

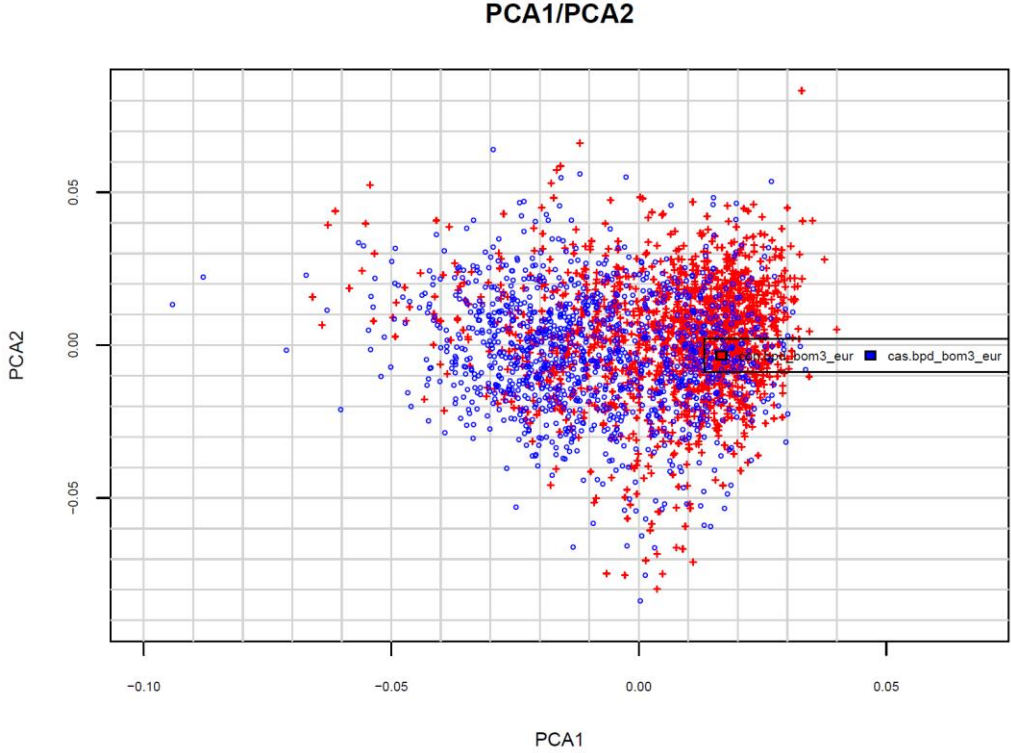
Supplementary Information for *Genome-wide association study of Borderline Personality Disorder reveals genetic overlap with the Major Psychoses*

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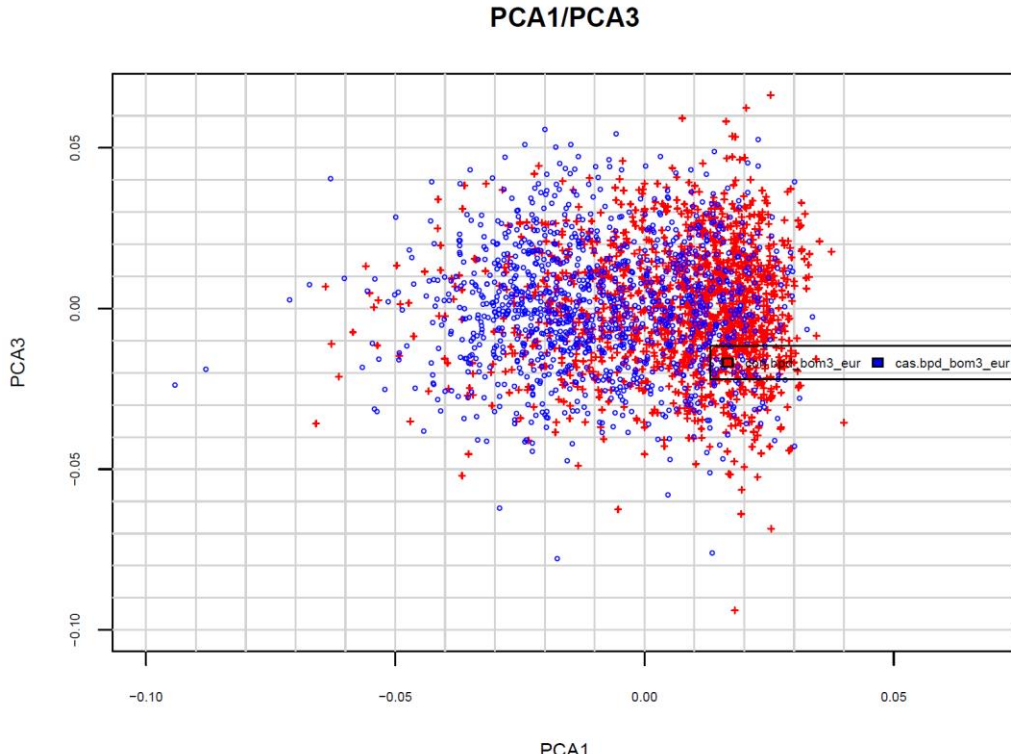
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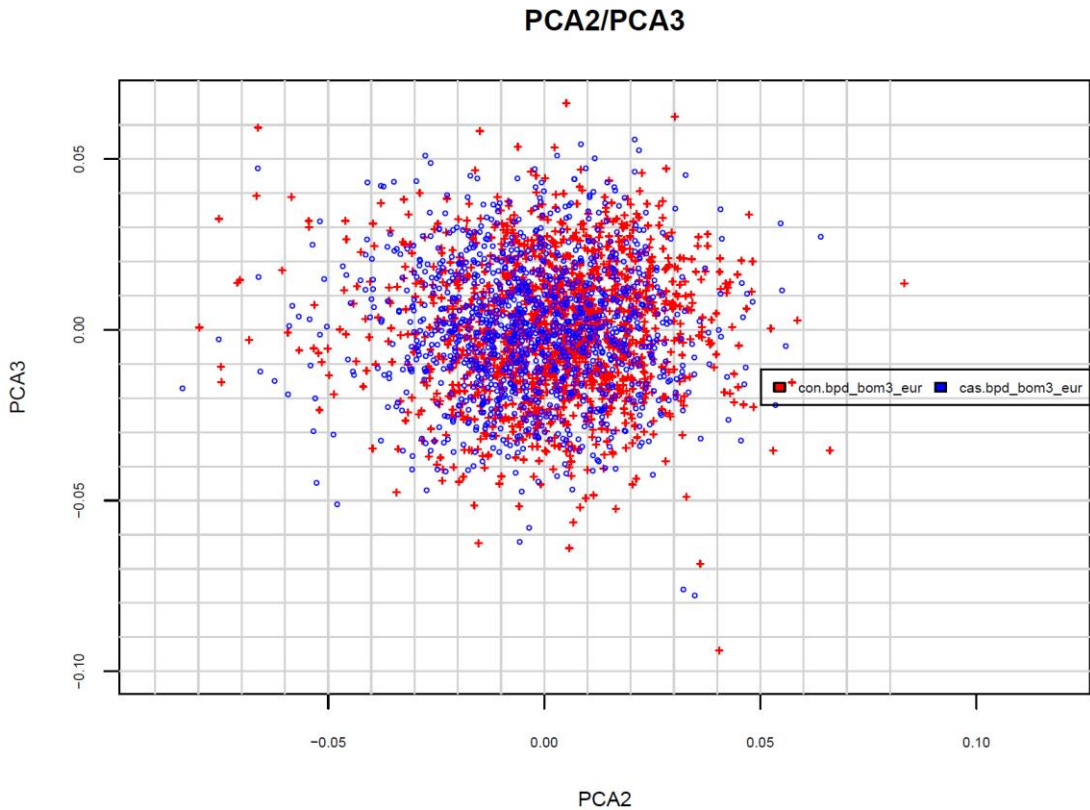
Supplementary Figure 1: Scatter plot of principal components 1 and 2



