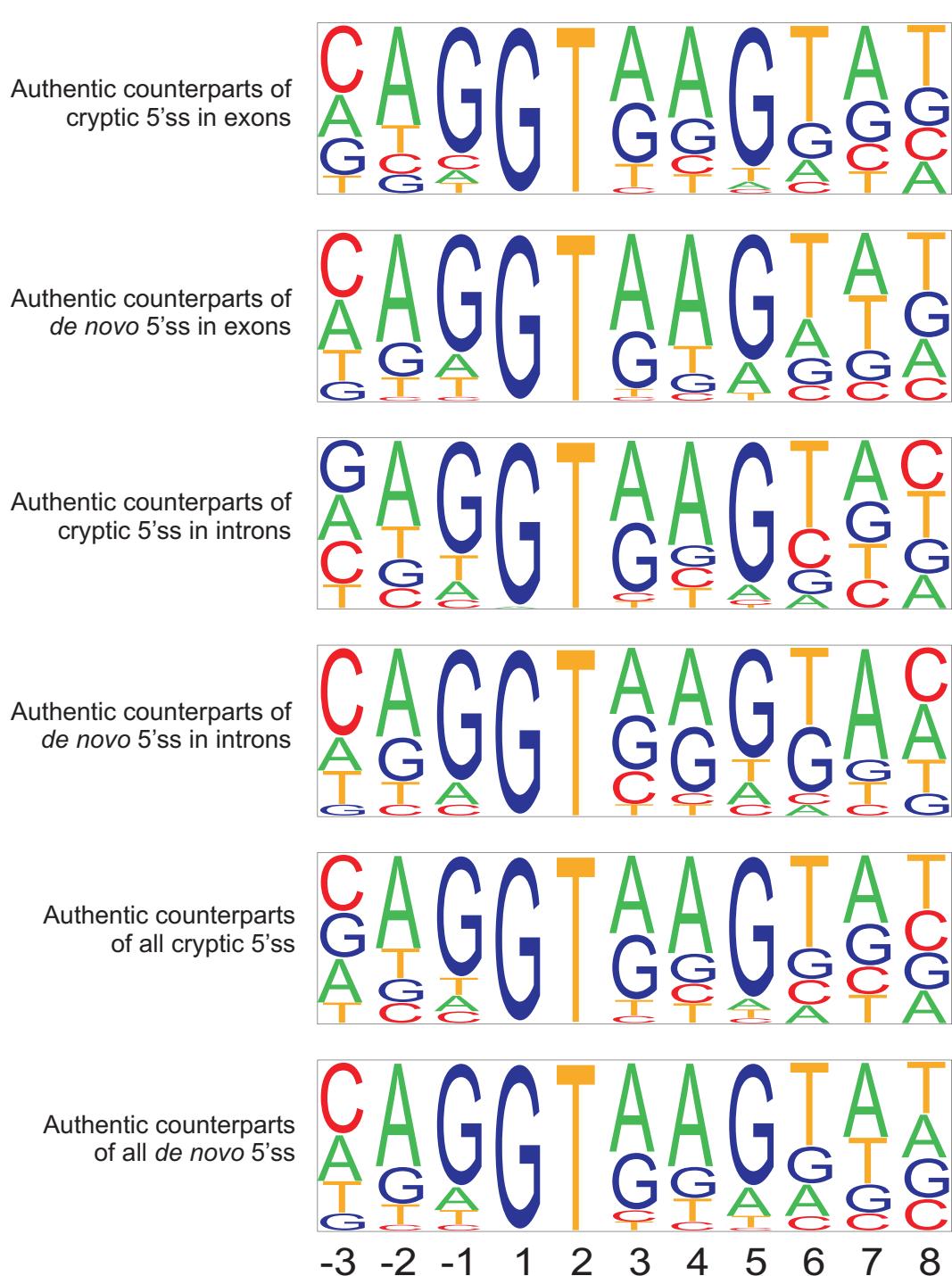
