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Original research

Whole-genome sequencing of monozygotic twins discordant for schizophrenia indicates multiple genetic risk factors for schizophrenia

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ABSTRACT

Schizophrenia is a common disorder with a high heritability, but its genetic architecture is still elusive. We implemented whole-genome sequencing (WGS) analysis of 8 families with monozygotic (MZ) twin pairs discordant for schizophrenia to assess potential association of *de novo* mutations (DNMs) or inherited variants with susceptibility to schizophrenia. Eight non-synonymous DNMs (including one splicing site) were identified and shared by twins, which were either located in previously reported schizophrenia risk genes (p.V24689I mutation in TTN, p.S2506T mutation in GCN1L1, IVS3+1G > T in *DOCK1*) or had a benign to damaging effect according to *in silico* prediction analysis. By searching the inherited rare damaging or loss-of-function (LOF) variants and common susceptible alleles from three classes of schizophrenia candidate genes, we were able to distill genetic alterations in several schizophrenia risk genes, including *GAD1*, *PLXNA2*, *RELN* and *FEZ1*. Four inherited copy number variations (CNVs; including a large deletion at 16p13.11) implicated for schizophrenia were identified in four families, respectively. Most of families carried both missense DNMs and inherited risk variants, which might suggest that DNMs, inherited rare damaging variants and common risk alleles together conferred to schizophrenia susceptibility. Our results support that schizophrenia is caused by a combination of multiple genetic factors, with each DNM/variant showing a relatively small effect size.

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1. Introduction

Schizophrenia is a severe chronic mental disorder affecting about 0.5% of the world populations (Saha et al., 2005). While it is believed that the etiology of schizophrenia is multifactorial, genetic epidemiological studies have indicated that schizophrenia is highly heritable (Sullivan et al., 2003). Genome-wide association studies (GWAS) have identified a number of common single nucleotide polymorphisms (SNPs) and copy number variations (CNVs) associated with schizophrenia (Schizophrenia Psychiatric Genome-Wide Association Study Consortium, 2011; Shi et al., 2011; Yue et al., 2011; Sullivan et al., 2012; Ma et al., 2013; Ripke et al., 2013; Wu et al., 2017). Although some CNVs were replicated and have not been found in mentally healthy cohort, they are not unique to schizophrenia (Stefansson et al., 2014). A recent large GWAS of schizophrenia reported 108 independent genetic variants that met the level of genome-wide significance (Schizophrenia Working Group of the Psychiatric Genomics Consortium, 2014). However, these common variants all together only explain a small portion of genetic risk. The “missing heritability” is still a preeminent case in schizophrenia (McClellan and King, 2010; Lee et al., 2012; Zuk et al., 2014).

Several studies had analyzed rare genetic variants or *de novo* mutations (DNMs) in schizophrenia through using either whole-genome sequencing (WGS) or whole-exome sequencing (WES) technology (Need et al., 2012; Fromer et al., 2014; Wang et al., 2015). While no specific genes associated with schizophrenia were identified, these studies demonstrated that patients with schizophrenia tend to have an enrichment of rare risk variants or DNMs in genes involved in synaptic function and in those encoding the activity-regulated cytoskeleton (ARC)-associated proteins and N-methyl-D-aspartate receptor (NMDAR) complexes, as well as fragile X mental retardation protein (FMRP) (Fromer et al., 2014). Mutations in the nongenic (Bae et al., 2014), noncoding RNA (Kwon et al., 2013), and large CNV regions (Liu et al., 2002; Xu et al., 2008; CNV and Schizophrenia Working Groups of the Psychiatric Genomics Consortium, 2017) may play important roles in the development of neuropsychiatric disorders such as schizophrenia. WGS is powerful for systematically evaluating various genetic risk factors such as common and rare variants, DNMs, and CNVs that may contribute to neuropsychiatric disorders (Jiang et al., 2013; Yuen et al., 2015). In this study, we performed WGS of 8 monozygotic (MZ) twin pairs discordant for schizophrenia and their parents to assess the potential role of DNMs or inherited genetic variants in liability of schizophrenia.

2. Results

2.1. Alignment and coverage statistics

WGS was performed at the BGI using the Illumina HiSeq platform with a depth of 30× (Table S1). Sequence calling was made for each individual using the Genome Analysis Toolkit (GATK) (McKenna et al., 2010). We detected an average of 3,141,201 single nucleotide variants (SNVs) (min = 3,096,496; max = 3,182,102; SD = 21,670) and 701,678 indels (min = 678,058; max = 720,000; SD = 11,501) per individual genome. Greater than 95% of these variations were SNPs in dbSNP142 (Bhagwat, 2010). Each sequenced individual had an average of 18,006 SNVs and 335 indels that were located in the coding region (Fig. S1A). Members of the 8 families (32 individuals) shared 735,818 SNVs and 169,308 indels, with no apparent family-specific pattern of SNVs and indels (Table S2; Fig. S1B and C). According to the minor allele frequency (MAF) in the 1000 Genomes Project (Sudmant et al., 2015), we detected around 6 million common SNVs (MAF > 5%), 94,344 low frequency SNVs (MAF = 0.5%–5%), and 73,453 rare SNVs (MAF < 0.5%) on average in each of the 32 individuals (Fig. S1D).

2.2. Detection and validation of DNMs

We applied forestDNM (Michaelson et al., 2012) to detect DNMs in trios (mother, father, and one offspring), combined the results of GATK (McKenna et al., 2010) and pindel (Ye et al., 2009) to find *de novo* indels, and employed four algorithms (Breakdancer (Chen et al., 2009), CNVnator (Abyzov et al., 2011), Delly (Rausch et al., 2012) and Lumpy (Layer et al., 2014)) to detect the *de novo* CNVs. Mutations shared by the MZ twins but not in their parents are counted as germline mutations. Mutations that were not shared by MZ twins are referred to as somatic mutations (Koren et al., 2012). While no *de novo* CNVs were detected, a total of 502 putative *de novo* SNVs (60.2 per individual) and 12 putative *de novo* indels (1.5 per individual) were detected in 8 MZ twin pairs (Fig. 1; Tables S3 and S4). There might be plausible mutation clusters across the genome, although our observation was based on a limited number of individuals (Fig. 1). We randomly selected 37 DNMs for validation on mother, father, and offspring using Sanger sequencing, of which 3 failed to be amplified, 20.6% (7/34) were false-positive variant calls, and 8.8% (3/34) were true-positive-inherited SNPs falsely called as negative in one parent (Table S5). The overall validation rate was comparable or higher than those of previous studies (Michaelson et al., 2012; Jiang et al., 2013; Genome of the Netherlands Consortium, 2014). We did not validate the 12 putative *de novo* indels, simply because they are all located in non-coding region, and the overall frequency of *de novo* indels in our sample was consistent with the previous study (Jiang et al., 2013).

2.3. DNMs and father's age at conception of child

We estimated the genome-wide mutation rate based on the germline DNMs per offspring (37–81 DNMs/offspring) as $1.18–2.58 \times 10^{-8}$ per generation, which is lower than the expected rate (Haldane, 2004). To gain more statistical power, we combined our data with others reported previously (Michaelson et al., 2012) to examine the association of the number of DNMs with parents' age at conception of child. We performed the correlation analyses of the number of DNMs in the offspring with maternal and paternal age at conception, respectively. Significant associations were observed with both maternal (p-value $= 2.60 \times 10^{-3}$) and paternal age (p-value $= 5.08 \times 10^{-5}$). Because the parent ages were highly correlated ($R^2 = 0.83$), we carried out a conditional analysis of multiple regression models to separate the age effect of the father and/or mother. We observed a significant positive correlation (p-value = 0.009) between father's age at conception of child and the number of DNMs in the offspring, whereas mother's age at conception (p-value = 0.976) was not significantly correlated with the number of DNMs (Fig. 2).

2.4. DNMs and potential pathogenicity

Nearly a half of DNMs were located in the intergenic region, only 8 DNMs (7 missense and 1 synonymous DNMs) shared between twins were located in the exon (Tables S4 and S6). In 8 probands, 4 (50%) were found to carry at least one *de novo* missense mutation. The *de novo* indels were located in the non-coding region. *In silico* functional analysis using both PolyPhen-2 (Adzhubei et al., 2010, 2013) and SIFT (Ng, 2003; Kumar et al., 2009) predicted that 4 of the 7 missense mutations would be deleterious (Table 1).

Approximately 63.3% (314/504 = 61.5%; excluding 10 unvalidated DNMs out of 37 randomly selected DNMs for validation) DNMs had been annotated with known and predicted regulatory

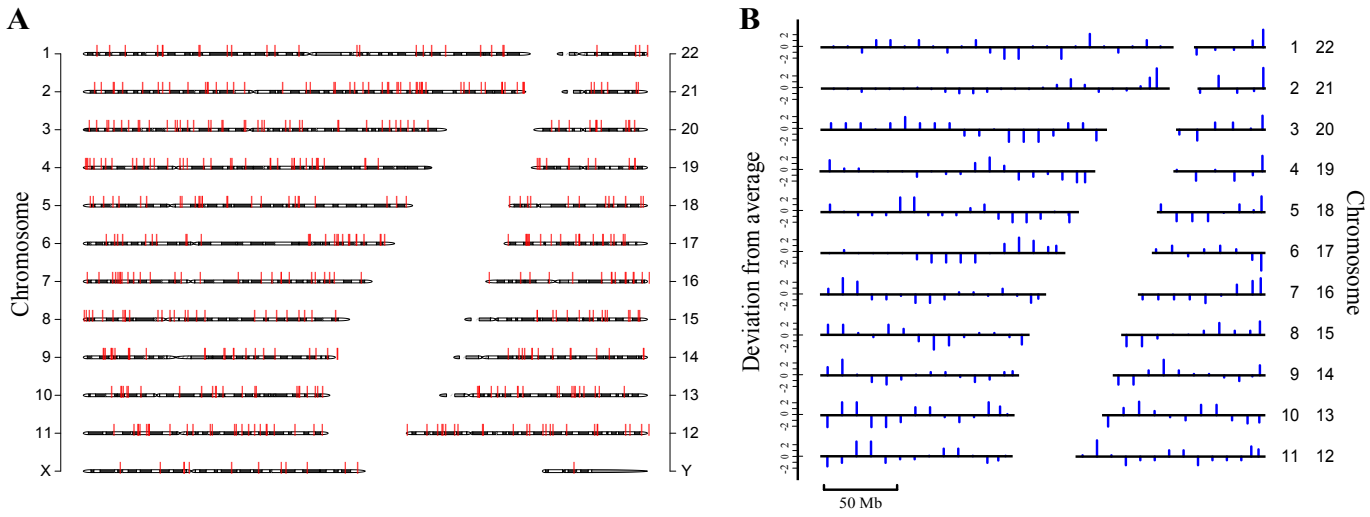

Fig. 1. The distribution of DNMs in the genomes of the 8 schizophrenia families. **A**: The location of DNMs in chromosomes 1–22, X and Y. The DNMs are marked with red "|". **B**: The distribution difference of DNMs located on different regions. The distribution difference is represented by the deviation from the average which was calculated by subtracting the average number of DNMs in 40 Mb genomic region from the average number of DNMs in whole chromosome, with a sliding window of 20 Mb genomic region for counting.

elements by RegulomeDB (Boyle et al., 2012), including DNAase hypersensitivity sites, binding sites of transcription factors, and promoter regions (Table S7). Approximately two-thirds of these DNMs (334/504 = 66.3%) are transitions. DNMs are located in CpG sites, including 4 in the CpG islands. Because all these DNMs are rare mutations, we could not retrieve the related information, such as expression quantitative trait loci (eQTL) (Yang et al., 2010). We arbitrarily classified these non-coding DNMs into different categories according to the location of each mutation in predicted regulatory elements of known schizophrenia susceptibility genes (3 classes of gene sets (see Methods and materials)), genes that were not reported to be associated with schizophrenia, and the intergenic regions. There was no apparent enriched pattern for more non-coding DNMs in regulatory elements in known schizophrenia genes (p-value > 0.01 for all three comparisons, fisher's exact test).

