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参考資料1: IMPA2遺伝子解析に用いたマーカー

SNP ID*	HUGO Nomenclature†	Database Information		Comment
		dbSNP ID‡	Nucleotide Position in the UCSC§	
SNP01	-1209T>G	rs971363	11970460	
SNP02	-1051G>T	rs971362	11970618	
SNP03	-708G>A <sup>  </sup>	-	11970961	
SNP04	-461C>T <sup>  ,¶</sup>	rs2075824	11971208	"C" was preferentially transmitted to bipolar <sup>¶</sup>
	-241_-237insGGGCT <sup>†</sup>	-	11971431	Minor allele (-241_-237ins) frequencies were 1% in both bioplar and control groups
SNP05	-207T>C <sup>¶</sup>	-	11971462	"T" was preferentially transmitted to bipolar <sup>¶</sup>
SNP06	-185A>G <sup>  ,¶</sup>	-	11971484	"A" was marginally preferentially transmitted to bipolar ( $P = 0.05$ ) <sup>¶</sup> "A" was Associated with schizophrenia <sup>  </sup>
SNP07	IVS1+1801C>T	rs3786305	11973565	
SNP08	IVS1+1916C>T	rs644710	11973680	
	IVS1-4890T>C	rs605964	11984163	Failed to design TaqMan probes
	IVS1-4800T>C	rs3786302	11984253	Failed to design TaqMan probes
	IVS1-4795G>A	rs590277	11984258	Failed to design TaqMan probes
SNP09	IVS1-4639C>T	rs607200	11984414	
	IVS1-4605C>G	rs3786299	11984448	Failed to design TaqMan probes
SNP10	IVS1-15G>A <sup>  ,¶</sup>	-	11989038	Associated with schizophrenia <sup>  </sup>
SNP11	159T>C (L53L) <sup>  ,¶</sup>	rs11545506	11989115	
SNP12	IVS2-3630C>T	rs3786291	11996252	
SNP13	IVS3+660G>A	rs3786284	12000646	
SNP14	IVS4+968A>C	rs3786282	12003182	
	IVS4-880C>T	rs628419	12003384	Ambiguous genotype
SNP15	IVS5+13-14insA <sup>  ,¶</sup>	-	12004385	
SNP16	558C>T (F186F) <sup>  </sup>	rs2075825	12018109	Associated with schizophrenia <sup>  </sup>
	IVS6+99A>G	rs2075826	12018249	Ambiguous genotype
SNP17	IVS6-261G>A	rs613993	12018580	
	IVS7-200DelG	rs3837885	12020142	Failed to design TaqMan probes
SNP18	Ex8+2108T>C	rs640088	12022984	
SNP19	Ex8+2157T>C	rs594235	12023033	

\*Only the SNPs whose genotypes were unambiguously determined and minor allele frequencies were more than 1% were assigned maker ID.

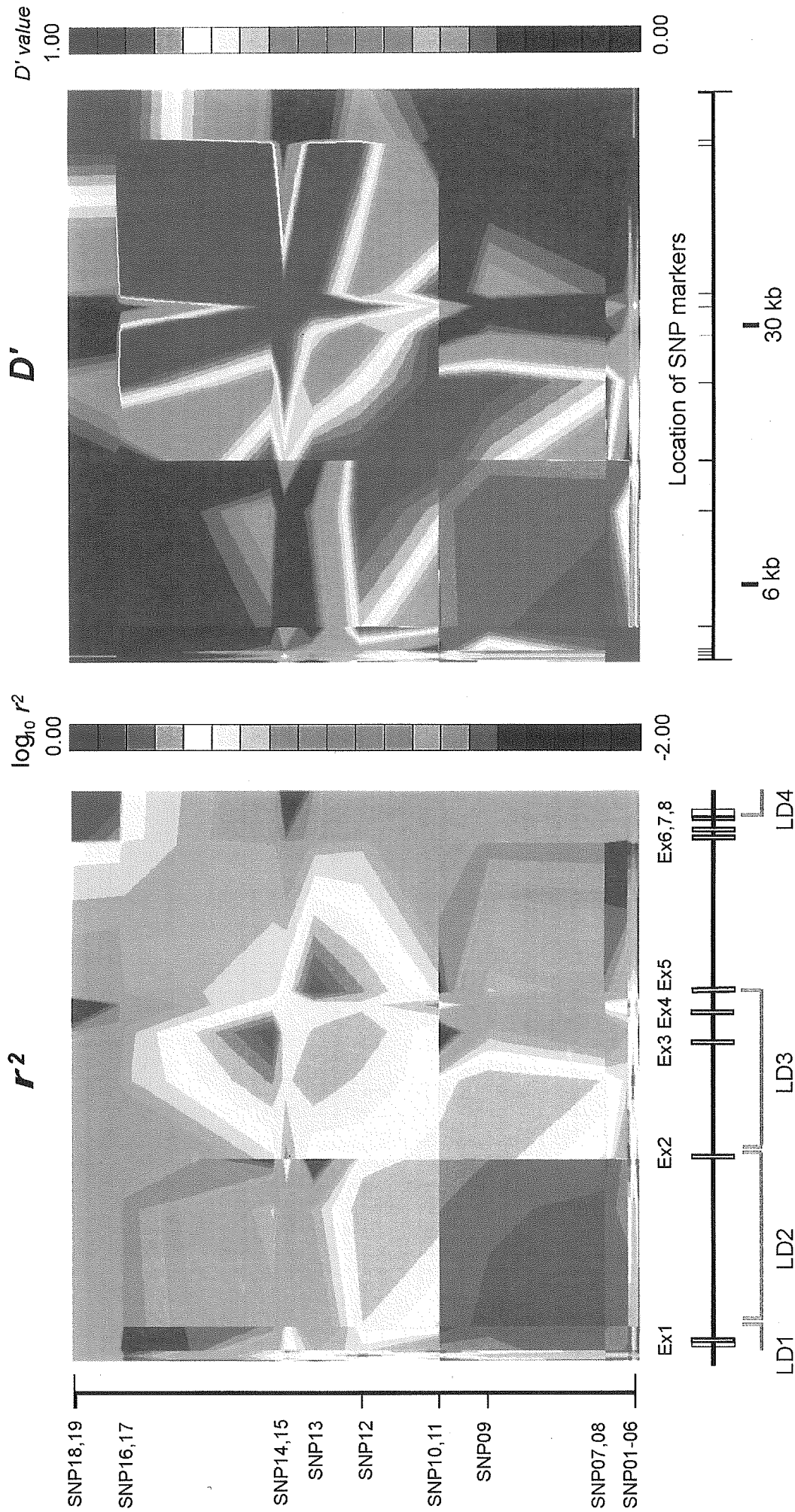
†<http://www.genomic.unimelb.edu.au/mdi/>