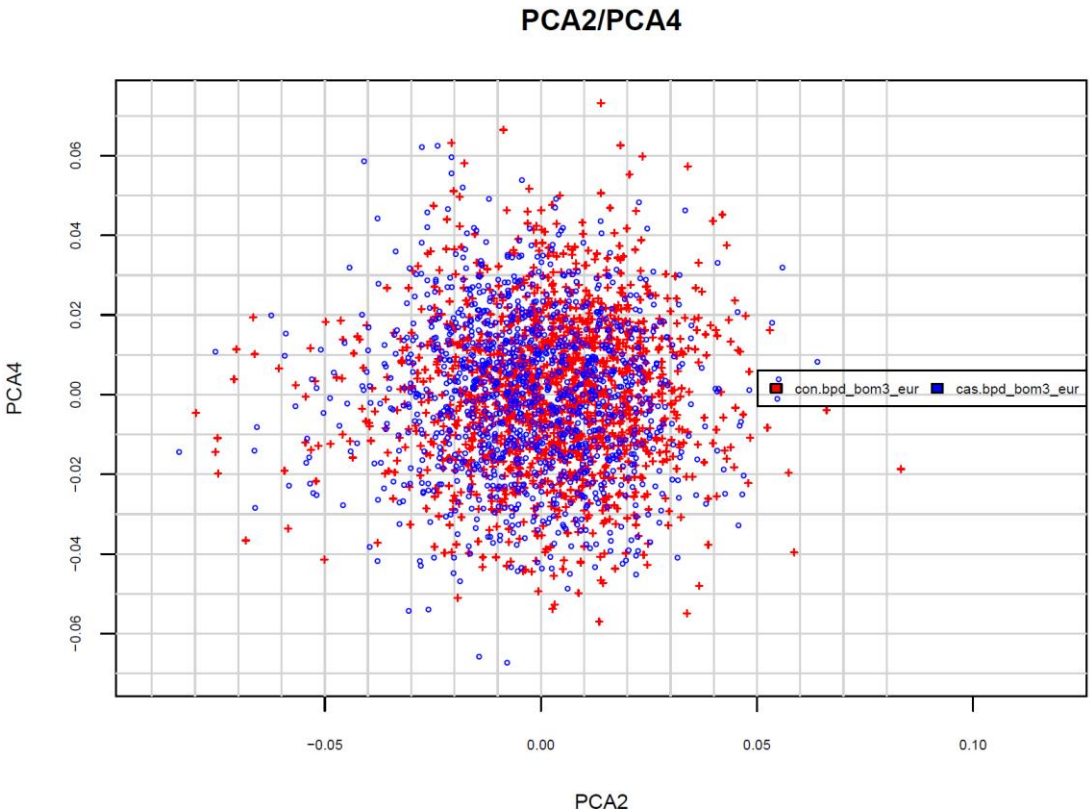
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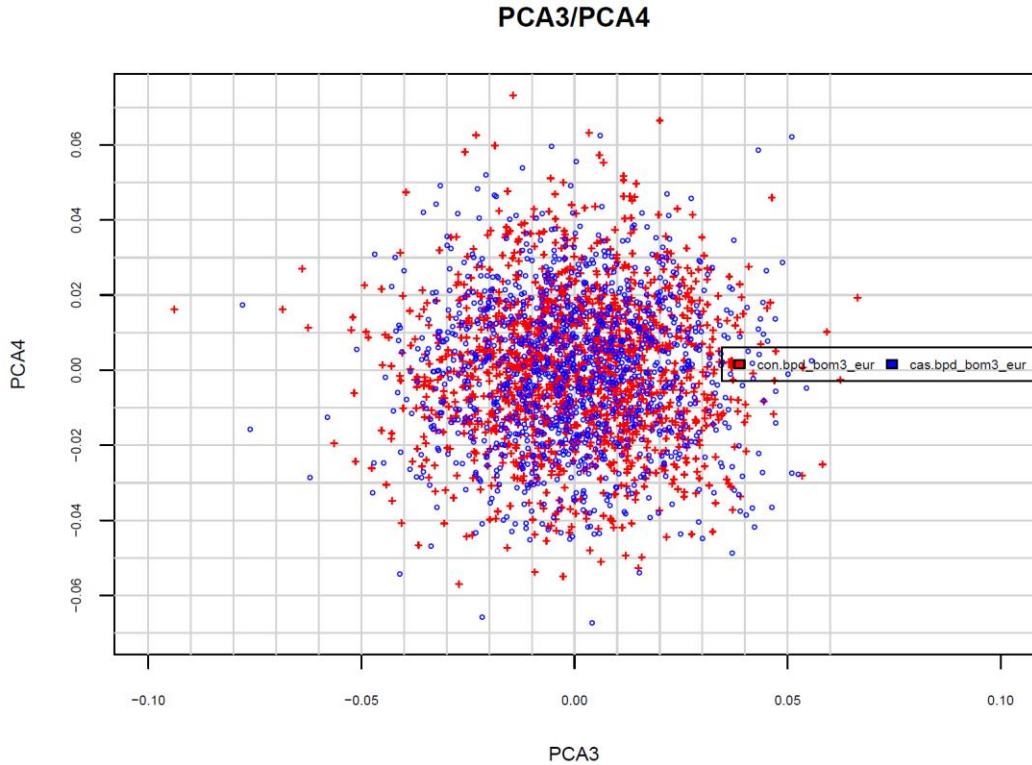
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