Most of the identified DNMs (92.1%; 464/504; excluding 10 unvalidated DNMs out of 37 randomly selected DNMs for validation) were shared by the paired twins, whereas only 40 DNMs were detected in one sibling of the twin, including 20 DNMs in the probands and 20 in the non-affected siblings. Among the 20 DNMs in the probands, 13 DNMs were located in the intron region, 1 DNM was located in the 5′-upstream region, and the remaining 6 DNMs were located in the intergenic region (Table 2). It is noteworthy that the majority of DNMs in the probands seemed to be enriched in the probands of 3 families (B84, 7 DNMs; B113, 5 DNMs; B124, 3 DNMs) (Table 2). Although these DNMs were not located in the coding region, we could not exclude the possibility that they had roles in gene regulation, as accumulating evidence showing a regulatory effect of mutations/variants in the non-coding region (Ward and Kellis, 2012; Roussos et al., 2014; Xiao et al., 2017). One intronic DNM in proband of B8 family was located in the intron of *NXPH1* (neurexophilin 1) – a gene located in a region with an autism risk CNV (Gai et al., 2012). The NXPH1 is a ligand for the NRXN1 protein, which has been reported to be involved in schizophrenia (Missler et al., 1998; Gauthier et al., 2011). The *NRXN1* gene had a significantly differential expression in schizophrenia patients compared with controls (Chen et al., 2013; Lanz et al., 2015) (Table 2).

DNMs shared by twins might also affect schizophrenia in the analyzed families. We focused on the 8 DNMs that were shared by the twins and would lead to residue changes or intron retention.

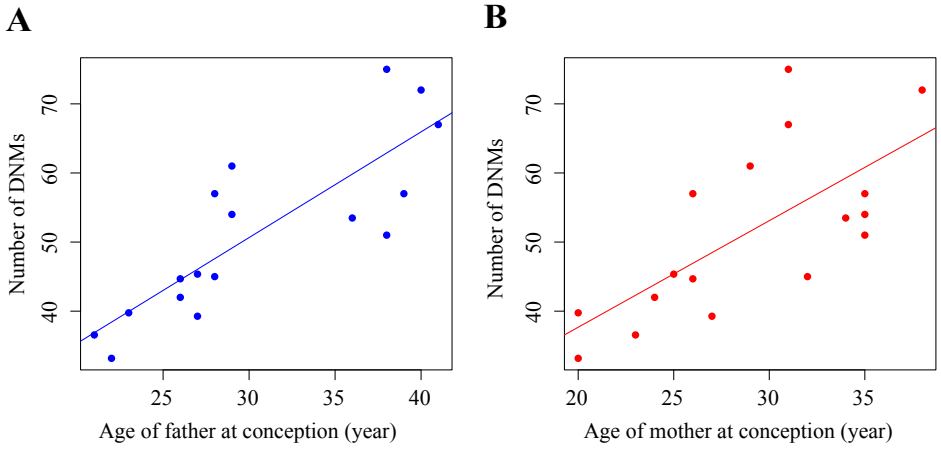

Fig. 2. Correlation between the number of DNMs and parents' age at conception. The number of DNMs in the offspring is plotted against the age of the father (**A**) and the mother (**B**). The number of mutations increases with the age of father at conception (p-value = 0.009). The data reported by Michaelson et al. (2012) were (re-)analyzed together with our data.

Table 1
The *de novo* missense and splice-site mutations in 8 pairs of MZ twins.

Sample	Location	Genotype	Gene	Transcript ID	Mutant	SIFT prediction	PolyPhen-2 prediction
B53 B54	chr12:120569035	C/G	*GCN1L1*	NM_006836	p.S2506T	Tolerated	Benign
B53 B54	chr17:39595049	G/T	*KRT38*	NM_006771	p.P265H	Damaging	Damaging
B53 B54	chr2:179436794	C/T	*TTN*	NM_001256850	p.V24689I	NA	NA
B62 B63	chr2:242283282	A/G	*SEPT2*	NM_001008491	p.N271S	Tolerated	Benign
B93 B94	chr19:57037004	C/G	*ZNF471*	NM_020813	p.A523G	Damaging	Damaging
B93 B94	chr16:2147410	G/A	*PKD1*	NM_000296	p.R3438W	Damaging	Damaging
B103 B104	chr10:128776253	G/T	*DOCK1*	NM_001290223	IVS3+1G > T	NA	NA
B123 B124	chr19:35449194	T/C	*ZNF792*	NM_175872	p.N522S	Tolerated	Damaging

The potential deleterious effect of each variant was predicted by using SIFT (Ng, 2003; Kumar et al., 2009) and PolyPhen-2 (Adzhubei et al., 2010, 2013). NA, not available. Tolerated, benign and damaging were defined according to SIFT scores (>0.05, tolerated; < 0.05, damaging) and PolyPhen-2 HDIV scores (>0.453, damaging; ≤ 0.452, benign).

Table 2
The *de novo* mutations in the probands and differential expression of the hit genes.

Sample	Location	Genotype	Gene and function	Differential expression (*p*-value)					RegulomeDB score[c]
				GSE53987[a]			GSE35978[b]		
				Hippocampus	Prefrontal cortex	Striatum	Cerebellum	Prefrontal cortex	
B104	chr2:153664053	G/A	Intergenic	NA	NA	NA	NA	NA	7
B113	chr4:122576011	T/C	Intergenic	NA	NA	NA	NA	NA	4
B113	chr20:52177664	A/G	RP4_724E16.2 intron	NA	NA	NA	NA	NA	6
B113	chr15:96155017	C/A	Intergenic	NA	NA	NA	NA	NA	6
B113	chr9:127918364	G/A	*PPP6C* intron	6.2E-06	0.057	0.007	0.75	0.007	6
B113	chr12:26742696	A/G	*ITPR2* intron	0.498	0.002	0.9	0.035	0.961	7
B124	chr1:230530315	T/C	*PGBD5* intron	0.465	0.231	0.003	0.647	7.7E-05	2b
B124	chr17:12638660	T/C	*MYOCD* intron	0.888	0.958	0.969	0.6826817	0.127	5
B124	chr17:12638661	A/G	*MYOCD* intron	0.888	0.958	0.969	0.6826817	0.127	5
B54	chr12:76731688	G/T	Intergenic	NA	NA	NA	NA	NA	4
B63	chr4:1331141	T/C	*MAEA* intron	2.3E-05	0.446	0.069	0.383	0.001	4
B84	chr17:41382052	T/C	LINC00854 5′-upstream	NA	NA	NA	NA	NA	2b
B84	chr13:45830708	C/T	*GTF2F2* intron	0.002	0.698	0.273	0.672	0.159	6
B84	chr9:94497890	T/C	*ROR2* intron	0.012	0.617	0.843	0.9733785	0.00468	6
B84	chr7:8681356	T/C	*NXPH1* intron	2.2E-06	0.865	0.007	0.086	0.00034	7
B84	chr4:36920104	A/C	Intergenic	NA	NA	NA	NA	NA	7
B84	chr6:149959476	T/A	*KATNA1* intron	0.73	0.766	0.995	0.4299414	0.358	7
B84	chr2:167878685	A/G	*XIRP2* intron	0.036	0.854	0.298	0.6894866	0.643	7
LJ	chr6:12749681	C/G	*PHACTR1* intron	0.008	0.939	0.043	0.4530216	0.0263	3a
LJ	chr19:3354318	T/C	Intergenic	NA	NA	NA	NA	NA	7

[a] Data from Lanz et al. (2015).
[b] Data from Chen et al. (2013).
[c] The RegulomeDB (Boyle et al., 2012) score refers to the following available datatypes for a single coordinate. 2b, transcription factor (TF) binding + any motif + DNase Footprint + DNase peak; 3a , TF binding + any motif + DNase peak; 4, TF binding + DNase peak; 5, TF binding or DNase peak; 6, other; 7, no data. NA, not avaliable.

Among the 7 shared non-synonymous mutations (Table 1), three (p.P265H in KRT38; p.A523G in ZNF471 and p.R3438W in PKD1) were predicted to be damaging according to the overlapping results of multiple *in silico* program affiliated predictions. We searched the 7 genes harboring the above non-synonymous mutations (Table 1) in the NPdenovo database (Li et al., 2016) to detect whether these genes had DNMs in schizophrenia or other neurodevelopmental disorders. We only found that the *TTN* gene contained one DNM according to database search, but the DNM is different from the one observed in this study. We observed a similar pattern of family-specific enrichment of shared missense DNMs as described above for DNMs in the probands, particularly for families B5 and B9 (Table 1). It is worth noting that the *TTN* (titin; p.V24689I) and *GCN1L1* (GCN1 eIF2 alpha kinase activator homolog; p.S2506T) genes identified in family B5 also exhibited *de novo* non-synonymous mutations in schizophrenia patients (Fromer et al., 2014; Wang et al., 2015) and autism patients (Iossifov et al., 2012; Fromer et al., 2014), but the mutations are different from this study. Using the DAPPLE web server (Rossin et al., 2011), we explored protein-protein interactions (PPIs) among 23 genes hit by DNMs in the coding regions, UTR regions, 3′-downstream regions or 5′-upstream regions, and the schizophrenia top genes (identified by convergent functional genomics (CFG) score) of the SZDB data set (Wu et al., 2017) (Fig. S2). We found that TTN and GCN1L1 proteins interacted with a variety of schizophrenia-related proteins, such as DISC1 (disrupted in schizophrenia 1) (Hodgkinson et al., 2004; Thomson et al., 2014; Huang, 2015) and AKT1 (encoding serine-threonine protein kinase) (Emamian et al., 2004; Schwab et al., 2005). However, we need to state that the exact role of these two genes in neurodevelopment of schizophrenia remains to be clarified.