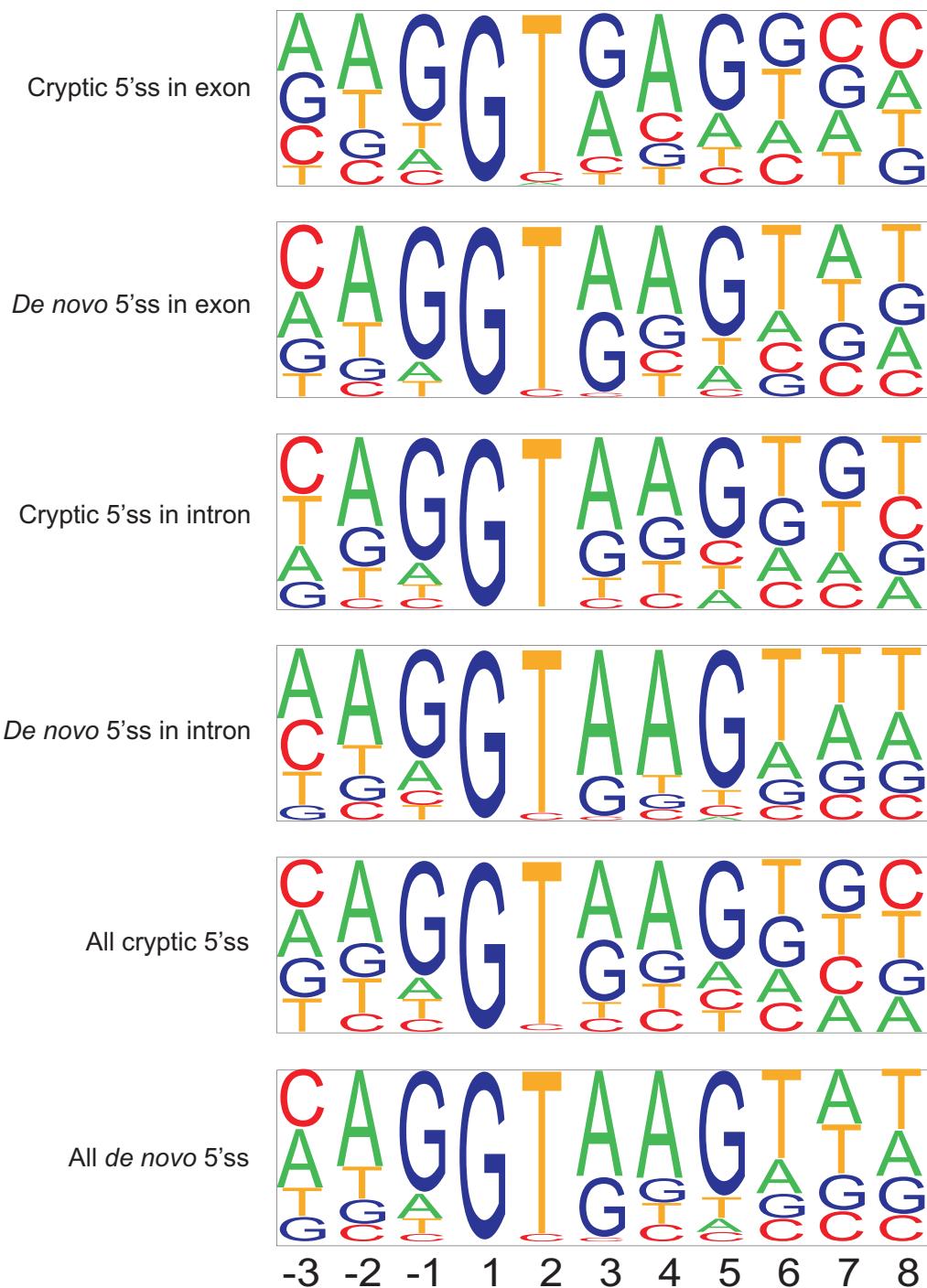


