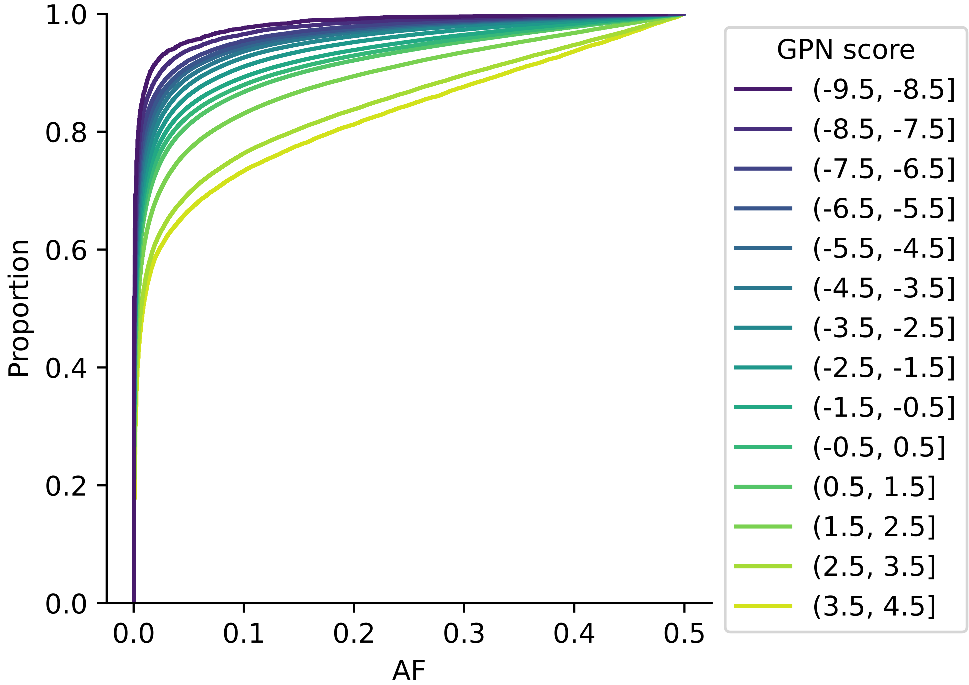


Figure S7: Cumulative distribution function of allele frequency (AF) for variants in different GPN score bins, as described in Fig. 5b.

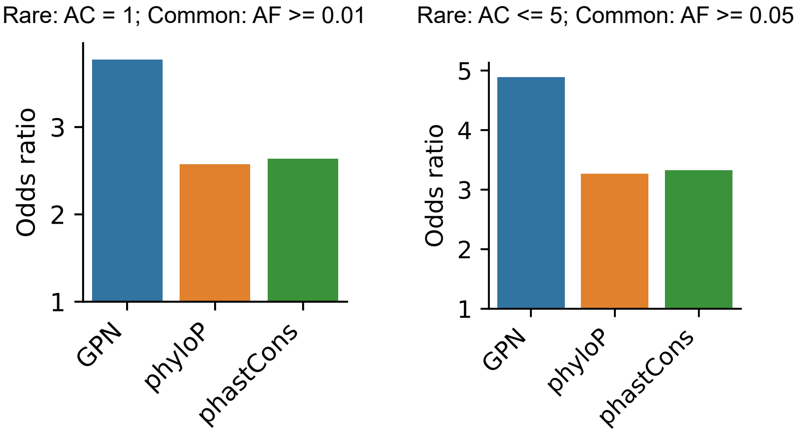


Figure S8: Rare vs. common odds ratios for different thresholds for defining rare and common variants. Odds ratios (OR) were calculated as described in Fig. 5c.