The splice site mutation (IVS3+1G > T) in the *DOCK1* (dedicator of cytokinesis 1) gene shared by the twins from B10 family would lead to an intron retention, which might cause the truncated protein (remaining only 57 of 1886 (3.0%) amino acid residues) to be non-functional. In the nervous system, the protein (Dock180) of the *DOCK1* gene was essentially involved in the regulation of axon guidance and dendritic spine morphogenesis (Li et al., 2008). The brain-specific angiogenesis inhibitor-1 (BAI-1), an interacting protein of Dock180-ELMO module, had been linked to a variety of neuropsychiatric disorders, including schizophrenia, bipolar disorder, and drug addiction (Park et al., 2007; Lanoue et al., 2013). The Dock proteins have been well recognized to be related to neurological diseases (Shi, 2013).

2.5. Inherited SNPs and schizophrenia

We performed a gene-based association analysis on the basis of all variants in 32 individuals from the 8 families that were scored relative to human reference genome (hg19; http://genome.ucsc.edu/). As our sample size was relatively small and had limited statistical power, we therefore chose an arbitrary threshold (p-value < 0.01) in order to sample more risk genes even there is a possibility of false positive hit. A total of 55 genes were captured by our analysis (p-value < 0.01) (Table S8). Among the most significant genes (p-value = 9.99×10^{-4}), *KCNQ2* encodes a subunit of KCNQ which is a voltage-gated K^+ channel regulating the excitability of neurons (Brown et al., 2007) and variants in this gene had been reported to be associated with schizophrenia (Lee et al., 2013). We further screened the recently reported 128 susceptibility SNPs that met the genome-wide significance (p-value $\leq 5 \times 10^{-8}$) (Schizophrenia Working Group of the Psychiatric Genomics Consortium, 2014) in our samples. A total of 607 hits from 8 families were identified. The polygenic risk scores based on the 607 hits showed that probands in general had substantially higher score than their mothers (p-value = 0.008, R^2 = 0.583), fathers (p-value = 0.353, R^2 = 0.022), and parents (average, p-value = 0.021, R^2 = 0.468). Because these SNPs used for the calculation of polygenic risk score were all common inherited SNPs and were shared by MZ twins, and the DNMs in each twin provided no additional information for the calculation, we could not distinguish the polygenic risk score between the proband and his/her sibling (Table S9 and Fig. 3). Furthermore, we focused on those risk alleles that were homozygous in the offspring but heterozygous in their parents, which would increase the risk of schizophrenia in the offspring. As a result, we identified 38 such variants. Eight of these 38 risk alleles occurred in multiple families (Table S10). 8 families all harbored at least 3 homozygous risk alleles, with the highest number in families B5 and B10 (each had 7). One of the homozygous risk variants in B8 family, rs2007044, influenced the expression of *CACNA1C* (encoding the α-1C subunit of the L-type voltage-gated calcium channel) according to the eQTL analysis (Yang et al., 2010). As an important schizophrenia risk gene, *CACNA1C* has been reported to be involved in neuronal development (Green et al., 2010; Nyegaard et al., 2010). Another homozygous risk variant in B10 family, rs10503253 (located in *CSMD1*), was associated with schizophrenia in several previous reports (Donohoe et al., 2013; Rose et al., 2013; Koiliari et al., 2014). Note that in LI family, we also detected a DNM that was located in the intron region of the *CSMD1* gene (Table S4).

Rare genetic variants with a large effect were thought to play a substantial role in causing schizophrenia (McClellan et al., 2007). We detected all the rare (MAF ≤ 0.01 in the 1000 Genomes Project (Sudmant et al., 2015)), inherited LOF (i.e., stop-gain or frameshift) and damaging missense variants (as predicted by at least two of the five prediction algorithms) in our samples. In total, 9480 rare damaging or LOF variants were found, with an average of 296 variants per individual (Table S11). However, we found no significant p-value for the gene-based rare variant burden test based on all rare damaging or LOF variants in all samples. It was noteworthy that *LAMC1* (laminin subunit gamma 1), with 5 rare damaging variants in 5 families (Table S12), had a marginal significance (p-value = 0.068). We further focused on these schizophrenia associated genes carrying the rare damaging or LOF variants by referring to 3 classes of gene sets (Materials and methods). In total, we identified 5 Class I genes, 17 Class II genes, and 45 Class III genes containing rare damaging or LOF variants (Table S13).

Among the 38 Class I genes, *RELN* (reelin), *GAD1*, *PLXNA2*, *MTHFR*, and *FEZ1* had rare damaging or LOF variants. The *RELN* gene encodes an extracellular matrix protein essential for cell positioning and neuronal migration during brain development (Tissir and Goffinet, 2003), which was previously reported to be involved in schizophrenia (Costa et al., 2002; Shifman et al., 2008), autism (Persico et al., 2001) and Alzheimer's disease (Seripa et al., 2008). The RELN variant p.M927V might contribute to schizophrenia-risk in family B11. The variant p.D180V in FEZ1 (fasciculation and elongation protein zeta-1) was found in LI family. FEZ1 was involved in nerve growth and fasciculation; together with DISC1, it regulates neuronal development that is related to schizophrenia (Kang et al., 2011). Genetic variants in *FEZ1* had been reported to be associated with schizophrenia in Japanese cohort (Yamada et al., 2004). B5 family had 2 Class I genes with rare damaging mutation: *GAD1* (glutamate decarboxylase 1) and *PLXNA2* (plexin A2). Previous study showed that schizophrenic brain had a decreased expression of the gamma-aminobutyric acid (GABA) synthetic enzyme glutamic acid decarboxylase 67 (GAD67), which is encoded by *GAD1* (Addington et al., 2005; Straub et al., 2007). PLXNA2 is a member of the semaphorin receptor family and plays a role in the development of axonal projections and in neural regeneration (Winberg et al., 1998). Co-expression network analysis showed that PLXNA2 and GAD1 had a similar expression pattern and potential interaction, suggesting a potential combined effect on schizophrenia (Fig. S3).

Among the 206 Class II genes, 2 genes (*GSN* and *ITPR1*) had more than one damaging non-synonymous variant. GSN variants p.K546R and p.R144Q were found in families B12 and B8, respectively. The *GSN* gene was found to be significantly down-regulated in a genome-wide expression analysis in a set of schizophrenia patients (Hakak et al., 2001). ITPR1 played a key role in synaptic plasticity by binding with disrupted-in-schizophrenia 1 (DISC1) (Tsuboi et al., 2015), a promising susceptibility factor for schizophrenia (Brandon and Sawa, 2011). The two missense variants p.I2030M (family B5) and p.I1421V (family LI) in ITPR1 were predicted to be damaging. It is plausible that these variants enacted an effect via altered binding activity with DISC1. Intriguingly, we found

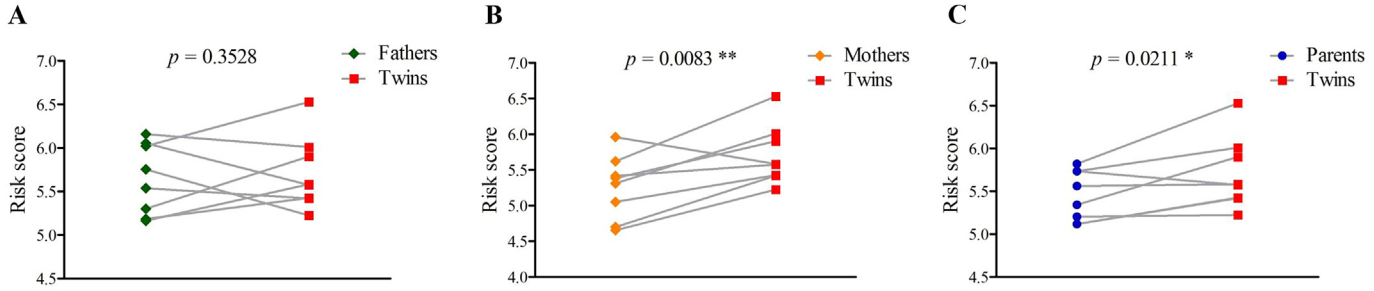

Fig. 3. Polygenic risk score based on 128 schizophrenia-risk SNPs in the comparison of probands and healthy parents. Comparison of the risk score between probands and their fathers (**A**), mothers (**B**) and parents (**C**) was evaluated by using paired t-test. *, $p < 0.05$; **, $p < 0.01$.

that families B5 and B8 appeared to have an enrichment of damaging missense variants (each had 5 variants) in Class II genes, and followed by families B9 and B11 (each had 2 variants) (Table S13).

We found 5 Class III genes that were shared by two families (Table S13). One of these genes was protocadherin alpha 6 (*PCDHA6*), which encodes a potential calcium-dependent cell-adhesion protein and is involved in the establishment and maintenance of specific neuronal connections in the brain (Wu and Maniatis, 1999). The interacting protein of PCDHA6, RELN, was reported as an important risk gene for schizophrenia (Costa et al., 2002; Shifman et al., 2008).

The enriched rare damaging or LOF variants in schizophrenia-related gene sets may significantly increase the risk of schizophrenia (Fromer et al., 2014). We compared the number of rare damaging variants or LOF variants in 17 core gene sets in parents and offspring (Table S14). Although we did not find a significant *p*-value for the odd ratio (OR) of rare damaging or LOF variants in these gene sets in offspring relative to their parents, partially due to the small sample size, the offspring did show a higher risk than parents in several gene sets (OR > 1.4) (Table S14). In the gene set of the NMDAR network, the offspring had 14 rare damaging variants whereas the parents only had 10 (OR = 1.40; *p*-value = 0.423). The NMDA signaling triggers multiple processes required for inducing synaptic plasticity (Malenka and Nicoll, 1993), and NMDA receptor hypofunction may contribute to the symptomatic features of schizophrenia (Goff and Coyle, 2001).