‡<http://www.ncbi.nlm.nih.gov/SNP/>

§Human May 2004, <http://genome.ucsc.edu/cgi-bin/hgGateway>

<sup>||</sup>Yoshikawa *et al.* 2001

<sup>¶</sup>Sjoholt *et al.* 2004



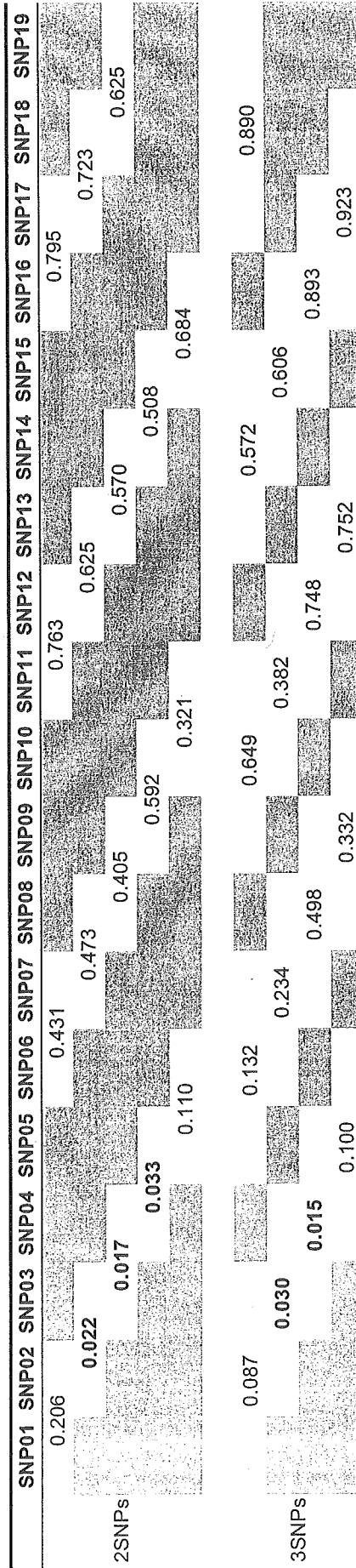
参考資料2  
IMPA2遺伝子のLDブロック構造

参考資料3:IMPA2遺伝子のSNP関連解析

Polymorphism	Sample	n	Allele Counts		P value	Genotype Counts			P value	HWE	Minor Allele
						T/T	T/G	G/G			Frequency
SNP01 -1209T>G	Bipolar	488	T	G	0.479	T/T	T/G	G/G	0.447	0.390	0.320
	Control	534	710	358		230	204	54			0.560
SNP02 -1051G>T	Bipolar	488	G	T	0.143	G/G	T/G	T/T	0.304	0.529	0.456
	Control	534	546	522		141	249	98			0.255
SNP03 -708G>A	Bipolar	486	G	A	0.462	G/G	A/G	A/A	0.511	0.583	0.147
	Control	537	903	171		382	139	16			0.442
SNP04 -461C>T	Bipolar	487	C	T	<b>0.042</b>	C/C	T/C	T/T	0.118	0.494	0.455
	Control	536	536	536		141	249	97			0.490
SNP05 -207T>C	Bipolar	489	T	C	<b>0.046</b>	T/T	T/C	C/C	0.114	0.202	0.450
	Control	537	543	531		141	256	92			0.363
SNP06 -185G>A	Bipolar	489	G	A	0.197	G/G	G/A	A/A	0.184	0.596	0.372
	Control	538	705	371		238	229	71			0.179
SNP07 IVS1+1801C>T	Bipolar	492	C	T	0.423	C/C	T/C	T/T	0.661	0.603	0.314
	Control	539	721	357		234	207	51			0.982
SNP08 IVS1+1916T>C	Bipolar	492	T	C	0.965	T/T	T/C	C/C	0.503	0.858	0.496
	Control	538	541	535		126	244	122			0.058
SNP09 IVS1-4639T>C	Bipolar	494	T	C	0.508	T/T	T/C	C/C	0.756	0.598	0.466
	Control	540	593	487		144	240	110			0.281
SNP10 IVS1-15G>A	Bipolar	492	G	A	0.157	G/G	A/G	A/A	0.339	0.573	0.307
	Control	538	714	362		239	204	49			0.984
SNP11 159T>C	Bipolar	490	T	C	0.713	T/T	T/C	C/C	0.263	0.346	0.232
	Control	541	839	243		293	167	30			0.192
SNP12 IVS2-3630C>T	Bipolar	488	C	T	0.519	C/C	T/C	T/T	0.347	0.614	0.130
	Control	539	927	151		368	113	7			0.114
SNP13 IVS3+660G>A	Bipolar	485	G	A	0.630	G/G	A/G	A/A	0.600	0.981	0.121
	Control	532	943	121		415	113	4			0.215
SNP14 IVS4+968C>A	Bipolar	486	C	A	0.615	C/C	A/C	A/A	0.865	0.539	0.382
	Control	537	676	398		215	246	76			0.677
SNP15 IVS5+13-14InsA	Bipolar	486	Del	Ins	0.394	Del/Del	Ins/Del	Ins/Ins	0.407	0.518	0.115
	Control	532	954	110		424	106	2			0.085
SNP16 558C>T	Bipolar	491	C	T	1.000	C/C	T/C	T/T	0.049	0.002	0.113
	Control	534	947	121		418	111	5			0.424
SNP17 IVS6-261G>A	Bipolar	492	G	A	0.523	G/G	A/G	A/A	0.348	0.860	0.271
	Control	541	774	308		262	193	37			0.032
SNP18 Ex8+2108T>C	Bipolar	494	T	C	0.627	T/T	T/C	C/C	0.493	0.674	0.281
	Control	542	768	316		282	204	56			0.039
SNP19 Ex8+2157T>C	Bipolar	486	T	C	0.591	T/T	T/C	C/C	0.510	0.639	0.283
	Control	534	754	314		276	202	56			0.040

Significant P values have been shown in bold type. HWE, P value for Hardy-Weinberg equilibrium.