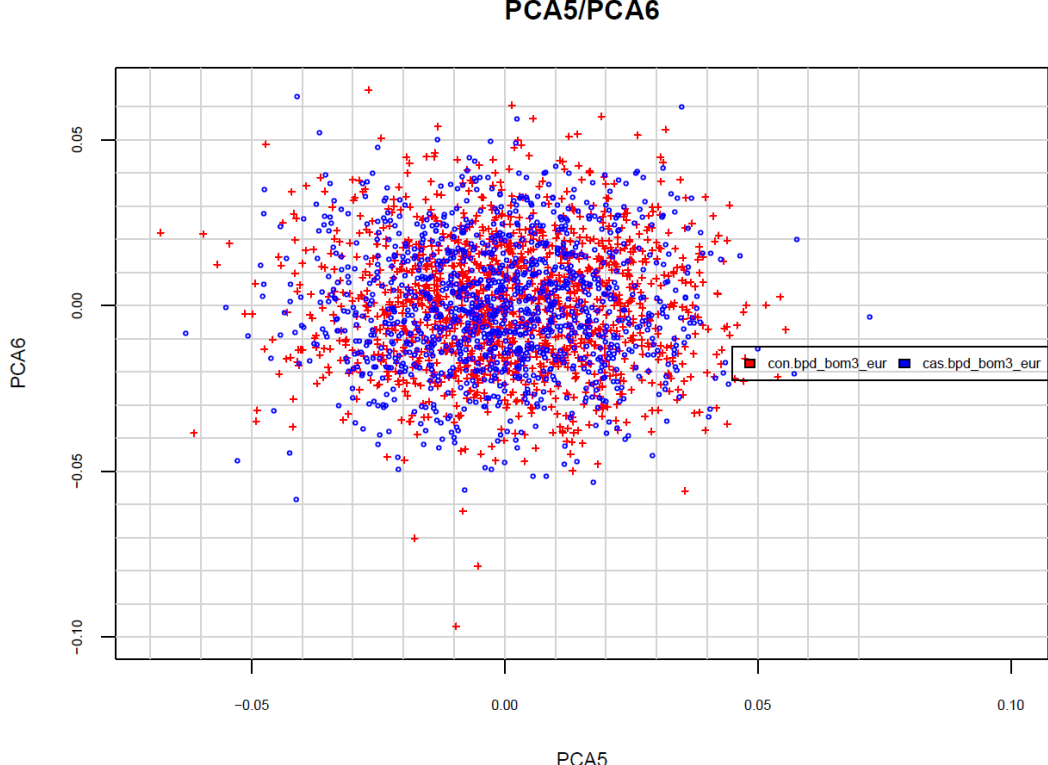
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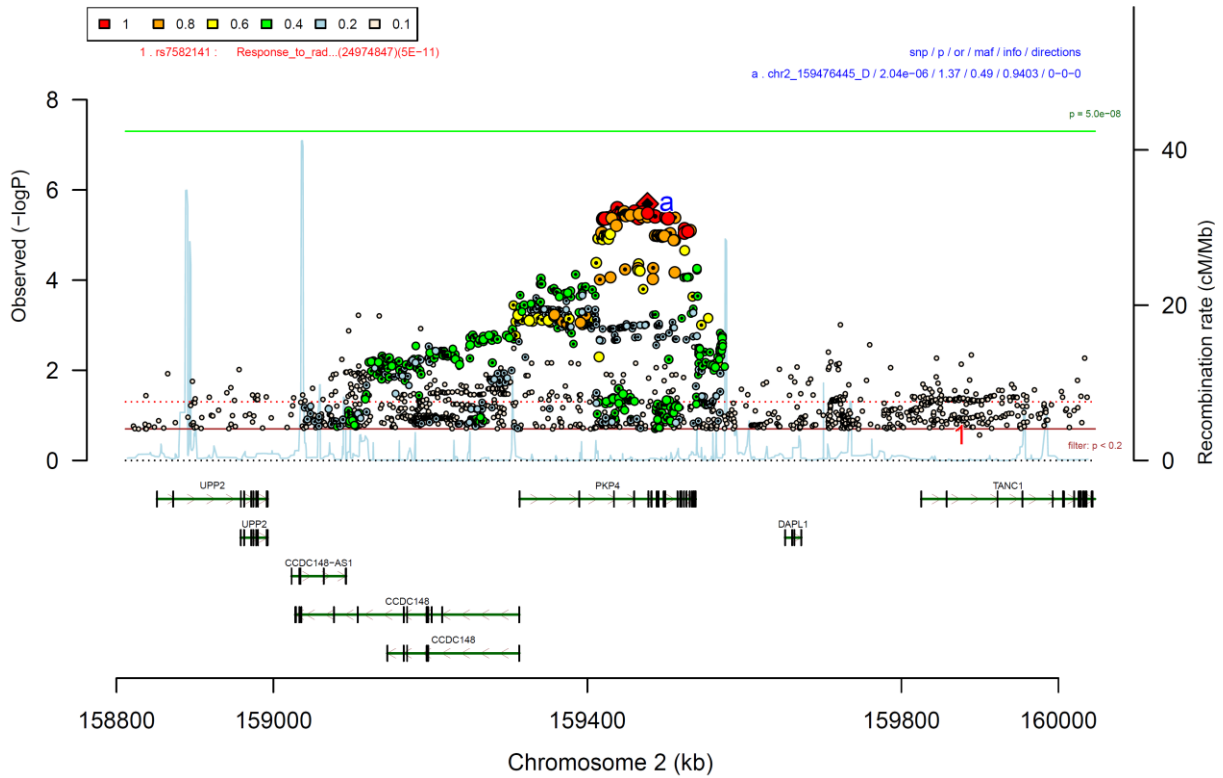