2.6. Inherited CNVs and schizophrenia

Combining the results from four algorithms (Breakdancer (Chen et al., 2009), CNVnator (Abyzov et al., 2011), Delly (Rausch et al., 2012) and Lumpy (Layer et al., 2014)), we detected 6619 CNVs (length > 1000 bp) in all probands and their parents, with an average of 276 variants per individual (Table S15). There were 4 inherited CNVs (both shared by twin pairs) in the previously reported schizophrenia-risk regions (Table S16) according to the data set that was based on 32 related studies ((Luo et al., 2014) and references therein). The proband from B11 family had a large inherited deletion (16p13.11; 2.9 Mb) contained 10 genes (*MPV17L*, *C16orf45*, *KIAA0430*, *NDE1*, *MYH11*, *C16orf63*, *ABCC1*, *ABCC6*, *NOMO3* and *XYLT1*) (Fig. S4). The *NDE1* (nuclear distribution gene E homolog 1) gene was involved in brain development, neuronal proliferation, migration and synapse formation (Shu et al., 2004). This gene was significantly associated with schizophrenia and had a biologically interaction with DISC1 and LISI proteins (Burdick et al., 2008; Bradshaw et al., 2011). Moreover, *NDE1*, *MYH11* and *ABCC1* were identified as top schizophrenia candidate genes by using a cumulative scoring to systematically prioritized genes affected by CNVs in schizophrenia (Luo et al., 2014).

3. Discussion

Multiple lines of evidence demonstrated that schizophrenia is a common disease with a complex etiology, involving heterogeneous genetic factors (Saha et al., 2005; Wu et al., 2017). We used WGS as a tool to identify genetic mutations/variants in 8 families with one MZ twin pair discordant for schizophrenia, to provide a comprehensive profile for genetic mutations and variants that might contribute to schizophrenia risk. Theoretically, the choice of identical twins discordant for schizophrenia would offer an extremely rare and valuable opportunity to pinpoint the pathogenic mutation(s) causing schizophrenia. We identified 504 DNMs in offspring and observed a significant positive correlation between father's age at conception of child and the number of DNMs in the offspring, whereas mother's age at conception was not significant. This finding is consistent with the result of previous studies (Kong et al., 2012; Michaelson et al., 2012; Genome of the Netherlands Consortium, 2014), and could explain the epidemiological observation that the risk of schizophrenia increased significantly with father's age at conception of child in Iceland (Malaspina, 2001).

To pinpoint the potential genetic alterations that would account for schizophrenia in the twin discordant for disease, we focused on DNMs, inherited common risk SNPs, inherited rare damaging or LOF variants in the family members. Among the identified DNMs only in probands, none of them were located in coding region. Nevertheless, whether these DNMs influence the discordance of schizophrenia remains to be elucidated. The missense DNMs shared by twins might also increase the risk of schizophrenia, such as p.V24689I in TTN, p.S2506T in GCN1L1 and IVS3+1G > T in *DOCK1*, to name a few, and functional assays should be performed to validate our speculation.

Previous studies found no discordant DNA variants in MZ twins with discordant disease (Baranzini et al., 2010; Chaiyasap et al., 2014; Petersen et al., 2014), suggesting that DNMs might partially account for disease, together with other genetic factors. All these variants with potentially small effect sizes were combined into one big effect network, rather than few independent variants, to increase the inherited risk of common diseases such as schizophrenia (International Schizophrenia et al., 2009; Kraft and Hunter, 2009). Presently, there were no single mutation that could have enough effect to cause schizophrenia (McCarthy et al., 2008). Given the characteristics of pathogenic complexity and high heritability in schizophrenia, we also analyzed the effect of inherited variants on schizophrenia. The gene-based burden analysis identified many genes that might increase the risk of schizophrenia, such as *KCNQ2*. We also found 38 common inherited variants that were homozygous in offspring but heterozygous in their parents, and these variants might be functional according to data-mining of available datasets. For instance, SNP rs2007044 from B8 family could change the expression of *CACNA1C* during the neuronal development (Green et al., 2010; Nyegaard et al., 2010), and SNP rs10503253 in *CSMD1* from B10 family was associated with schizophrenia (Donohoe et al., 2013; Rose et al., 2013; Koiliari et al., 2014). Further analysis of polygenic risk score based on the 128 schizophrenia susceptible SNPs (Schizophrenia Working Group of the Psychiatric Genomics, 2014) indicated that probands had a higher risk for schizophrenia than their parents.

Rare genetic variants, as well as common variants, are thought to contribute to schizophrenia, but rare variants may exert a larger effect than common variants (McClellan et al., 2007; Sebat et al., 2009). By searching the rare damaging variants or LOF variants from three classes of schizophrenia-related genes, we found some important genetic alterations in these schizophrenia-related genes that might be linked to the onset of schizophrenia, such as *GAD1*, *PLXNA2*, *RELN* and *FEZ1*. Moreover, multiple genetic factors, including DNMs, common inherited variants, rare inherited damaging variants and LOF variants, might increase risk for schizophrenia in these families (Fig. 4). For instance, B5 family not only had two missense DNMs in *TTN* and *GCN1L1*, but also had rare damaging variants in *GAD1* and *PLXNA2*, which suggested a combined effect of DNMs and inherited rare damaging variants on schizophrenia. It is noteworthy that B8 and B12 families shared two schizophrenia-associated genes which had rare damaging variants, *GSN* (p.R144Q mutation in B8 family, and p.K546R mutation in B12 family) and *PCDHA6* (p.N403T mutation in B8 and B12 families). These results indicated that the two families might have been subjected to similar genetic risk factors of schizophrenia.

Furthermore, we systematically investigated the interaction network among proteins encoded by genes harboring *de novo*

missense mutations and inherited highly risky or rare damaging mutations mentioned above (Tables 1, S10, S13 and S16). We found that all genes contained in the GeneMANIA database (Montojo et al., 2014) would constitute a network (Fig. S5). This result suggested these genes are involved in one shared interaction network to increase risk for schizophrenia with small additive effects.

The current study had several limitations. First, we collected only 8 families with MZ twin pairs discordant for schizophrenia. The sample size might have limited statistical power in the context of genome-wide sequence data, but this constitutes the largest sample for WGS of MZ twin pairs discordant for schizophrenia. Second, we analyzed the genetic alterations in blood samples, but we could not exclude a possibility of *in situ* DNMs in the schizophrenia-associated brain regions. Moreover, gene expression and epigenetic changes in these families were not analyzed, which had been reported to be an important factor in the pathogenesis of psychiatric disorders (Roth et al., 2009; Dempster et al., 2011; Zong et al., 2015). Finally, we did not perform functional assays to elucidate the biological implications of these missense DNMs and to validate the combined effect of estimated polygenic factors (Li et al., 2017b).

In short, we analyzed the potential risk factors of schizophrenia by using WGS data in 8 families with one MZ twin pair showing a discordance of schizophrenia. We did not identify any specific genes that played a prevalent role in schizophrenia, but we found a potentially combined effect of many genetic alterations, including DNMs, common inherited variants, rare inherited damaging variants and LOF variants, on schizophrenia. These results confirmed the heterogeneous nature of schizophrenia and might have important implications for clinical diagnosis of patients with schizophrenia. When investigating the genetics of schizophrenia, we need to search for rare and common variants, CNVs, and DNMs.

4. Materials and methods

4.1. Sample collection

Eight families of MZ twins discordant for schizophrenia were recruited in this study. These samples were analyzed for mtDNA heteroplasmy in our recent study (Li et al., 2017a). The schizophrenia patients were diagnosed independently by two psychiatrists following the criterion of Diagnostic and Statistical Manual of Mental Disorders, Fourth Edition (DSM-IV). Informed consents conforming to the tenets of the Declaration of Helsinki were obtained from each participant or guardian prior to this study. The institutional review board of the Second Xiangya Hospital approved this study.

4.2. Whole genome sequencing

Genomic DNAs from 8 identical twin pairs and their parents were isolated from the peripheral blood by using AxyPrep™ Blood Genomic DNA Miniprep Kit (Axygen, USA) according to the manufacturer's instruction. Deep (~30×) whole genome sequencing was performed at the BGI in Shenzhen using the Illumina HiSeq 2000 Platform (500 bp library, 90 bp reads). One individual (B114) of family B11 was sequenced at the Novogene corporation in Beijing using Illumina HiSeq X Ten Platform (500 bp library, 150 bp reads).

4.3. Alignment and variant calling

Low-quality bases of raw reads were removed using Trimmomatic-0.32 (Bolger et al., 2014) with the parameters as "LEADING:3 TRAILING:3 SLIDINGWINDOW:4:15 MINLEN:36". Quality-filtered reads were mapped to hg19 reference genome by BWA version 0.79a (Li and Durbin, 2009). The BAM file was then sorted by genomic position and indexed using SortSam in picard-tools-1.107 (https://github.com/broadinstitute/picard). The BAM files for each sample were then merged into a single BAM file using MergeSamFiles in picard-tools-1.107. To prevent PCR artifacts from influencing the downstream analysis of our data, we used MarkDuplicates in picard-tools-1.107 to mark the duplicate reads, which were ignored in downstream analysis. GATK version 2.8 (DePristo et al., 2011) was used for SNV discovery by the parameters as recommended (http://www.broadinstitute.org/gsa/wiki/index.php/Best_Practice_Variant_Detection_with_the_GATK_v3). We used the GATK unified genotyper (UG) for SNV discovery and calculation of genotype likelihoods on each family with "trio" mode. We considered all SNVs called by the GATK UG with a Phred-quality score > Q10 as a starting point before filtering in order to

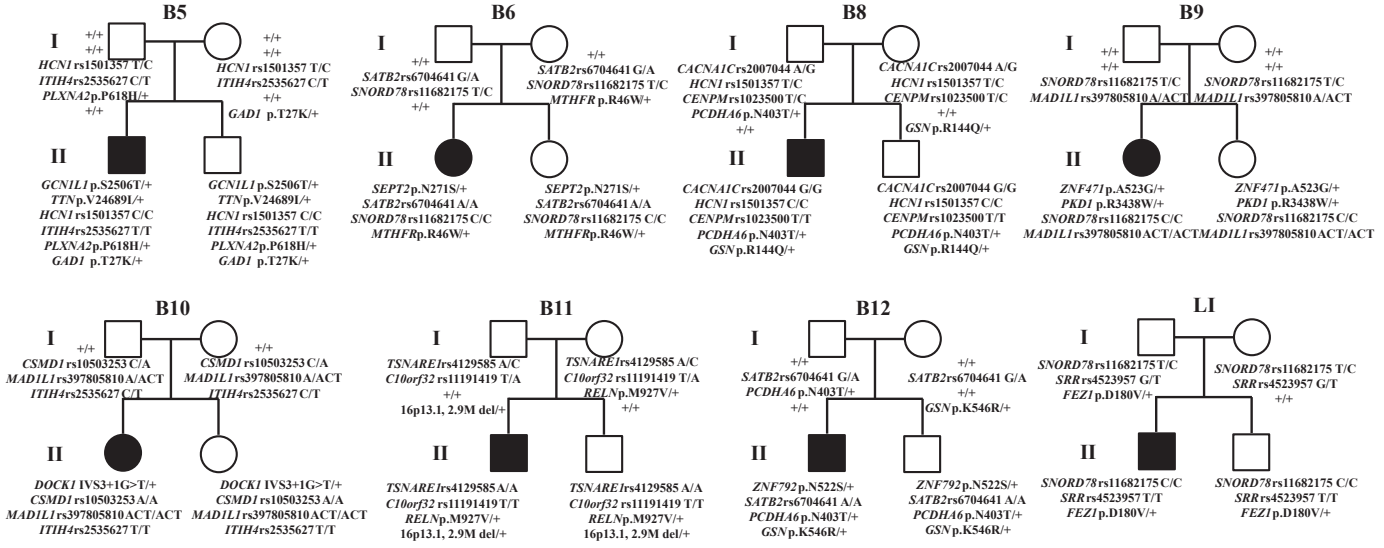

Fig. 4. Pedigrees of 8 families with schizophrenia-risk genetic variants showing the polygenic burden. The *de novo* mutations or inherited variants are shown for each family member. Males are denoted by squares and females by circles. Black symbols indicate individuals diagnosed with schizophrenia. "+" indicates the allele containing the reference (wild-type) sequence.

maximize sensitivity. We used the GATK Variant Quality Score Recalibration (VQSR) to filter spurious SNVs due to sequencing errors and mapping artifacts. We annotated all variants according to RefSeq gene transcripts (accessed from the UCSC Genome Browser, http://genome.ucsc.edu) using our house script.