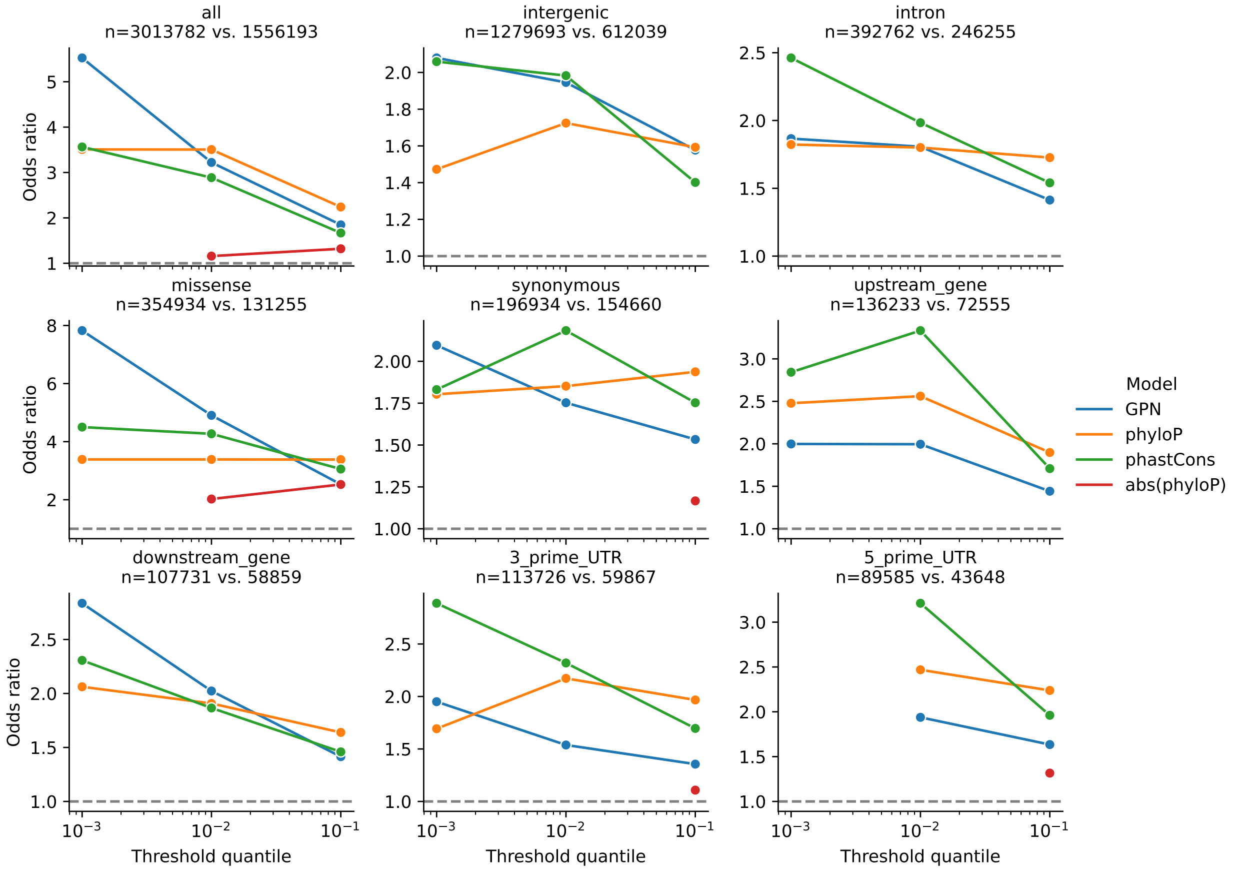


Figure S9: Rare vs. common odds ratios for specific variant categories and different thresholds for defining functional scores. Odds ratios (OR) were calculated as described in Fig. 5c. Only significant odds ratios are shown. The most stringent threshold in 5' UTR was excluded due to certain models having less than 10 counts in an entry of the contingency table.