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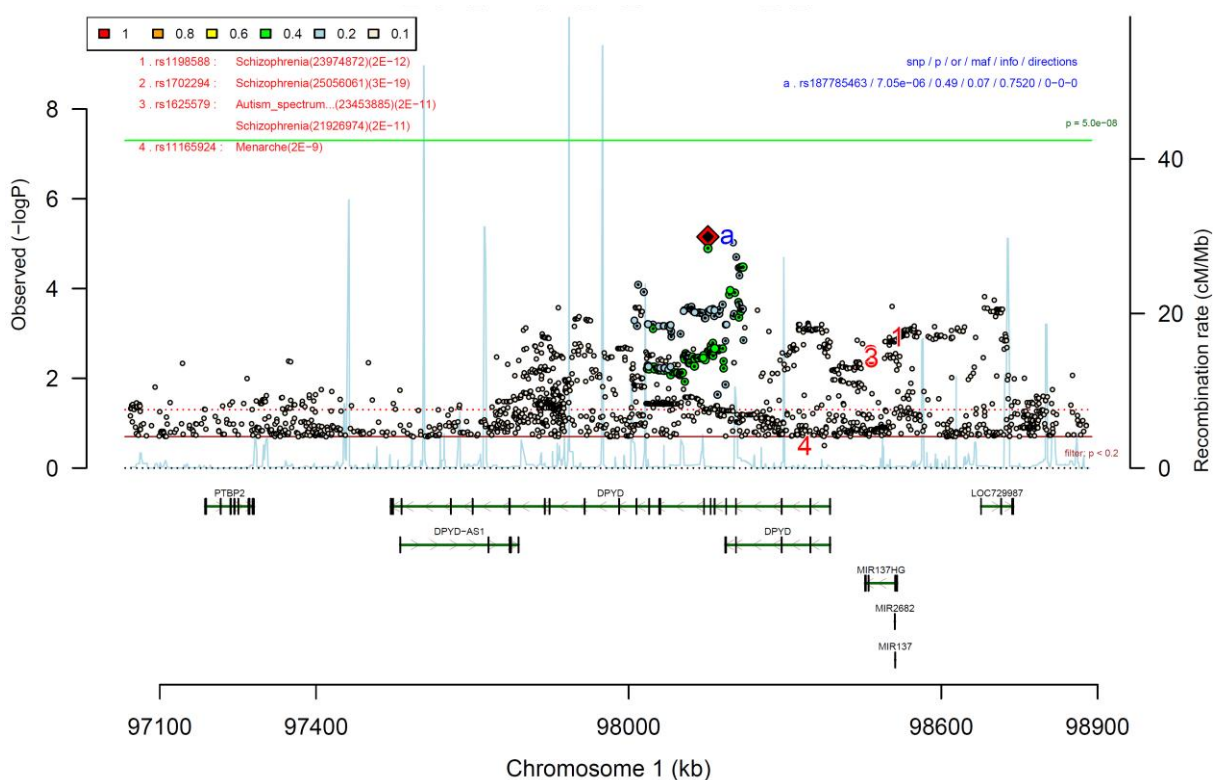
Supplementary Figure 6: Scatter plot of principal components 5 and 6