4.4. De novo SNPs and indel detection

In order to accurately distinguish true *de novo* SNPs from errors, we used machine-learning classifier called forestDNM (Michaelson et al., 2012) to predict the validation status of the putative DNMs in all families. The method was designed to distinguish *de novo* variants from false-positive DNMs that arose from errors in sequencing, alignment, or variant calling. We applied following filtering criteria for *de novo* indel detection: a) Phred-scaled quality score (QUAL) of all family members more than Q30; b) QualByDepth (QD: variant confidence from the QUAL field/unfiltered depth) more than 10; c) MappingQualityZero (MQ0: total counts across all samples that have reads with a mapping quality of zero) less than 4; d) The call from the offspring needed to be supported by at least 30% of the reads, and there needed to be at least a 17× read depth; e) The *de novo* indel calls where both parents were genotyped as homozygous reference and at the offspring was genotyped as heterozygous; f) The *de novo* indels were also detected by PINDEL (Ye et al., 2009).

4.5. Detection of CNVs

The CNVs (>1 Kb) were calculated by combining the algorithms of Breakdancer (Chen et al., 2009), CNVnator (Abyzov et al., 2011), Delly (Rausch et al., 2012) and Lumpy (Layer et al., 2014). The putative inherited CNVs were detected by 2 or more algorithms (80% reciprocal overlap) and were present in at least one parent. Putative *de novo* CNVs were selected from calls detected by 4 algorithms (80% reciprocal overlap) and were not present in their parents or in any other individuals. The total list of the SNPs, indels and CNVs has been included in the SZDB dataset (www.szdb.org) (Wu et al., 2017).

4.6. Experimental validation

37 candidate DNMs were selected randomly for validation by Sanger sequencing. Primers were designed to span at least 150 bp upstream and downstream of the candidate DNMs (Table S5).

4.7. DNMs and father's age at conception of child

We combined the data reported by Michaelson et al. (2012) with our data to detect the potential correlation between the number of DNMs and parents' age at conception. As the DNMs that we identified had a false-positive variant calling rate around 21.6%, we adjusted the number of DNMs by multiplying the positive rate. The age of the fathers and mothers in our study were strongly correlated ($R^2 = 0.83$), therefore it is difficult to distinguish a father's age effect from a mother's age effect. To separate the age effect of the father and/or mother, we used multiple regression to test the relationship between number of DNMs and fathers' or/and mother's age at conception. B11 family was excluded from further analysis because we generated the sequencing data from 2 different sequencing platforms.

4.8. Polygenic risk score analysis and comparison

Polygenic risk scores were calculated using the method described by Ahn et al. (2016). We scored our samples using the most recent 128 schizophrenia-risk SNPs (Schizophrenia Working Group of the Psychiatric Genomics, 2014). The log of odds ratio of an allele was multiplied by 0, 1 or 2 depending on the number of reference alleles. The total polygenic score is a sum of across SNPs. For the comparisons of probands' polygenic risk scores and their mothers, fathers or parents (average), we used the paired *t*-test.

4.9. Identification of rare inherited variants in schizophrenia-related gene sets

Five prediction algorithms (SIFT (Ng, 2003; Kumar et al., 2009), PolyPhen-2 HumDiv, PolyPhen-2 HumVar (Adzhubei et al., 2010, 2013), LRT (Chun and Fay, 2009) and MutationTaster (Schwarz et al., 2014)) were used for predicting potential damaging effect. Missense variants were rated as damaging when at least 2 of the 5 predictions suggested a potential deleterious effect. We followed the prediction of the dbNSFP database (Liu et al., 2016). Rare (MAF $\leq$ 0.01 in the 1000 Genomes Project (Sudmant et al., 2015)) and potential damaging variants including nonsense, frameshift, and damaging missense variants were selected for further analysis. All schizophrenia-related genes carrying rare damaging or LOF variants were considered and were grouped into 3 classes:

a) Class I – 38 genes listed in the core gene set of SZGR (Jia et al., 2010).
b) Class II – 206 genes in schizophrenia-related pathways or complexes, including postsynaptic density protein 95 (PSD-95) complexes, neuronal activity-regulated cytoskeleton-associated protein (ARC) postsynaptic signaling complexes, N-methyl-d-aspartate receptor (NMDAR) network and mGluR5 complexes. The entire list was compiled by Kirov et al. (2012).
c) Class III – 352 genes were taken from the108 schizophrenia susceptibility loci showing a genome-wide significance (Schizophrenia Working Group of the Psychiatric Genomics, 2014).

4.10. Gene-based test

The gene-based burden test was performed by using the PLINK/seq (https://atgu.mgh.harvard.edu/plinkseq/) based on all variants in the analyzed subjects relative to human reference sequence (hg19; http://genome.ucsc.edu/). We also performed the same test based on rare damaging variants or LOF variants.

4.11. Polygenic burden analysis of rare variants for schizophrenia-related gene sets

We selected the following gene sets for polygenic burden analysis:

a) 685 genes from the postsynaptic density (PSD) “human core” gene sets compiled by Kirov et al. (2012), which include NMDAR complex, PSD-95 and mGluR5 subsets.
b) 431 genes from pre-synapse gene set compiled by Kirov et al. (2012), which include pre-synaptic active zone and synaptic vesicle.
c) 352 genes related to the 108 schizophrenia susceptibility loci of genome-wide significance (Schizophrenia Working Group of the Psychiatric Genomics, 2014).
d) 611 genes with a non-synonymous DNM reported by Fromer et al. (2014).
e) 1589 genes listed as targets of FMRP were taken from Darnell et al. (2011) and Ascano et al. (2012).
f) 432 genes related to axon guidance pathways, 237 genes related to nerve impulse, 1847 genes related to nervous

system development, 309 genes related to neuronal cell body, and 699 genes related to neuron projection were taken from Yuen et al. (2015).

The enrichment analysis of rare variants in each gene set was conducted using the Fisher's exact test.

4.12. Other published datasets for data mining

Distinction of rare variants and common variants depended on the MAF in the 1000 Genomes Project (Sudmant et al., 2015): the cut-off of rare variants is MAF $\leq$ 0.01 and the cut-off of common variants is MAF > 0.01. The eQTL analysis was performed by Genevar analysis (Yang et al., 2010). The *p*-values of differential expression were taken from SZDB (Wu et al. (2017) and references therein), which was estimated by using datasets of GSE53987 (Lanz et al., 2015) and GSE35978 (Chen et al., 2013) from Gene Expression Omnibus (GEO) (Clough and Barrett, 2016). *de novo* SNVs had been annotated with known and predicted regulatory elements by RegulomeDB (Boyle et al., 2012), including DNAase hypersensitivity sites, binding sites of transcription factors, and promoter regions.

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Supplementary data

Supplementary data related to this article can be found at http://dx.doi.org/10.1016/j.jgg.2017.05.005.

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Supplemental Figures

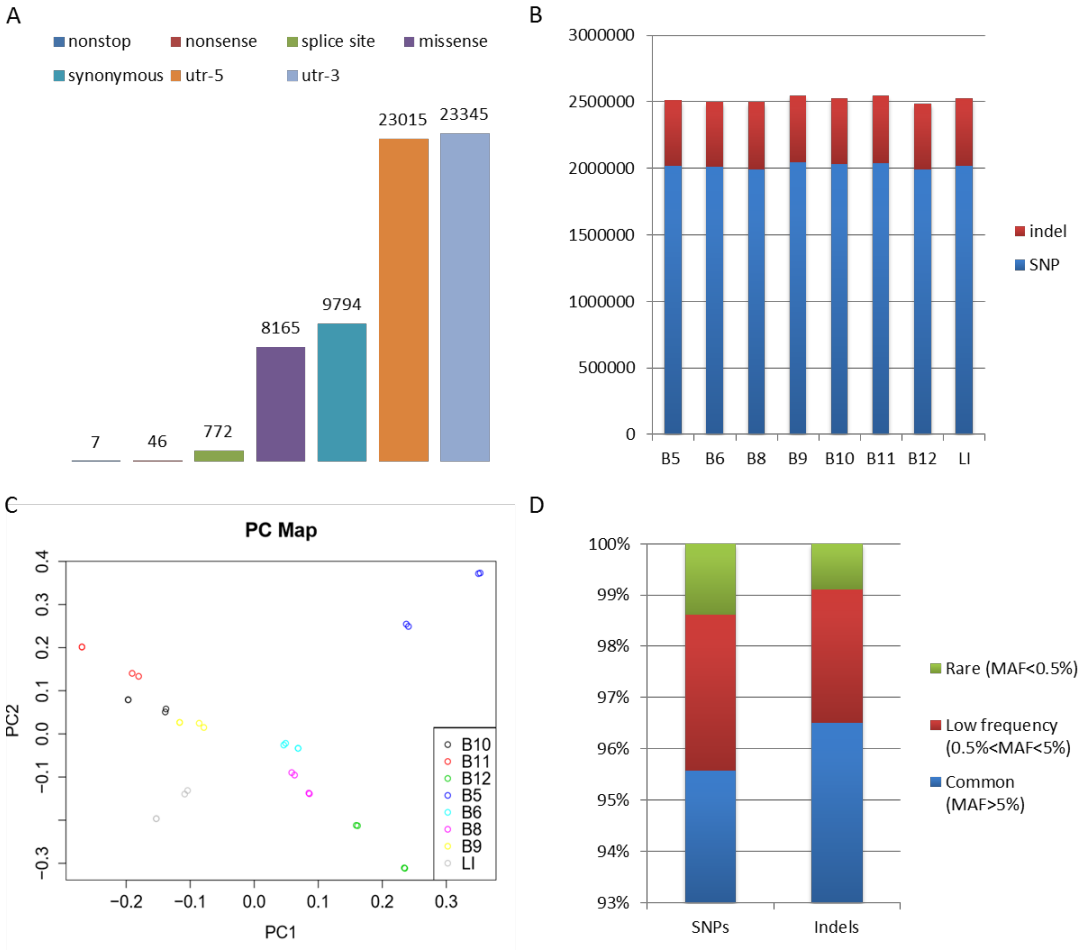

Fig. S1. The average distribution of SNVs in eight families (32 samples). The average number of various types of variations (**A**) and frequency (**B**) per individual; **C**: Principal component analysis (PCA) plot (the first and the second principal components) for 32 samples. The PCA was implemented by GCTA (Yang et al., 2011). **D**: The plot of shared SNVs in each family.