参考資料4:IMPA2遺伝子のハプロタイプsliding window解析

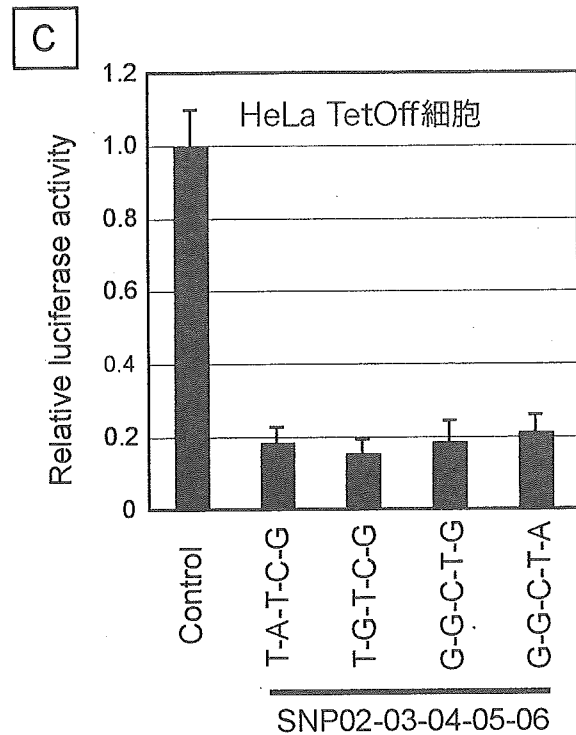
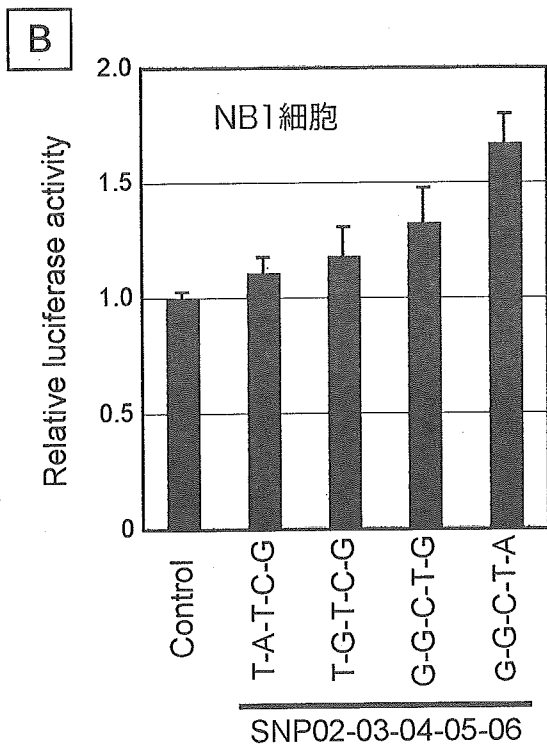
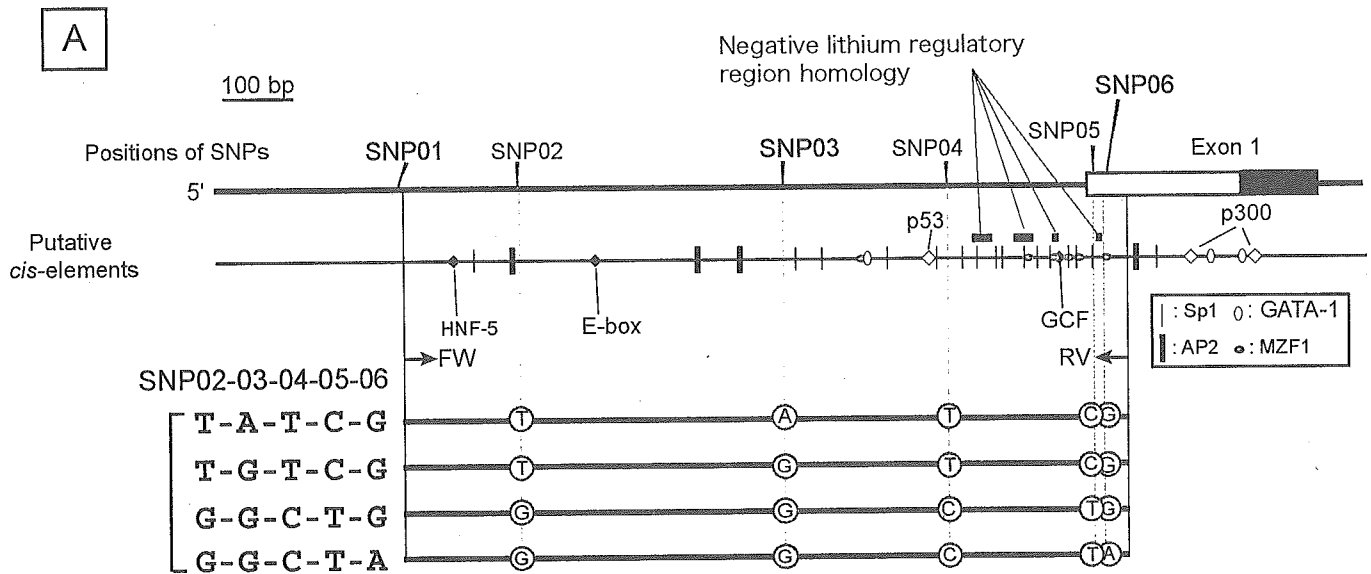


Significant *P* values have been shown in bold type.