Supplementary Figure 7: Region plot of PKP4



Supplementary Figure 8: Region plot of DPYD



Tables

Supplementary Table 1: Demographic and clinical information on the final sample

	Cases	Controls
N	998	1,545
Sex (female/male)	914 / 84	868 / 677
Age in years (SD)	29.58 (8.64)	44.19 (13.24)
Comorbidity		
Depression		
(yes / no / missing)	666 / 262 / 40	-
Alcohol dependency		
(yes / no / missing)	163 / 781 / 54	-

Supplementary Table 2: Association results for single markers

Markers are reported with $p < 1 \times 10^{-5}$, and sorted according to chromosomal position. CHR = chromosome, BP = base pair position, A = allele, FRQ = frequency, SNP = single nucleotide polymorphism, OR = odds ratio, INFO = imputation info score, SE = standard error, GT = genotyped, IMP = imputed

CHR	SNP	BP	Gene	A1	A2	FRQ cases	FRQ controls	INFO	OR	SE	P	GT
1	rs187785463	98152125	DPYD	A	G	0.0531	0.0739	0.752	0.4935	0.1572	7.05E-06	IMP
1	rs6683957	98200719	DPYD	A	G	0.078	0.1128	1.002	0.6117	0.111	9.46E-06	IMP
2	rs57726666	16203326	GACAT3	A	G	0.9407	0.9125	0.912	1.8855	0.1341	2.24E-06	IMP
2	rs78068563	16208690	GACAT3	A	G	0.9397	0.9127	0.9227	1.8395	0.1324	4.19E-06	IMP
2	rs115689122	52606079	AC087073.1	A	T	0.016	0.0054	0.7809	6.1189	0.4062	8.20E-06	IMP
2	rs62127626	52719291	AC139712.4	A	C	0.0163	0.0057	0.7744	5.9639	0.3984	7.38E-06	IMP
2	rs150592717	89936117		T	C	0.8884	0.869	0.3247	2.2187	0.1796	9.14E-06	IMP
2	rs4664975	159418438	PKP4	A	C	0.4677	0.5152	0.8943	0.7399	0.0678	8.77E-06	IMP
2	rs3771608	159419739	PKP4	A	C	0.5405	0.4847	0.9831	1.345	0.0646	4.42E-06	IMP
2	rs12052933	159420140	PKP4	A	G	0.54	0.4841	0.9852	1.3451	0.0645	4.29E-06	IMP
2	rs10174340	159421412	PKP4	A	T	0.54	0.4841	0.9856	1.3451	0.0645	4.27E-06	IMP
2	rs3771609	159422239	PKP4	T	C	0.46	0.516	0.9859	0.7434	0.0645	4.25E-06	IMP
2	rs3771610	159422317	PKP4	A	G	0.54	0.484	0.9859	1.3451	0.0645	4.25E-06	IMP
2	rs10187426	159425104	PKP4	A	G	0.5399	0.4839	0.9869	1.3451	0.0644	4.21E-06	IMP
2	rs7577672	159429019	PKP4	A	G	0.4748	0.5324	0.9886	0.7523	0.0643	9.56E-06	IMP
2	rs3771614	159430579	PKP4	T	G	0.5318	0.4731	0.9904	1.3443	0.0643	4.26E-06	IMP
2	rs3771616	159430991	PKP4	A	G	0.5318	0.4731	0.9903	1.3444	0.0643	4.23E-06	IMP
2	rs3821291	159431031	PKP4	A	G	0.5318	0.4731	0.9903	1.3444	0.0643	4.23E-06	IMP