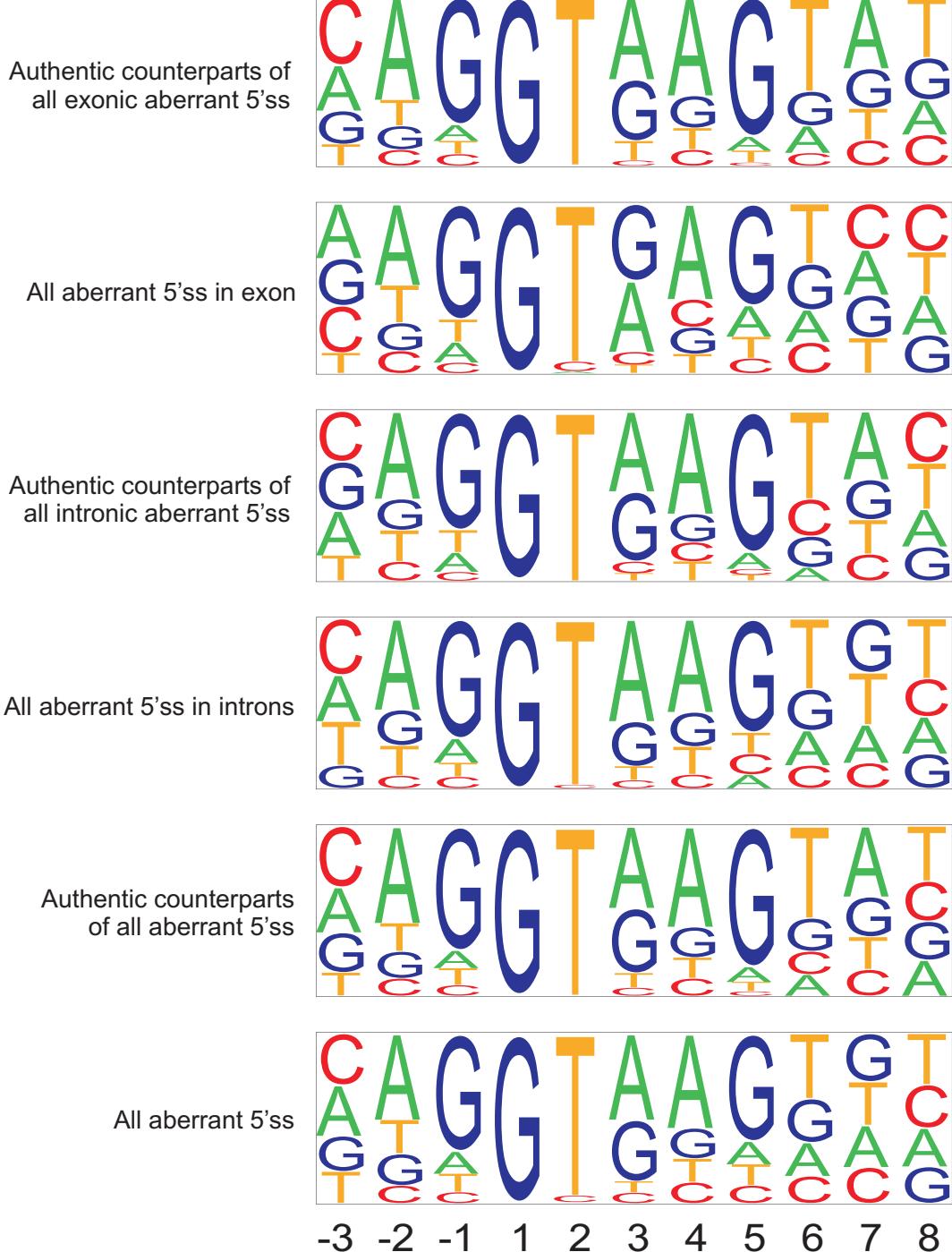
Supplementary Fig. 1



Supplementary Fig. 2



Supplementary Fig. 3



Supplementary Fig. 4

A

Search DBASS5 - Microsoft Internet Explorer provided by MEDIS

<http://www.soton.ac.uk/research/geneticsdb/dbass5/adsearch.aspx>

File Edit View Favorites Tools Help

Google Search DBASS5

University of Southampton School of Medicine

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Research : Human Genetics Division

Search DBASS5 (Advanced)

Advanced Search

Construct a search criteria by selecting values to search for from the items below. This will return any records that match all of the criteria selected. Note that you can select multiple values for the list boxes by holding down CTRL when clicking on the value.