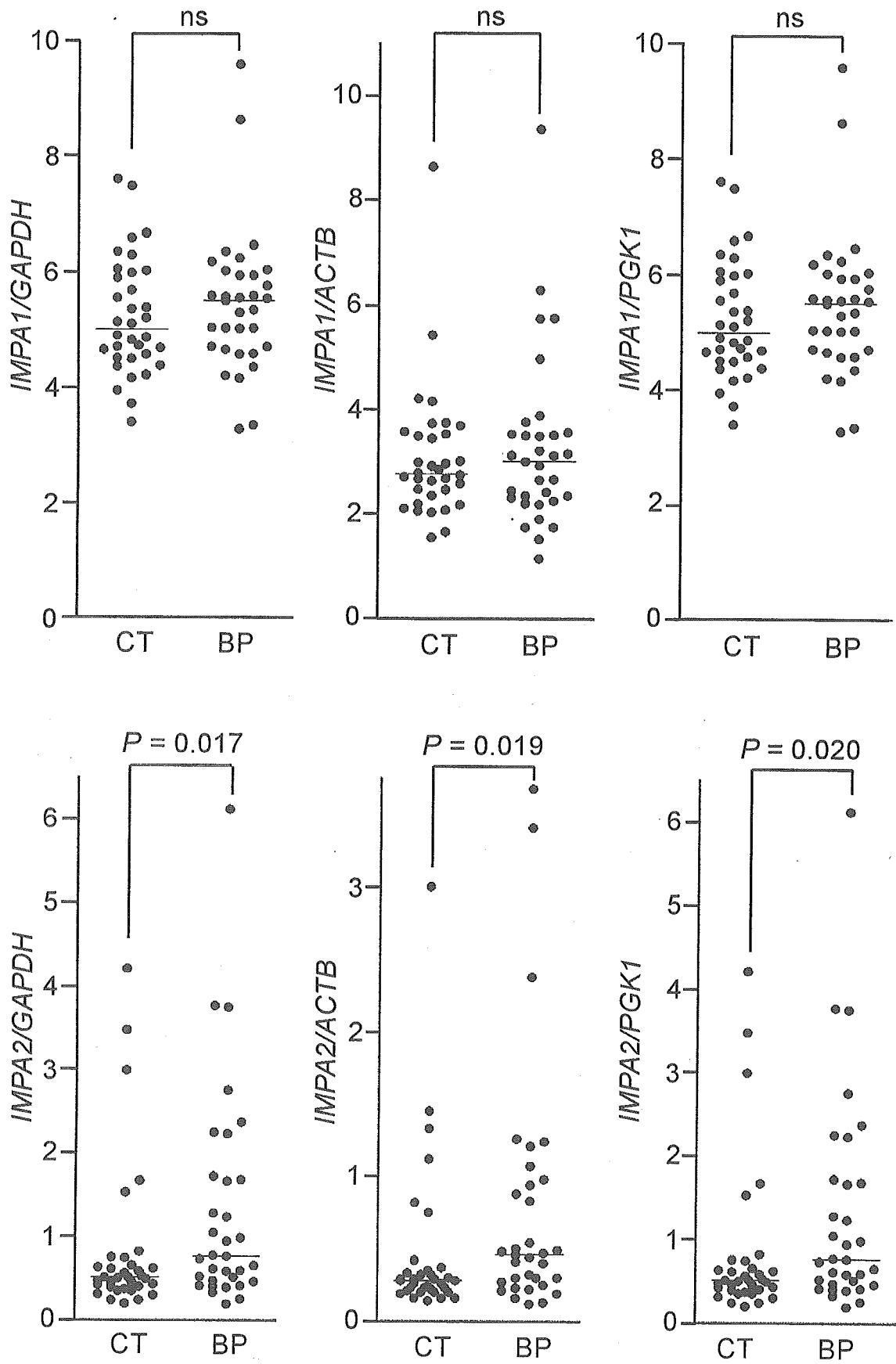
参考資料5: IMPA2遺伝子のプロモーターハプロタイプ頻度

Haplotype (SNP02-03-04-05-06)	Bipolars (n = 496)	Controls (n = 543)	$\chi^2$	P value	Odds ratio
T-A-T-C-G	0.136	0.155	1.386	0.239	0.859
T-G-T-C-G	0.320	0.335	0.462	0.497	0.936
G-G-C-T-G	0.165	0.159	0.124	0.725	1.045
G-G-C-T-A	0.380	0.352	1.576	0.209	1.126

Haplotype frequencies were computed by the COCAPHASE program.