2	rs12473797	159431663	PKP4	T	C	0.5319	0.4732	0.9898	1.3449	0.0644	4.14E-06	IMP
2	rs3755408	159431842	PKP4	T	C	0.5318	0.4731	0.9903	1.3445	0.0643	4.21E-06	IMP
2	chr2_159432313_D	159432313	PKP4	I2	D	0.5496	0.4914	0.9516	1.3537	0.0658	4.13E-06	IMP
2	rs2356189	159434500	PKP4	A	G	0.5402	0.4839	0.9869	1.3482	0.0644	3.55E-06	IMP
2	rs3755413	159436702	PKP4	A	G	0.528	0.4703	0.9825	1.3387	0.0646	6.23E-06	IMP
2	rs3771620	159438129	PKP4	T	C	0.4591	0.5157	0.9808	0.7375	0.0647	2.52E-06	IMP
2	rs4664979	159439898	PKP4	T	C	0.4597	0.5161	0.9868	0.7414	0.0644	3.44E-06	IMP
2	rs3771627	159443266	PKP4	T	C	0.4596	0.5161	0.9865	0.741	0.0645	3.31E-06	IMP
2	rs2108215	159446818	PKP4	T	G	0.4674	0.5269	0.9909	0.7404	0.0643	3.01E-06	GT
2	rs11891131	159447509	PKP4	T	C	0.532	0.4731	0.9898	1.347	0.0644	3.71E-06	IMP
2	rs1465236	159448238	PKP4	A	G	0.4682	0.527	0.9894	0.7424	0.0644	3.71E-06	IMP
2	rs999232	159448306	PKP4	C	G	0.468	0.5269	0.9899	0.7422	0.0644	3.62E-06	IMP
2	rs3771631	159448926	PKP4	A	C	0.468	0.5269	0.9898	0.7422	0.0644	3.62E-06	IMP
2	rs3771632	159448946	PKP4	C	G	0.5341	0.4763	0.9806	1.3485	0.0646	3.75E-06	IMP
2	rs2051946	159454789	PKP4	T	G	0.532	0.4732	0.9886	1.3474	0.0644	3.66E-06	IMP
2	rs2051947	159459863	PKP4	A	G	0.5321	0.4733	0.9884	1.3472	0.0644	3.71E-06	IMP
2	rs12694965	159460218	PKP4	T	C	0.4594	0.5159	0.9836	0.7396	0.0646	3.00E-06	IMP
2	rs10191923	159460807	PKP4	T	C	0.5321	0.4735	0.9882	1.3467	0.0644	3.83E-06	IMP
2	rs10191934	159460832	PKP4	T	G	0.5322	0.4736	0.9877	1.3461	0.0644	3.97E-06	IMP
2	rs10191939	159460846	PKP4	A	T	0.4679	0.5267	0.9885	0.7423	0.0644	3.72E-06	IMP
2	chr2_159465312_I	159465312	PKP4	I5	D	0.4498	0.5053	0.9315	0.7371	0.0663	4.28E-06	IMP
2	rs3771643	159466149	PKP4	T	C	0.532	0.4731	0.9889	1.3483	0.0644	3.46E-06	IMP
2	rs3771647	159469420	PKP4	T	C	0.5321	0.4735	0.9893	1.3459	0.0644	3.93E-06	IMP
2	rs7607589	159469920	PKP4	T	C	0.4594	0.5159	0.9845	0.7404	0.0645	3.18E-06	GT

2	chr2_159476445_D	159476445	PKP4	I13	D	0.5489	0.4908	0.9403	1.3685	0.0661	2.04E-06	IMP
2	rs10180170	159476635	PKP4	T	C	0.532	0.4735	0.9892	1.3459	0.0644	3.95E-06	IMP
2	rs10180277	159476732	PKP4	T	C	0.5325	0.4735	0.99	1.3493	0.0644	3.23E-06	IMP
2	rs2883873	159486346	PKP4	T	C	0.4596	0.5149	0.9828	0.742	0.0646	3.84E-06	IMP
2	rs2528638	159500560	PKP4	A	G	0.4597	0.5145	0.98	0.7423	0.0647	4.08E-06	IMP
2	rs2711070	159502533	PKP4	C	G	0.4598	0.5145	0.9798	0.7428	0.0647	4.27E-06	IMP
2	rs929390	159503016	PKP4	A	G	0.4598	0.5145	0.9798	0.7428	0.0647	4.28E-06	IMP
2	rs2711068	159503757	PKP4	A	G	0.4561	0.5082	0.9498	0.7482	0.0657	9.98E-06	IMP
2	rs2711067	159503758	PKP4	C	G	0.4558	0.5082	0.9496	0.7476	0.0657	9.41E-06	IMP
2	chr2_159505404_D	159505404	PKP4	I7	D	0.5375	0.4842	0.9621	1.3353	0.0652	9.12E-06	IMP
2	rs2711062	159511527	PKP4	A	T	0.537	0.4807	0.967	1.3501	0.0652	4.09E-06	IMP
2	chr2_159523551_D	159523551	PKP4	I2	D	0.5395	0.4867	0.969	1.3392	0.0651	7.17E-06	IMP
2	rs2711072	159523803	PKP4	C	G	0.5399	0.4876	0.9688	1.335	0.0651	9.00E-06	IMP
2	rs2711073	159525516	PKP4	A	T	0.4611	0.5138	0.9713	0.7487	0.065	8.46E-06	IMP
2	rs2528590	159527845	PKP4	A	T	0.4586	0.5105	0.9641	0.7477	0.0653	8.33E-06	IMP
2	rs2528591	159527936	PKP4	T	C	0.5389	0.4862	0.9713	1.336	0.065	8.31E-06	IMP
2	rs2528592	159529272	PKP4	C	G	0.5391	0.4864	0.971	1.3359	0.065	8.43E-06	IMP
2	rs2711034	159530651	PKP4	A	T	0.5386	0.4858	0.9652	1.3379	0.0652	8.02E-06	IMP
3	chr3_77677315_D	77677315		I2	D	0.8496	0.9014	0.9652	0.6434	0.0991	8.57E-06	IMP
3	rs283386	115415834	GAP43	A	G	0.858	0.8958	0.7665	0.6098	0.1104	7.42E-06	IMP
4	rs57800857	140863365	MAML3	A	C	0.6855	0.6365	0.9212	1.3684	0.0707	9.16E-06	IMP
4	rs35319653	140903155	MAML3	T	C	0.3002	0.3514	0.9691	0.7328	0.0698	8.37E-06	IMP
4	chr4_140905595_I	140905595	MAML3	D	I3	0.6984	0.6467	0.9682	1.3655	0.0697	7.86E-06	IMP
4	rs12505942	140906390	MAML3	T	C	0.6996	0.6475	0.968	1.369	0.0698	6.84E-06	IMP