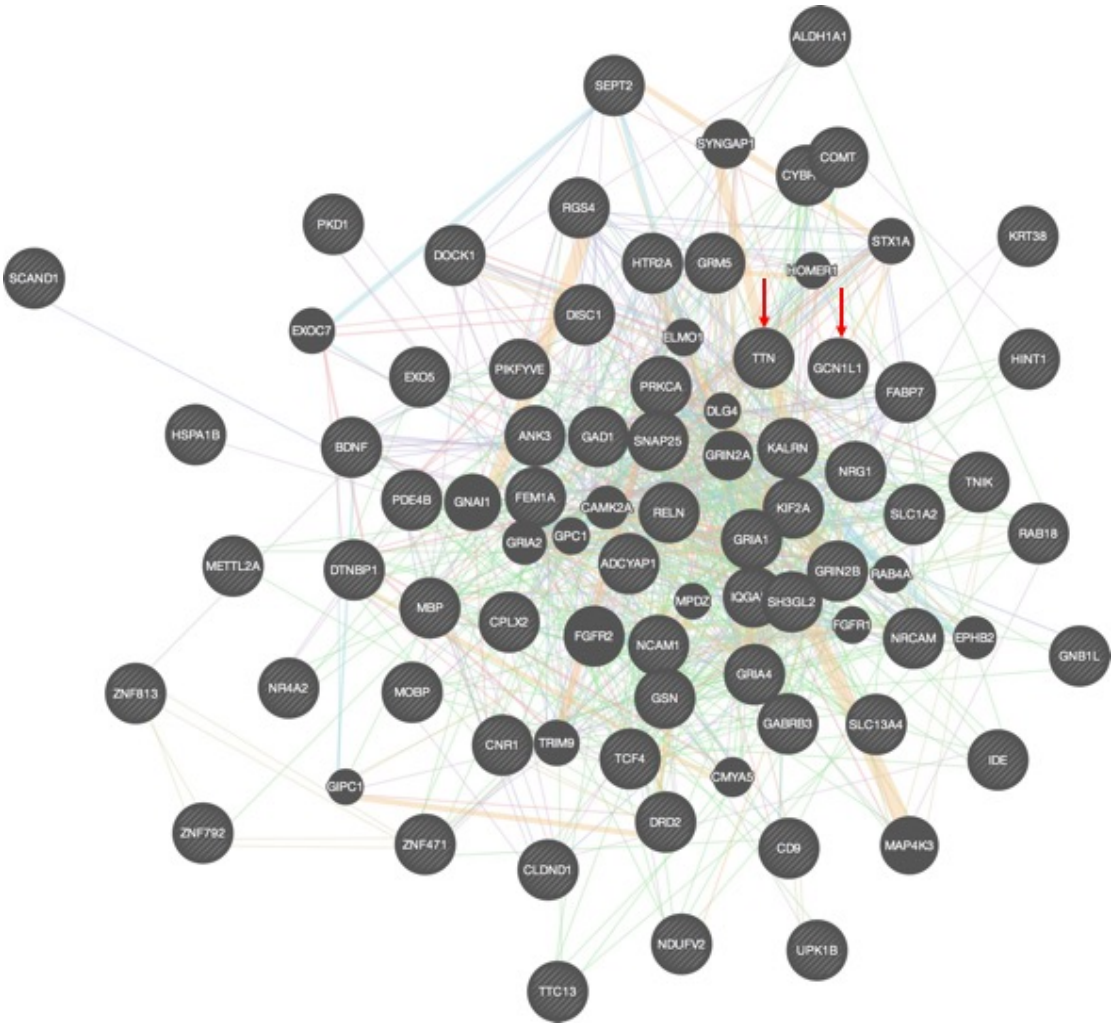

Fig. S2. Protein–protein interaction (PPI) network of 23 genes with DNMs and 42 schizophrenia top genes. The network was based on the genes retrieved from GeneMANIA (Montojo et al., 2014) by inputting 23 genes hit by *de novo* mutations in the coding region, UTR region, 3′-downstream region or 5′-upstream region, and 42 SCZ top genes (identified by convergent functional genomics [CFG] score) of the SZDB data set (www.szdb.org). The *TTN* (titin; p.V24689I) and *GCN1L1* (GCN1 eIF2 alpha kinase activator homolog; p.S2506T) genes identified in family B5 were marked by arrows.

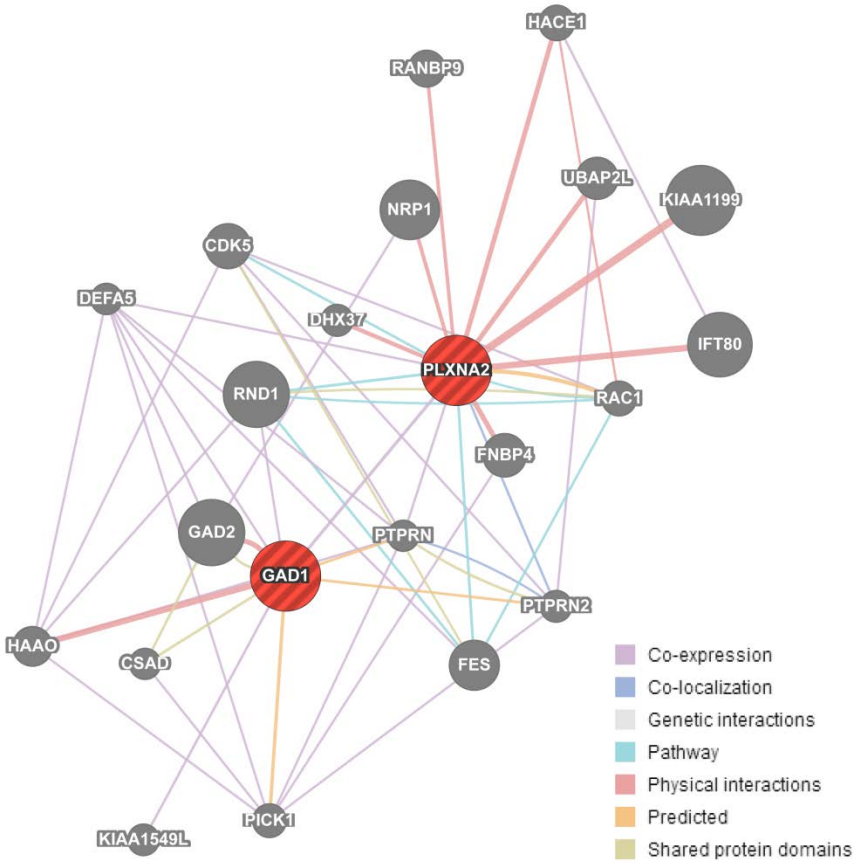

Fig. S3. GeneMANIA network showing potential interactions with GAD1 and PLXNA2. The network was based on the genes retrieved from GeneMANIA (Montojo et al., 2014) by inputting GAD1 and PLXNA2. GAD1 and PLXNA2 are marked with red color, and the other genes that interacted with GAD1 or PLXNA2 are marked with grey color.

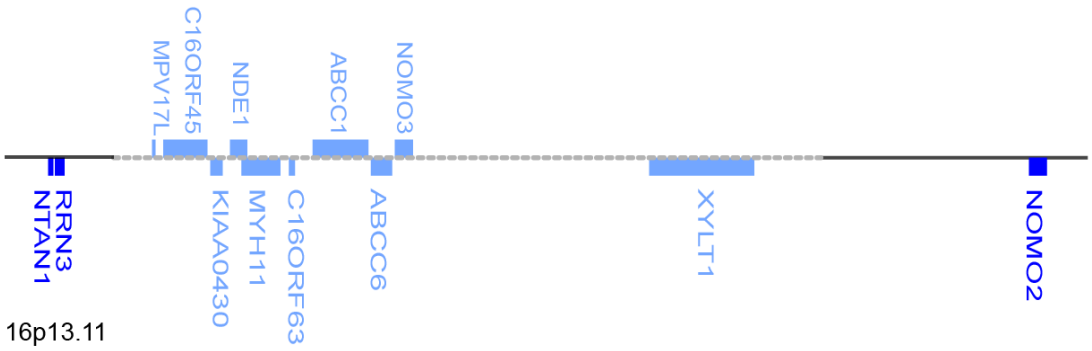

Fig. S4. Schematic profile of the large deletion (16p13.11; 2.9M) in proband of B11 family. The deletion contains 10 genes (*MPV17L*, *C16orf45*, *KIAA0430*, *NDE1*, *MYH11*, *C16orf63*, *ABCC1*, *ABCC6*, *NOMO3* and *XYLT1*) marked with lighter color and grey dotted line.