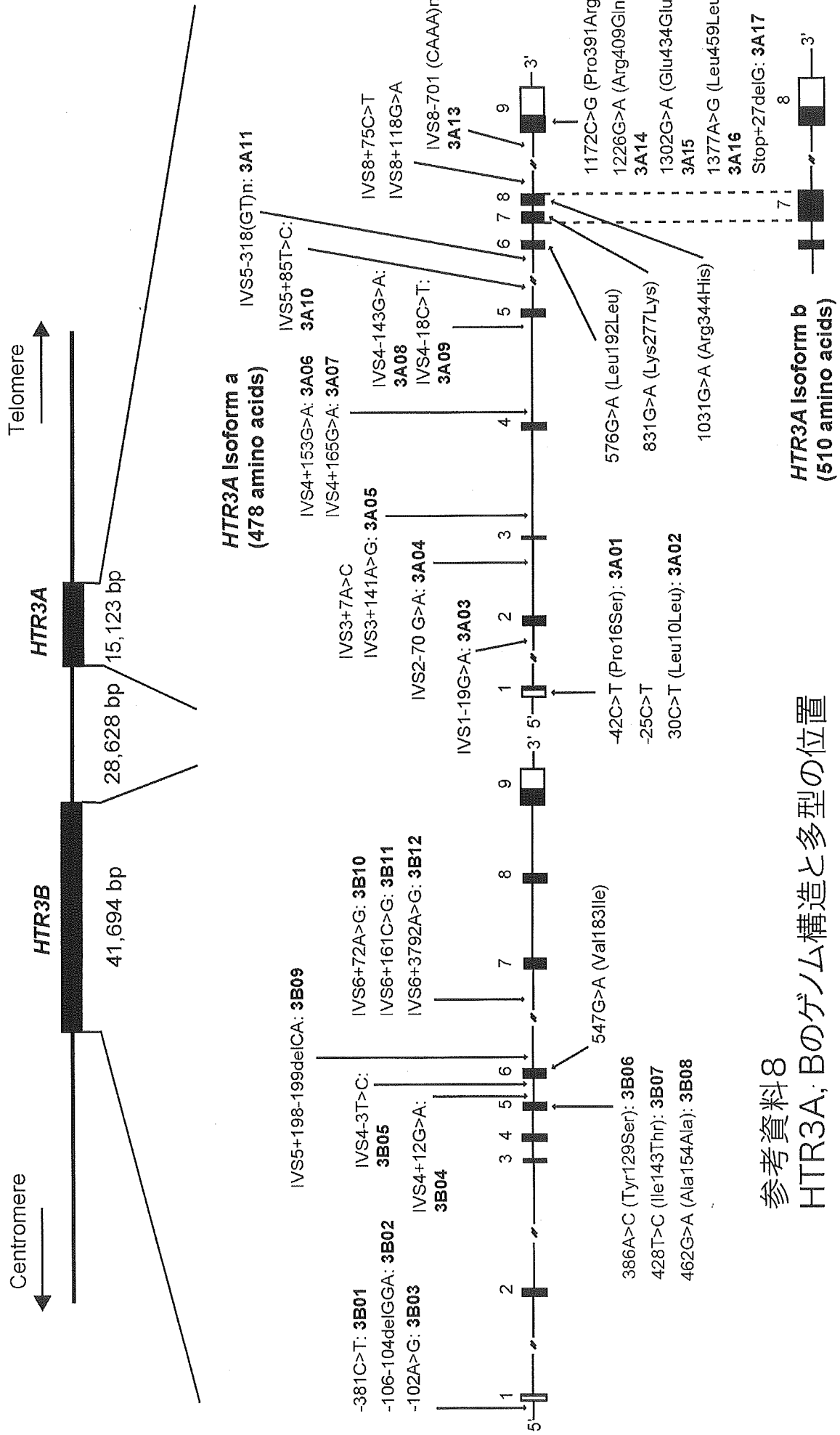
参考資料6  
 IMPA2プロモーターコンストラクトの設計と  
 機能解析