4	rs13136239	140908755	MAML3	A	G	0.3012	0.3534	0.9664	0.7293	0.0698	6.14E-06	IMP
4	chr4_140909205_I	140909205	MAML3	D	I9	0.686	0.6321	0.9323	1.3646	0.0703	9.75E-06	IMP
4	rs6823515	140925403	MAML3	C	G	0.3265	0.3825	0.9602	0.7372	0.0686	8.85E-06	IMP
5	rs34416917	36406618		A	G	0.1156	0.1561	0.8749	0.6316	0.1026	7.49E-06	IMP
5	rs35571570	36410724		T	C	0.1979	0.2522	0.9345	0.6981	0.0808	8.61E-06	IMP
5	rs10512650	36419793		T	C	0.1892	0.2459	0.943	0.6943	0.0816	7.89E-06	IMP
6	rs2145737	123879922	TRDN	T	C	0.6008	0.5422	0.9379	1.3526	0.0671	6.85E-06	IMP
6	rs9365376	158198680	SNX9	T	C	0.0693	0.1009	0.8188	0.5493	0.133	6.67E-06	IMP
6	rs6922614	158206364	SNX9	T	C	0.9205	0.888	0.8136	1.7794	0.1255	4.41E-06	IMP
7	rs6975373	4467479		T	C	0.1056	0.0793	0.8489	1.7113	0.1186	5.95E-06	IMP
7	rs75047832	4470076		C	G	0.1081	0.0814	0.8539	1.6776	0.1171	9.97E-06	IMP
7	rs12530503	4493125		A	C	0.9241	0.9495	0.9481	0.5499	0.1344	8.58E-06	IMP
7	rs114497090	19217524		A	G	0.0151	0.005	0.815	6.1683	0.4103	9.25E-06	IMP
7	chr7_27669577_D	27669577		I5	D	0.3572	0.2951	0.9038	1.3789	0.0718	7.68E-06	IMP
7	chr7_76213662_D	76213662		D	I4	0.229	0.189	0.8719	1.4694	0.084	4.63E-06	IMP
8	rs11784341	96957730	RP11-31K23.1	T	C	0.0245	0.0118	0.6765	4.2643	0.3038	1.81E-06	IMP
8	rs140121824	96958935	RP11-31K23.1	A	C	0.0245	0.0118	0.6765	4.2643	0.3038	1.81E-06	IMP
8	rs150918060	96966136		A	T	0.0243	0.0119	0.6873	4.0694	0.2994	2.77E-06	IMP
8	rs149724328	96977529		A	G	0.0239	0.012	0.7081	3.7974	0.293	5.27E-06	IMP
8	rs183820757	96983770		T	C	0.0242	0.0121	0.7254	3.5986	0.2882	8.85E-06	IMP
9	rs7859734	89562354	GAS1	T	C	0.3234	0.3899	0.9284	0.7309	0.0699	7.34E-06	IMP
10	rs184224980	12665138	CAMK1D	T	C	0.7877	0.8293	0.6115	0.6174	0.1039	3.44E-06	IMP
10	rs12768957	82187293	FAM213A	A	G	0.1559	0.1232	0.9723	1.5119	0.0928	8.44E-06	IMP
10	rs7074356	82191334	FAM213A	A	G	0.1586	0.1251	0.9643	1.5278	0.0926	4.75E-06	IMP

10	rs35828718	82193877		A	G	0.156	0.1237	0.9611	1.5096	0.0932	9.98E-06	IMP
12	rs113507694	7866609	DPPA3	A	G	0.9427	0.9654	0.591	0.3524	0.2006	2.01E-07	IMP
12	chr12_7867117_I	7867117	DPPA3	I2	D	0.1707	0.1402	0.6188	1.6577	0.111	5.27E-06	IMP
12	rs144017301	78915948	RP11-171L9.1	T	C	0.8577	0.8111	0.9081	1.5204	0.0918	5.03E-06	IMP
12	rs146457125	78916053	RP11-171L9.1	A	T	0.8575	0.8107	0.9091	1.5209	0.0917	4.84E-06	IMP
12	rs11110253	78918441	RP11-171L9.1	A	G	0.8705	0.8255	0.8742	1.5641	0.0973	4.32E-06	IMP
12	rs11110304	78928494		T	C	0.8494	0.8049	0.9059	1.4918	0.09	8.71E-06	IMP
13	rs675828	103350733	METTL21C	A	G	0.4471	0.5172	0.9842	0.7496	0.0644	7.63E-06	IMP
13	rs9520569	108402551	FAM155A	T	C	0.7808	0.7353	0.8278	1.4449	0.0824	7.86E-06	IMP
14	rs187058036	87892807	RP11-594C13.1	T	C	0.9611	0.9766	0.7328	0.3808	0.2159	7.74E-06	IMP
17	rs76695126	9426724	STX8	T	C	0.486	0.44	0.9092	1.3671	0.0674	3.47E-06	IMP
17	rs1114532	9434564	STX8	T	C	0.5205	0.565	0.9992	0.7517	0.0642	8.83E-06	GT
17	chr17_9437507_D	9437507	STX8	D	I3	0.4379	0.4772	0.804	0.7232	0.0719	6.54E-06	IMP
17	rs4791850	9439561	STX8	T	C	0.5222	0.5665	0.9862	0.7511	0.0647	9.57E-06	IMP
17	rs1985762	48404076		T	C	0.1225	0.0832	0.814	1.6842	0.1164	7.56E-06	IMP
19	rs12327596	54192903	MIR526B	A	G	0.5877	0.6468	0.9698	0.7412	0.0666	6.90E-06	IMP
19	rs8104156	54193170	MIR526B	T	G	0.3995	0.3394	0.9527	1.3601	0.0677	5.47E-06	IMP
19	rs1978714	54194677	MIR526B	T	C	0.5168	0.5755	0.9689	0.7479	0.0653	8.52E-06	IMP
19	rs7248039	54196520	MIR525	A	G	0.5185	0.577	0.9765	0.7495	0.065	9.32E-06	IMP
19	rs7251482	54196955	MIR525	A	G	0.5197	0.5789	0.9734	0.747	0.0652	7.65E-06	IMP
19	rs10413288	54197633	MIR525	A	G	0.5175	0.5767	0.9793	0.7481	0.0649	7.88E-06	GT
19	rs8100449	54199017	MIR525	T	C	0.5362	0.5955	0.94	0.7405	0.0665	6.25E-06	IMP
19	rs8099860	54199251	MIR525	T	C	0.4812	0.4218	0.9741	1.3388	0.0651	7.51E-06	IMP
19	rs1989486	54200619	MIR525	A	G	0.477	0.4181	0.9616	1.3469	0.0657	5.82E-06	IMP