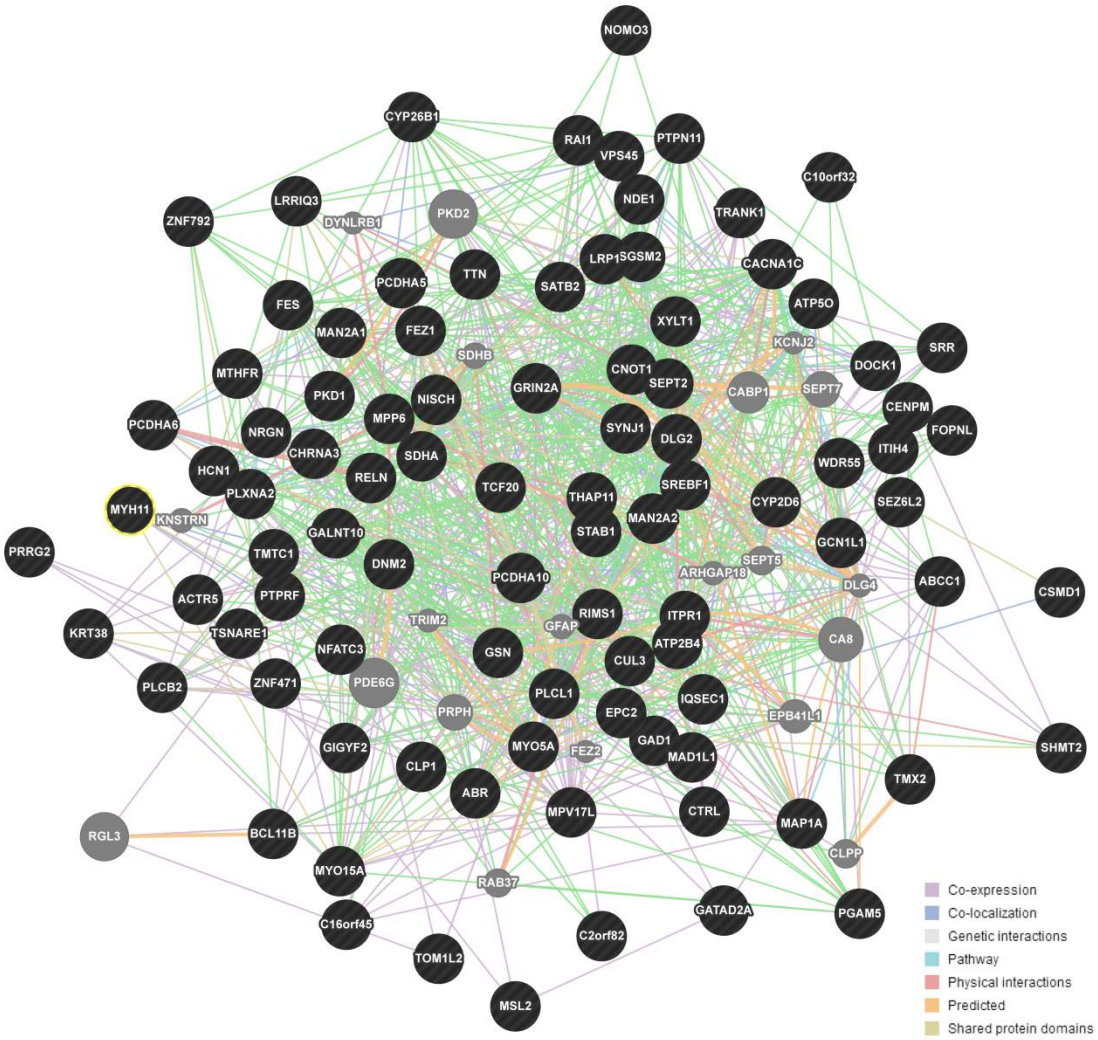

Fig. S5. GeneMANIA network categories. The network analysis was based on genes from the GeneMANIA (Montojo et al., 2014) by inputting genes harbored *de novo* missense mutations and inherited highly risky or rare damaging mutations (Tables 1, S10, S13 and S16). Inputted genes are marked with black color, and the other genes that interacted with the inputted genes are marked with grey color.

Supplemental Tables

Table S1. Demographical information and sequencing depth for each member of the 8 families with MZ twins

Family ID	Sample	Original sample ID	Sex [a]	Age	Kinship	Onset age	Duration of psychosis (year)	Average sequencing depth (X)
B5	B5-F	B51	M	45	Father	-	-	35.77
	B5-M	B52	F	47	Mother	-	-	31.91
	B5-T1	B53	M	24	Son	-	-	30.78
	B5-T2	B54	M	24	Son	23	1	31.4
B6	B6-F	B64	M	52	Father	-	-	30.81
	B6-M	B61	F	50	Mother	-	-	31.61
	B6-T1	B62	F	25	Daughter	-	-	32.31
	B6-T2	B63	F	25	Daughter	21	4	32.92
B8	B8-F	B81	M	44	Father	-	-	31.44
	B8-M	B82	F	42	Mother	-	-	31.45
	B8-T1	B83	M	22	Son	-	-	31.37
	B8-T2	B84	M	22	Son	12	10	33.27
B9	B9-F	B91	M	62	Father	-	-	33.92
	B9-M	B92	F	60	Mother	-	-	33.66
	B9-T1	B94	F	26	Daughter	-	-	30.13
	B9-T2	B93	F	26	Daughter	25	1	31.2
B10	B10-F	B101	M	50	Father	-	-	31.55
	B10-M	B102	F	50	Mother	-	-	30.91
	B10-T1	B103	F	24	Daughter	-	-	32.13
	B10-T2	B104	F	24	Daughter	21	3	31.22
B11	B11-F	B111	M	76	Father	-	-	32
	B11-M	B112	F	68	Mother	-	-	31.79
	B11-T1	B114	M	40	Son	-	-	31.11
	B11-T2	B113	M	40	Son	32	8	33.6
B12	B12-F	B121	M	46	Father	-	-	31.7
	B12-M	B122	F	46	Mother	-	-	31.58
	B12-T1	B123	M	19	Son	-	-	34.88
	B12-T2	B124	M	19	Son	17.5	1.5	32.29
LI	LI-F	LF	M	49	Father	-	-	28.46
	LI-M	LM	F	46	Mother	-	-	29.75
	LI-T1	LS	M	26	Son	-	-	31.51
	LI-T2	LJ	M	26	Son	24	2	28.3

The demographic information for each sample was described in our recent study (Li et al., 2016). [a] M - male; F - female.

Table S2. The number of shared SNVs in each family

Family ID	Shared SNPs	Shared indels
B5	2021376	495686
B6	2010400	489202
B8	1995282	504487
B9	2049020	497705
B10	2036478	491321
B11	2040822	505594
B12	1995286	494165
LI	2020038	504579
Mean	2021087.8	497842
SD	20190.49	6380.7

All variants in 32 individuals from the 8 families were scored relative to human reference genome (hg19; http://genome.ucsc.edu/).

Table S3. The number of DNMs in 8 pairs of MZ twins

Family ID	T1 unique SNPs [a]	T2 unique SNVs [a]	Shared SNVs	Shared indels
B5	0	1	54	1
B6	3	2	67	4
B8	5	7	49	1
B9	0	0	79	2
B10	0	1	66	1
B11	8	5	36	1
B12	2	3	58	1
LI	2	2	52	1
Total	20	21	461	12

[a] T1 - twin 1; T2 - twin 2; Twin 2 of each family had schizophrenia.

Table S4. The distribution of DNMs in 8 pairs of MZ twins.

This table was listed as a separate Excel file, because the table is too big. The 8 DNMs that were not validated in this study were excluded.

Table S5. Validation of DNMs in 8 pairs of MZ twins

Sample	SNP	Allele	Gene	Predicted function	Validation	Primer Sequence (5'-3')	Sequencing region & Product length
B53 B54	chr2:179436794	C/T	*TTN*	p.V24689I	Validated	U: TCTTTATGCCAGGTTACAGC	chr2:179436604-179437094; 491bp
						L: TACTATTTCAGGGTTCTCGC	
B53 B54	chr1:40982533	A/G	*EXO5*	3'-downstream	Validated	U: CAATAGCAGAAGCCTAGAAGC	chr1:40982362-40982602; 241bp
						L: CTTCTAAGATGGCTTGGGATT	
B53 B54	chr4:3769798	T/C	*ADRA2C*	3'-utr	False positive	U: TTCGTGCTCTGCTGGTTCCC	chr4:3769504-3770011; 508bp
						L: ACCCTGACCTCACAGCCACA	
B53 B54	chr12:120569035	C/G	*GCN1L1*	p.S2506T	Validated	U: CAAGTGCTGGTCCAAGGG	chr12:120568834-120569295; 462bp
						L: GCCAAGATAGCAGGGAGG	
B53 B54	chr15:90200559	A/G	*KIF7*	5'-upstream	False positive	U: AAACCCATCACTCTAGGAGG	chr15:90200283-90200730; 448bp
						L: ACATCCACCAGAAAGAGCAA	
B53 B54	chr17:39595049	G/T	*KRT38*	p.P265H	Validated	U: ACATCCTGGCGGTTGGTC	chr17:39594971-39595341; 371bp
						L: TGAGGTGGGTCTGAGGGTC	
B53 B54	chr17:41382051	T/C	*LINC00854*	5'-upstream	Unknown	U: CAGTTTGTAGAAAGCAGGAG	
						L: TTTAGTACCGCATAGAGCC	
B103 B104	chr9:17578171	A/C	*SH3GL2*	5'-upstream	Validated	U: CGGATGGAGGACCCTT	chr9:17577961-17578276; 316bp
						L: GGAGCAGAGCAGCACTAA	
B103 B104	chr3:118923152	T/C	*UPK1B*	3'-utr	Validated	U: CTTACATTGCCAAGTCAGA	chr3:118922923-118923330; 408bp
						L: GGCATCAAGAGAATCACC	
B103 B104	chr11:69923089	A/C	*ANO1*	5'-upstream	False positive	U: CCAGCCTGAGAACCAAAT	chr11:69922865-69923308; 444bp
						L: ACAGCCGATGCTCAATAA	
B123 B124	chr6:56978734	T/G	RP11_203B9.4	3'-downstream	Validated	U: CATGTTAGCCAGGATGCTC	chr6:56978620-56978821; 202bp
						L: TTCTTGAACAATTCACTCTCG	
B123 B124	chr2:172411647	G/C	*CYBRD1*	3'-utr	Validated	U: TGCTTCTCCTATTAGCCATAT	chr2:172411394-172411722; 329bp
						L: ATTGTCTCCCAGAATTAACAG	