Phenotype	Acute intermittent porphyria Acute lymphoblastic leukemia Adenosine deaminase deficiency Alpha mannosidase Alpha-1 antitrypsin deficiency
Gene	ABCD1 ADA ADAMTS13 ALG3 AR
Mutation	[IVS1+198G>T] [IVS1+1G>A] [IVS1+1G>C] [IVS1+1G>T] [IVS1+2T>A]
Location of Aberrant 5' Splice Sites:	IVS1 IVS10 IVS100 IVS11 IVS12
Distance from Natural Splice Site:	= <input type="text"/> (in nt, with "+" for exons, or "-" for introns)
Terminal Exon?	N/A
Nucleotide Sequence of the Authentic or Aberrant 5' Splice Sites	<input type="text"/>
Reference	<input type="text"/>
Score	All <input type="button" value="Shapiro & Senapathy Matrix"/> = <input type="button"/>
<input type="button" value="Search"/> <input type="button" value="Reset"/>	

B

DBASS5 - Aberrant Splice Site Database - Microsoft Internet Explorer provided by MEDIS				
File Edit View Favorites Tools Help Live Search				
Home View Splice Sites Search Submit Subscribe Consensus Sequences Credit Terms of Use Contact	DBASS5 - Splice Site Details			
	Gene	<i>HMBS</i>	Location of Abberant 5' Splice Site	Exon
	Phenotype	Acute intermittent porphyria	Distance between Authentic and Aberrant 5' Splice Site (nt)	-9
	Mutation	E10+114G>T	Change in the Reading Frame	0
	Splice Site Strength	Authentic 5'ss		Cryptic 5'ss
	Shapiro & Senapathy Matrix Score ^[1]	84.05		69.33
	Maximum Entropy Model ^[2]	9.46		3.69
	Maximum Dependence Decomposition Model ^[3]	13.08		7.88
	First-Order Markov Model ^[4]	9.04		5.1
	Weight Matrix Model ^[5]	6.11		6.11
	H-Bond Score ^[6]	15.7		9.5
	Free Energy ^[5]	-9.2		-8.6
	Base Pairing with U1 snRNA ^[6]	8		8
	Neural Network ^[7]	0.63		0.13
	Reference(s)	Delfau, M. H., Picat, C., De Rooij, F., Voortman, G., Deybach, J. C., Nordmann, Y., and Grandchamp, B. (1991). Molecular heterogeneity of acute intermittent porphyria: identification of four additional mutations resulting in the CRM-negative subtype of the disease. Am J Hum Genet 49, 421-428. Pub Med Article # 1714233		
	Comments	-		
	Nucleotide Sequence ^[8]	<pre> cttaaagtcgt agagctggaa gatgactcaa agtca gggccag cttagagggg aaaggacagac tcaggcagag ggaaaccgcac ggccggcccg atggccgac actgttgtcc tttagcaacttc tccacatc CGG GGAAACCTCA ACACCGGCT TCCGAAGCT GACGCCAGC AGGGATTCAG TGCCATCATC CTGGCAACAG CTGGCCCTCA GGGCATGGG TTGGCACAAAC GG</GGGGG A(GT)tag ggcctggccccc tatccctcc ccggctcatc tgcattctcc ttctgcctta cagtcattcca caatgtttaga ttttttagat ttatgtatgt gtggaaaggcg tatacgttca tgcggaaactcg tacttttgatcc cccataggcc atctttgttt tttatcttcg tacatgcgtatcata </pre>		
	Terminal Exon?	No		
	Percent Utilisation	Not determined		
Legend <i>, /</i> , cryptic or <i>de novo</i> splice sites; <i>></i> nucleotide substitutions (in parentheses); <i>()</i> , deletions; <i>[]</i> , duplications or insertions. Intronic sequences are in lower case (in blue), exons are in upper case.				