19	rs10417113	54201088	MIR525	T	C	0.5184	0.5773	0.9732	0.7467	0.0652	7.39E-06	IMP
19	rs10417538	54201286	MIR525	T	C	0.5183	0.5773	0.9731	0.7464	0.0652	7.24E-06	IMP
19	chr19_54202088_I	54202088	RPS9	I5	D	0.4321	0.3786	0.836	1.3694	0.071	9.61E-06	IMP

Supplementary Table 3: Overview of Gene Set Excytosis (GO: 0006887)

SNP = single nucleotide polymorphism; BP = Base pair position; CHR = chromosome

<i>Significant Genes</i>	<i>SNP</i>	<i>BP</i>	<i>P-Value</i>	<i>CHR</i>	<i>Gene Start</i>	<i>Gene End</i>
YKT6	rs1004558	44240407	7,08E-05	7	44240567	44253893
NLGN1	rs9821957	173858380	4,30E-04	3	173114074	174004434
LIN7A	rs17662201	81220111	6,04E-04	12	81186299	81331704
RABEPK	rs393721	127985775	0,0045	9	127962821	127996437
RIMS1	rs6931045	72631747	0,0069	6	72596406	73112845
SYT1	rs10861133	79294033	0,0111	12	79257773	79845788
PKDREJ	rs41302599	46657308	0,0126	22	46651560	46659219
CADPS	rs11708441	62548081	0,0146	3	62384022	62861054
AKAP3	rs2074866	12680454	0,0151	7	4724674	4758213
SCIN	rs55903142	160514	0,0161	17	12610203	12693228
RPH3AL	rs1990313	4736086	0,0190	12	62293	236045
CPLX1	rs6816868	779670	0,0253	4	778745	819986
CCL8	rs8075846	32632483	0,0367	17	32646055	32648421
SEPT05	rs1557627	19677207	0,0380	22	19701987	19712295
RAB26	rs36232	2199788	0,0397	16	2190804	2204166
<i>Non-Significant Genes</i>						
SCRN1	rs1049394	29960804	0,0554	7	29959719	30029905
CPLX2	rs1544923	175268492	0,0563	5	175223313	175311023
ARFGEF2	rs1997850	47526360	0,1248	20	47538427	47653230
CCL5	rs1065341	34198593	0,1940	17	34198495	34207797
ARFGEF1	rs10957384	68219564	0,2926	8	68085747	68255912
VAMP3	rs2071987	7834026	0,4460	1	7831329	7841492
CCL3	rs9972960	34420079	0,5845	17	34415602	34417515
<i>Genes without mapped variants</i>						
SYTL4	No Variant					
VTI1B	No Variant					
SYTL4	No Variant					

Supplementary Table 4: Association of polygenic risk scores for Bipolar Disorder (BIP), Schizophrenia (SCZ), and Major Depressive Disorder (MDD) with Borderline Personality Disorder case control status

R² = Nagelkerke's R², N SNPs = Number of included SNPs

<i>P</i> -value threshold	<i>BIP</i>			<i>SCZ</i>			<i>MDD</i>		
	<i>N SNPs</i>	<i>R²</i>	<i>P</i>	<i>N SNPs</i>	<i>R²</i>	<i>P</i>	<i>N SNPs</i>	<i>R²</i>	<i>P</i>
5*10 ⁻⁸	17	6.09E-05	0.74481	108	0.00493	0.00337	0	-	-
0.000001	73	0.00031	0.46040	246	0.01009	2.70E-05	5	1.49E-05	0.87201
0.0001	556	0.00310	0.02005	1,278	0.02211	4.78E-10	125	0.00012	0.65288
0.001	2,180	0.00497	0.00324	3,419	0.02993	4.04E-13	784	0.00468	0.00431
0.01	9,408	0.00670	0.00063	10,649	0.02929	7.23E-13	5,010	0.01211	4.20E-06
0.05	27,346	0.00691	0.00051	24,741	0.02714	5.04E-12	16,687	0.02115	1.14E-09
0.1	43,365	0.00731	0.00036	35,710	0.02757	3.40E-12	27,188	0.01736	3.52E-08
0.2	67,753	0.00856	0.00011	51,621	0.02765	3.18E-12	43,315	0.01841	1.36E-08
0.5	116,624	0.00778	0.00023	80,828	0.02993	4.03E-13	73,493	0.01515	2.64E-07
1	154,455	0.00670	0.00063	101,718	0.03062	2.17E-13	94,078	0.01511	2.73E-07

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