

Supplemental Material

Titles and Legends of Supplemental Material

Supplemental Table I

List of all the AGRE individuals and families used in the sequencing and genotyping stages. First two columns on the left list the 48 individuals and families included in stage 1 sequencing. Next two columns on the center indicate the 981 individuals (327 trios) genotyped in stage 2. Last 2 columns on the right list the 1011 individuals (337 trios) genotyped in the third stage.

Supplemental Table II

List of all the AGRE individuals, used in the complex indel screening. First two columns on the left list the families and the 189 autistic individuals. Next two columns on the right indicate the families and 319 unaffected siblings used as controls.

Supplemental Table III

SNPs used in stage 3 genotyping. The 18 tagging SNPs with a MAF=5% and higher used in stage 3 are listed. The first column on the left indicates the SNP ID. The second column denotes the SNP location in bp within the *AHI* gene (Ensembl). The third column lists the alleles and the last column on the right indicates the SNP # for the analysis. All the SNPs were selected using HapMap data NCBI 35 build for the *AHII* gene and Tagger.

Supplemental Figure 1.

Pedigree of the 87405 Complex Indel in 2 AGRE Families. Family trees of families 1 (A) & 2 (B) are shown. Indel transmission is denoted by (-) when there is no indel and (+) when heterozygous indel is found. Filled figures indicated “autism” diagnosis, patched figure denotes “broad spectrum”. Squares are males and circles females.

First Stage (Sequencing)		Second Stage (Genotyping)		Third Stage (Genotyping)	
Family Code	Indiv. Code	Family Code	Indiv. Code	Family Code	Indiv. Code
AU0044	AU004403	AU0008	AU000801	AU0012	AU001201
AU0055	AU005504	AU0008	AU000802	AU0012	AU001202
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AU0107	AU010703	AU0017	AU001701	AU0017	AU001701
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AU0121	AU012103	AU0018	AU001804	AU0018	AU001804
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AU0159	AU015903	AU0024	AU002402	AU0021	AU002102
AU0175	AU017504	AU0024	AU002403	AU0021	AU002106
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AU0379	AU037906	AU0097	AU009703	AU0037	AU003705
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AU0493	AU049304	AU0133	AU013302	AU0044	AU004402
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AU0703	AU070303	AU0157	AU015703	AU0053	AU005303
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AU0733	AU073303	AU0161	AU016103	AU0055	AU005504
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		AU0217	AU021701	AU0081	AU008101
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		AU0217	AU021704	AU0081	AU008103
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		AU0231	AU0231301	AU0084	AU008403
		AU0238	AU023801	AU0085	AU008501
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		AU0283	AU028302	AU0095	AU009502
		AU0283	AU028304	AU0095	AU009503
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		AU0308	AU030801	AU0102	AU010204
		AU0308	AU030802	AU0102	AU010205
		AU0308	AU030804	AU0102	AU010209
		AU0316	AU0316201	AU0110	AU011001
		AU0316	AU0316202	AU0110	AU011002
		AU0316	AU0316301	AU0110	AU011004
		AU0358	AU035803	AU0116	AU011601
		AU0358	AU035804	AU0116	AU011602
		AU0358	AU035810	AU0116	AU011604
		AU0385	AU038501	AU0120	AU012001
		AU0385	AU038502	AU0120	AU012002
		AU0385	AU038503	AU0120	AU012003
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		AU0399	AU039902	AU0132	AU013202
		AU0399	AU039903	AU0132	AU013203
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		AU0436	AU043603	AU0140	AU014005
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		AU0455	AU045507	AU0146	AU0146201
		AU0455	AU045508	AU0146	AU0146202

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		AU0461	AU046103	AU0148	AU014803
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		AU0688	AU068802	AU0259	AU025902
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		AU0700	AU070002	AU0275	AU027502
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		AU0744	AU074403	AU0311	AU031104
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		AU0763	AU076302	AU0361	AU036102
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		AU0773	AU077303	AU0386	AU038603
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		AU0780	AU0780301	AU0399	AU039903
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		AU0782	AU0782301	AU0423	AU042303
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		AU0786	AU0786202	AU0427	AU042702
		AU0786	AU0786303	AU0427	AU042704
		AU0788	AU078801	AU0432	AU043201
		AU0788	AU078802	AU0432	AU043202
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		AU0811	AU081103	AU0461	AU046103
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		AU0836	AU083602	AU0504	AU050402
		AU0836	AU083603	AU0504	AU050403
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		AU0839	AU0839301	AU0509	AU050908
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		AU0841	AU084104	AU0521	AU052103
		AU0842	AU084201	AU0533	AU053301
		AU0842	AU084202	AU0533	AU053302
		AU0842	AU084203	AU0533	AU053303
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		AU0845	AU084503	AU0535	AU053503
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		AU0852	AU0852303	AU0538	AU053803
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		AU0862	AU0862302	AU0541	AU054103
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		AU0866	AU0866202	AU0543	AU054302
		AU0866	AU0866301	AU0543	AU054303
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		AU0868	AU086804	AU0545	AU054503
		AU0871	AU0871201	AU0550	AU055001
		AU0871	AU0871202	AU0550	AU055002
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		AU0875	AU0875201	AU0551	AU055101
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		AU0885	AU0885303	AU0555	AU055503
		AU0895	AU0895201	AU0561	AU056101
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		AU0899	AU089903	AU0565	AU056503
		AU0901	AU0901201	AU0566	AU056601
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Supplemental Table I

Affected Individuals		Controls	
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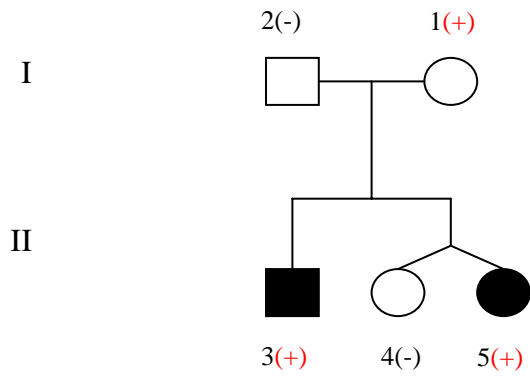
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Supplemental Table II

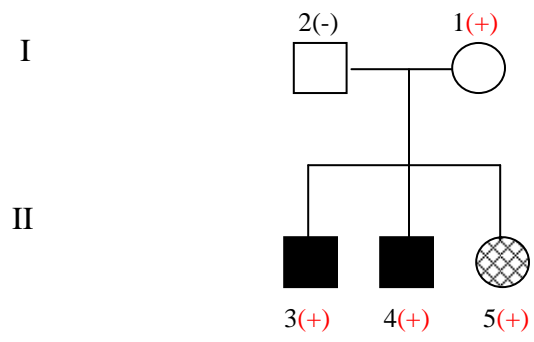
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rs7766656	135659294	A/G	5
rs4896141	135663293	C/G	6
rs4896142	135664512	T/G	7
rs2223802	135665277	C/T	8
rs2207000	135665526	C/T	9
rs6570000	135669367	A/G	10
rs6914831	135681337	C/T	11
rs4896144	135688385	T/C	12
rs17707754	135713904	T/C	14
rs2246943	135733209	A/T	15
rs6570004	135748959	A/T	16
rs17778438	135773887	G/A	17
rs7759971	135788577	C/T	20
rs717120	135806822	T/C	21
rs4526212	135846324	C/A	24

Supplemental Table III

A)



B)



Supplemental Figure 1.