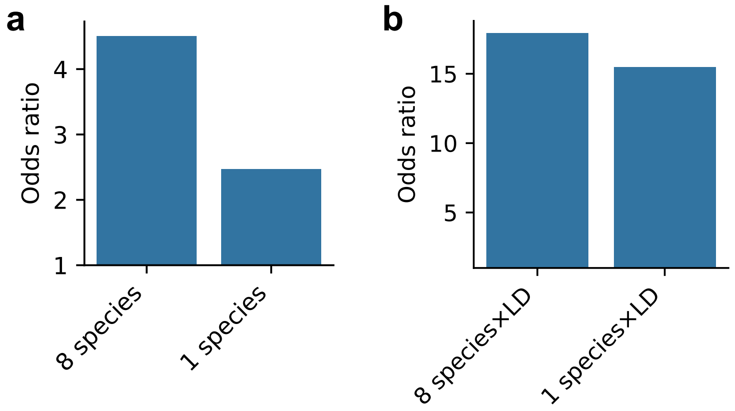


Figure S10: Comparison of GPN models trained on a different number of species. (a) Odds ratios for rare (AC = 1) vs. common (AF ≥ 5%), as described in Fig. 5c. AC: allele count. AF: allele frequency. (b) Odds ratios for GWAS hits, as described in Fig. 6c.

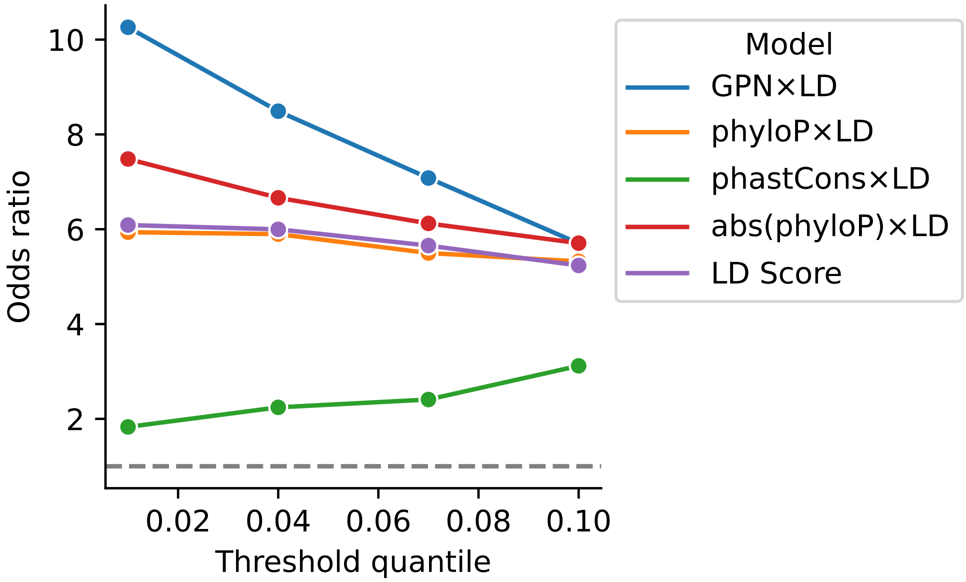


Figure S11: GWAS hit odds ratios for different thresholds for defining functional-tagged scores. Odds ratios (OR) were calculated as described in Fig. 6c.

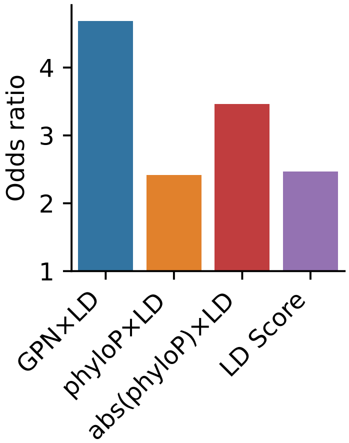


Figure S12: Odds ratios for GWAS hits, using the Bonferroni correction instead of permutation-based significance threshold, as described in Fig. 6c. Only significant odds ratios are shown.