参考資料7  
死後脳でのIMPA2 mRNAの発現解析

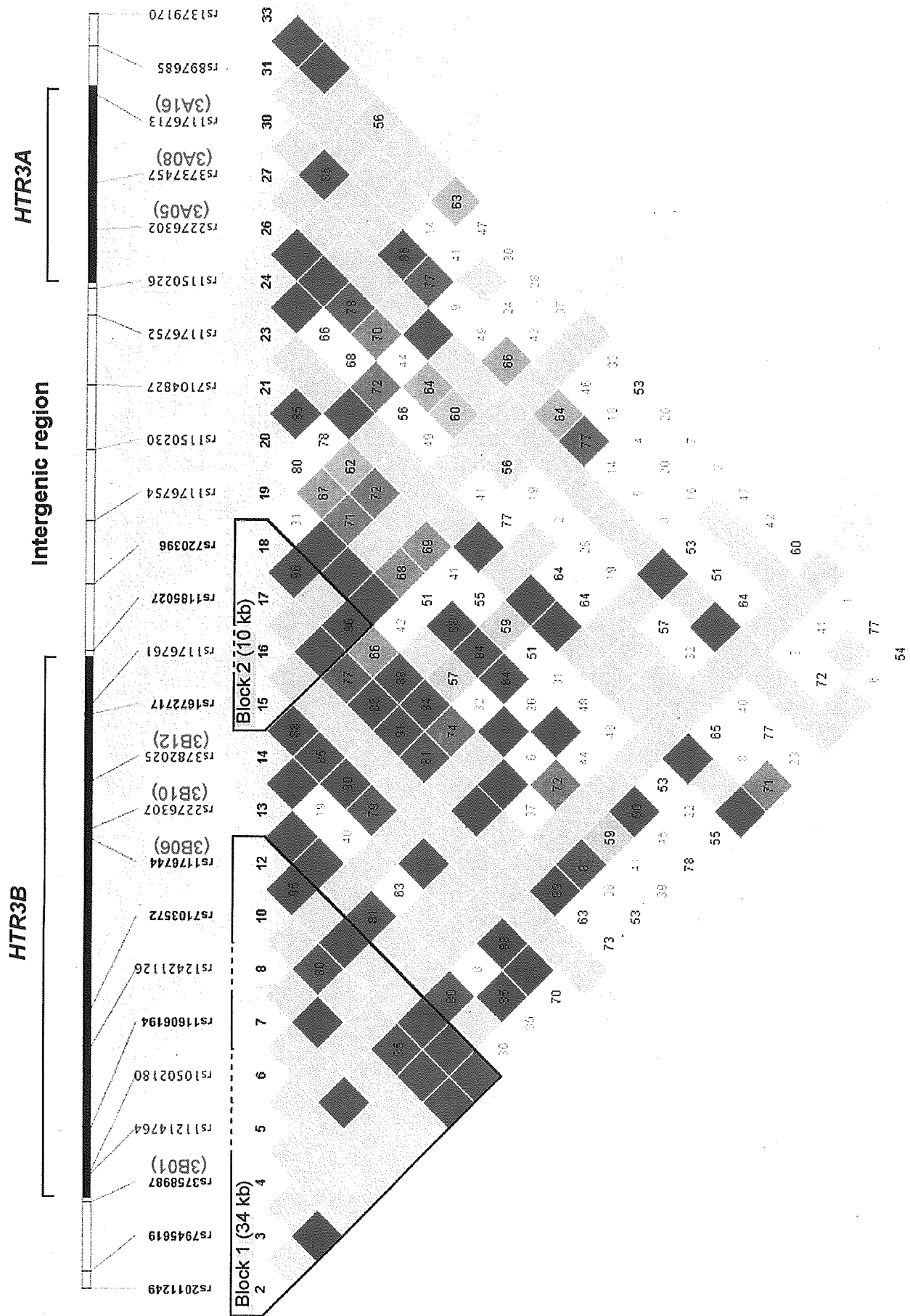


Chromosome 11q23

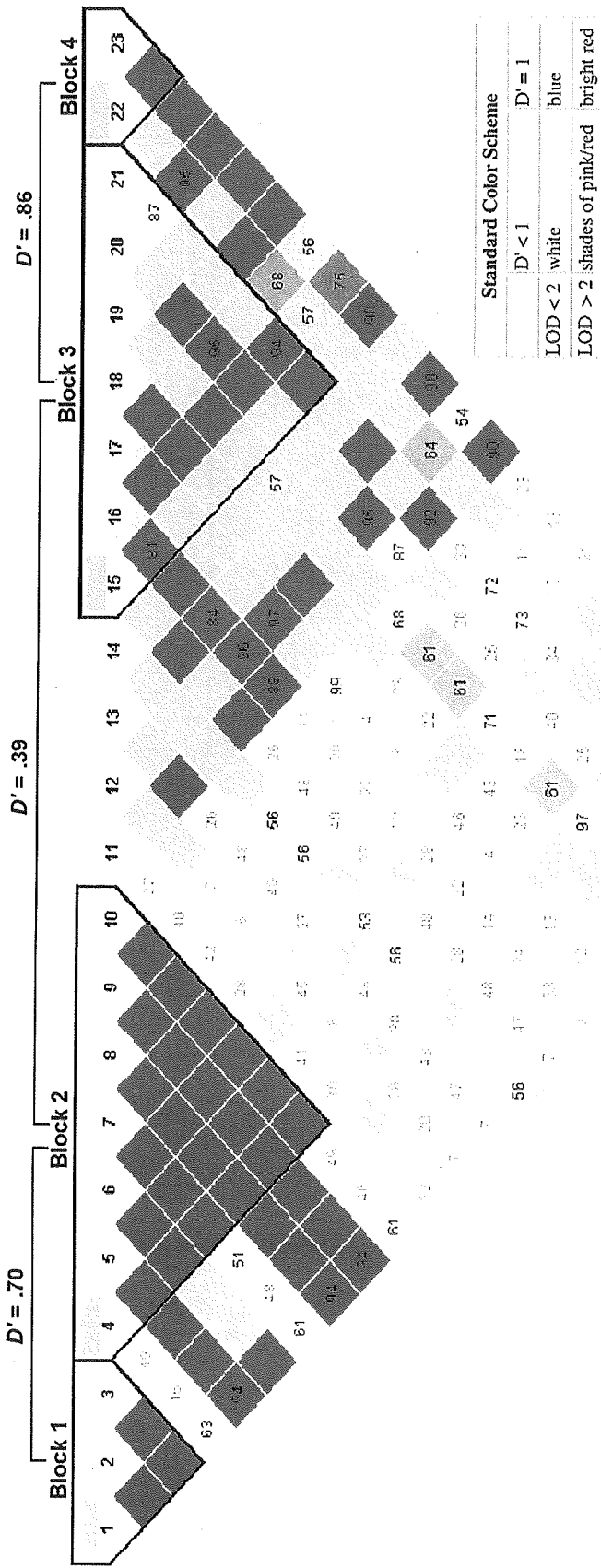
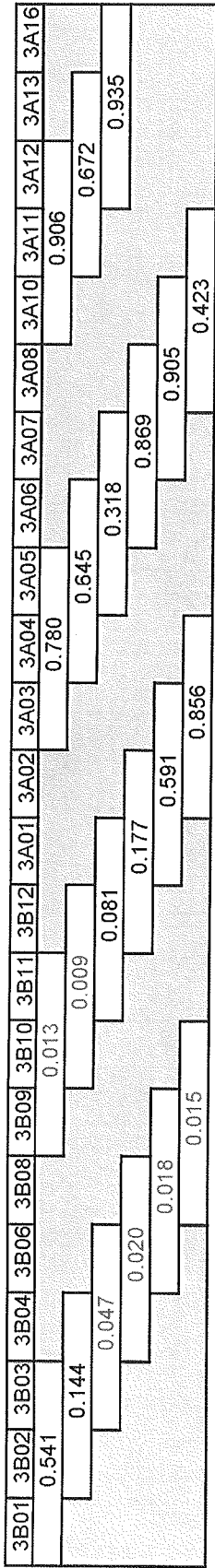


参考資料8

HTR3A, Bのゲノム構造と多型の位置



参考資料9: CaucasianにおけるHTR3A, B遺伝子のハプロタイプブロック構造



Block-based haplotype analysis

Block 1

T I A  
C I A  
T D G

case control  
.593 .517  
.233 .313  
.174 .171  
P = .377

Block 2

G A G I A C A  
G C G I G G A  
A A G D A C G  
G A A I A C A

case control  
.415 .303  
.195 .321  
.134 .253  
.256 .124  
P = .002

Block 3

A G G T A G  
A G G C A G  
G A G T A A  
A A A T T G  
A A A G T A G

case control  
.611 .633  
.125 .101  
.111 .108  
.111 .108  
.042 .051  
P = .984

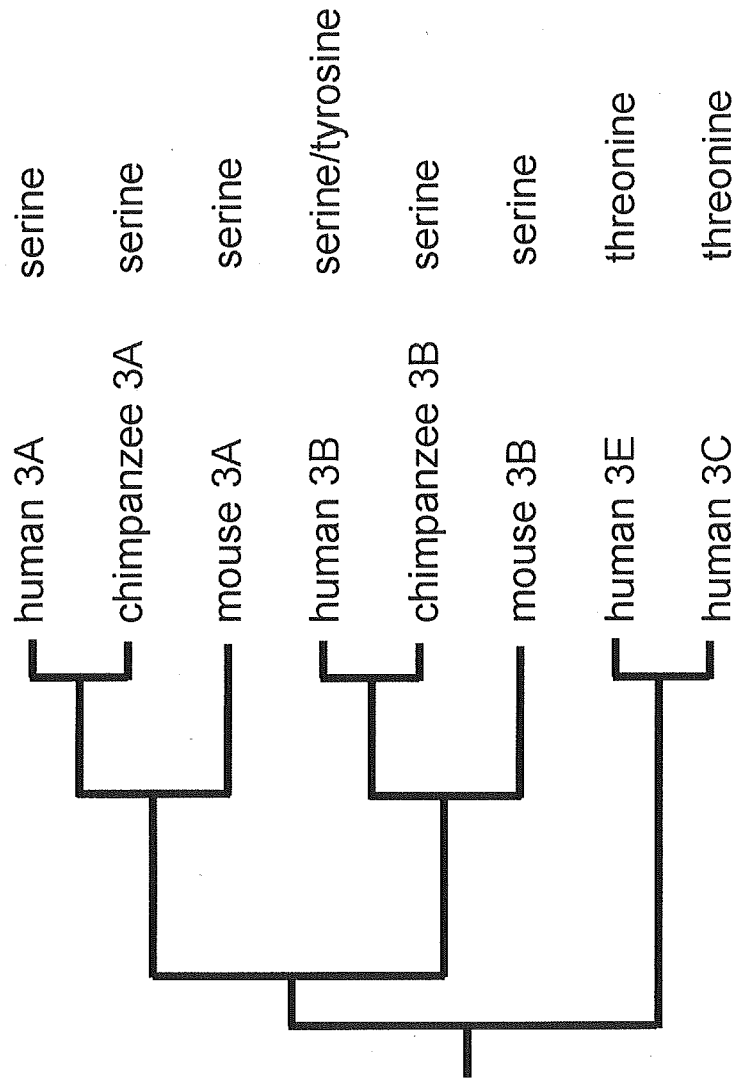
Block 4

T A  
T G  
A G

case control  
.750 .771  
.125 .100  
.125 .130  
P = .832

参考資料10:日本人におけるHTR3A, B遺伝子のハプロタイプブロック構造と  
ハプロタイプ頻度解析

129th amino acid



参考資料11:セロトニン受容体3型遺伝子の系統樹