B123 B124	chr4:80994100	C/A	*ANTXR2*	5'-utr	False positive	U: GCCGGAACTCTTGACGAATC	chr4:80993933-80994251; 319bp
						L: CGGGAGGAAAGTTTCGGAGT	
B123 B124	chr17:60527119	A/G	*METTL2A*	3'-utr	Unknown	U: CTGGGCAACAAAGCAAGACTCT	
						L: CTCGATCTCCTGACCTCGTGA	
						U: TGATCGTGGTCTTCGCTTTA	
						L: GACGGAGTCTTCCTCTGTTGC	
B123 B124	chr19:35449194	T/C	*ZNF792*	p.N522S	Validated	U: CAGCGCCCTGATAAGGTCTG	chr19:35448916-35449411; 496bp
						L: CACGGGTGTGGTGAGTGTG	
B84	chr17:41382052	T/C	*LINC00854*	5'-upstream	Unknown	U: AGAGGGTCCCATTGAACT	
						L: TGCTCTTTAGTACCGCATAG	
B83 B84	chr2:209224680	C/T	*PIKFYVE*	3'-downstream	Validated	U: GGATAGAGCGTACCCTGA	chr2:209224567-209224976; 410bp
						L: GGCGCTCTTCTTACCTGT	
B83 B84	chr8:21966891	C/A	*NUDT18*	5'-utr	False positive	U: TCACCTGCTCGCTGAGGAAC	chr8:21966646-21967128; 483bp
						L: ACTGACAGGCGGCAGAAGC	
B83 B84	chr20:9966603	G/A	*LOC101929371*	5'-upstream	Validated	U: CCATTTACACTGGACTGGCACA	chr20:9966366-9966703; 338bp
						L: AAATGTGGGCAGGAATAAGGA	
B83 B84	chr19:4796824	G/A	*FEM1A*	3'-downstream	Validated	U: AACGTCCGGTTAATCCCTG	chr19:4796713-4796954; 242bp
						L: AACCCACTCCTTTGATGCA	
B93 B94	chr10:94297199	C/A	*IDE*	synonymous	Validated	U: TACCAATAGCTTTACGAGGGT	chr10:94296973-94297280; 308bp
						L: GAATAATCCAGCCATCAAGAG	
B93 B94	chr16:2147410	G/A	*PKD1*	p.R3438W	Validated	U: TGTGGGTGTCTTGGGTAGG	chr16:2147174-2147480; 307bp
						L: GGGAACGCTCAGTTGGC	
B93 B94	chr7:135414085	A/G	*SLC13A4*	5'-upstream	Validated	U: GAGCAGAGTCAAAGGGTTA	chr7:135413881-135414168; 288bp
						L: AAATGAGGCCAGAATGTAG	
B93 B94	chr19:57037004	C/G	*ZNF471*	p.A523G	Validated	U: AAACCGTATGAATGCAAGGA	chr19:57036805-57037107; 303bp
						L: ATTGGAAGTTTGGCTGAAGG	

LJ LS	chr12:88176981	C/A	*MKRN9P*	non-coding	False positive	U: AGCAGACCAGTTCACACGC L: GAAATACAAGGAGGCAATGAGC	chr12:88176864-88177267; 404bp
LJ LS	chr7:31375247	C/G	*NEUROD6*	3'-downstream	False positive	U: CCATTGCATTTTTGCATCC L: TCAGAATCTTGGTTCAGAC	chr7:31375077-31375366; 290bp
B62 B63	chr15:90929885	T/C	*IQGAP1*	5'-upstream	Validated	U: GGAGTCAGGCTATGATTTGGC L: CCTCTTCCGTTTATTCTTTGC	chr15:90929609-90930097; 489bp
B62 B63	chr2:242283282	A/G	*SEPT*	p.N271S	Validated	U: TAATGTTCTCGTGTTGCCAGTG L: ACTGAGGGCGGCAATGAA	chr2:242283093-242283431; 339bp
B62 B63	chr4:10042236	C/T	*SLC2A9*	5'-upstream	False positive	U: ATCATCGGGTAATGCCAA L: AAGCTGACCCTCCAATGTT	chr4:10041979-10042430; 452bp
B62 B63	chr1:231116417	A/G	*TTC13*	5'-upstream	Validated	U: TCTGCCTCCCAAGTTCAAGCAA L: TGCGGGTGGGAATGTGACTAAA	chr1:231116166-231116682; 517bp
B63	chr18:55144502	A/T	*ONECUT2*	3'-utr	False positive	U: CCTTCCTGGAGCGAGTTA L: GAGCGCAAGTGAACTGAC	chr18:55144240-55144714; 475bp
B113 B114	chr3:98242563	T/C	*CLDND1*	5'-upstream	Validated	U: CTGGCAGACAGTATCCATCAA L: TAAGATGTCCCTGCTTCCTGT	chr3:98242291-98242659; 369bp
B113 B114	chr3:13974618	C/G	*FGD5P1*	non-coding	False positive	U: CAAAATCTTTCCTCTTGTGGCG L: GGAGTGGAGGGCAAGGGT	chr3:13974381-13974894; 514bp
B113 B114	chr19:53997821	C/T	*ZNF813*	3'-downstream	Validated	U: AGGCACCCAGAAGGATAAG L: GAGTTTCGCTCTTGTTGACC	chr19:53997571-53997891; 321bp
B84	chr13:45830708	C/T	*GTF2F2*	intronic	Validated	U: ACCCCTGGTGAATAAAGTAAGT L: GTTCGTTTATGTTTCTCAGTCT	chr13:45830487-45830883; 397bp
B84	chr2:167878685	A/G	*XIRP2*	intronic	Validated	U: TCAGATTTATTTTCTGTATTTCACC L: ATTATGCAATTGATATGTTCTGAGA	chr2:167878435-167878940; 506bp
B84	chr6:149959476	T/A	*KATNA1*	intronic	Validated	U: GTCTTGCTATGTTGTCTAGGCT L: CTCTGCGATGGTCTATTATCA	chr6:149959302-149959615; 314bp

Table S6. The genomic locations of DNMs in 8 pairs of MZ twins.

Genomic region	Number
CDS (missense)	7
CDS (synonymous)	1
3′-UTR	3
5’-UTR	0
3′-upstream	5
5’-upstream	9
Splice site	1
Intronic	223
Intergenic	255

Table S7. The regulatory elements hit by *de novo* SNVs in 8 pairs of MZ twins.

This table was listed as a separate Excel file, because the table is too big.

Table S8. Gene-based associations with schizophrenia

Gene symbol	Transcript ID	Position	NVAR[a]	*P*[b]	I[c]	DESC[d]
KCNQ2	ENST00000360480	chr20:62038277..62103437	564	0.000999	0.001	3245/2910
LIMK2	ENST00000331728	chr22:31608618..31674200	235	0.000999	0.001	1256/1054
COPS4	ENST00000503682	chr4:83956665..83995802	146	0.000999	0.001	712/588
PHYKPL	ENST00000308158	chr5:177639942..177659519	112	0.000999	0.001	658/475
AIF1L	ENST00000372298	chr9:133972131..133996503	102	0.000999	0.001	622/420
ALKBH4	ENST00000292566	chr7:102098010..102104491	34	0.000999	0.002	232/172
POLD2	ENST00000223361	chr7:44154865..44160926	18	0.001998	0.001	109/83
SNAP47	ENST00000366759	chr1:227923986..227968107	132	0.001998	0.001	749/579
LTV1	ENST00000367576	chr6:144164990..144182969	62	0.001998	0.001	286/200
HCK	ENST00000375852	chr20:30640592..30688137	121	0.001998	0.001	861/573
CCDC125	ENST00000396499	chr5:68579047..68616331	171	0.001998	0.001	1182/833
KRT7	ENST00000543899	chr12:52627215..52661714	143	0.001998	0.001	892/781
BATF3	ENST00000243440	chr1:212861303..212873096	60	0.002997	0.001	284/218
EPHA1	ENST00000275815	chr7:143088823..143105830	53	0.002997	0.001	268/218
RGL4	ENST00000382833	chr22:24034288..24040735	10	0.002997	0.001	104/74
MET	ENST00000397752	chr7:116339672..116436110	208	0.002997	0.001	1048/869
EPC2	ENST00000258484	chr2:149402806..149543718	523	0.003996	0.001	3275/2425
ATP5C1	ENST00000335698	chr10:7830319..7849341	69	0.003996	0.001	475/352
ANXA1	ENST00000376911	chr9:75773632..75784327	27	0.003996	0.001	188/122
PSMG1	ENST00000380900	chr21:40547848..40555160	33	0.003996	0.001	224/180
TMEM106B	ENST00000396667	chr7:12254545..12271452	146	0.003996	0.001	1487/937
C5orf45	ENST00000403396	chr5:179264564..179285752	66	0.003996	0.001	474/333
C11orf65	ENST00000525729	chr11:108181511..108332126	489	0.003996	0.001	3693/2618
BPI	ENST00000262865	chr20:36932660..36964414	178	0.004995	0.001	1201/1019
TAPBPL	ENST00000266556	chr12:6561603..6571298	38	0.004995	0.001	321/217
C14orf182	ENST00000399206	chr14:50459315..50472405	45	0.004995	0.001	386/240
FEZF1	ENST00000427185	chr7:121942621..121943610	11	0.004995	0.001	54/33
ATM	ENST00000452508	chr11:108098459..108235433	359	0.004995	0.001	2758/1947
SQSTM1	ENST00000510187	chr5:179248642..179263515	75	0.004995	0.001	599/396
HEATR5A	ENST00000543095	chr14:31763142..31871390	427	0.004995	0.001	2069/1567
LRWD1	ENST00000292616	chr7:102105852..102113256	50	0.005994	0.001	404/306
PLA2G4E	ENST00000399518	chr15:42276176..42342824	378	0.005994	0.001	2229/1658
VAMP1	ENST00000400911	chr12:6572048..6579565	43	0.005994	0.001	256/181
C12orf28	ENST00000547771	chr12:70220331..70345690	485	0.005994	0.001	2456/2052
DEGS1	ENST00000323699	chr1:224371225..224379317	31	0.006993	0.001	172/142
KRT81	ENST00000327741	chr12:52680766..52685213	23	0.006993	0.002	136/107
C4orf34	ENST00000511809	chr4:39554679..39606533	227	0.006993	0.001	1320/1042
THAP9	ENST00000536314	chr4:83822334..83839836	64	0.006993	0.002	374/276
GDA	ENST00000238018	chr9:74764627..74865422	279	0.007992	0.001	1801/1356
RIN1	ENST00000311320	chr11:66099987..66103046	14	0.007992	0.001	85/64
SMAD5	ENST00000545620	chr5:135489891..135513085	63	0.007992	0.002	671/438

CHRNA10	ENST00000250699	chr11:3687985..3692121	18	0.008991	0.001	216/147
SPC25	ENST00000282074	chr2:169728069..169745594	78	0.008991	0.001	464/375
PEX11A	ENST00000300056	chr15:90226947..90233145	17	0.008991	0.001	200/114
CTC1	ENST00000315684	chr17:8132763..8150923	55	0.008991	0.003	454/297
INSR	ENST00000341500	chr19:7117426..7293898	1310	0.008991	0.001	6621/5679
UBE2D3	ENST00000357194	chr4:103719434..103789810	223	0.008991	0.001	2192/1542
FXYD5	ENST00000423817	chr19:35647174..35660508	71	0.008991	0.001	412/302
SYT8	ENST00000535046	chr11:1853107..1857270	20	0.008991	0.001	128/101
SLC39A2	ENST00000298681	chr14:21467913..21469268	6	0.00999	0.001	68/48
SP3	ENST00000310015	chr2:174775562..174829798	181	0.00999	0.001	860/563
TMPRSS11F	ENST00000356291	chr4:68919659..68995529	313	0.00999	0.001	2174/1881
CHI3L2	ENST00000369748	chr1:111770423..111784816	79	0.00999	0.001	551/460
CDKN2AIP	ENST00000504169	chr4:184366145..184367749	8	0.00999	0.001	46/32
CCDC149	ENST00000504487	chr4:24810094..24914338	359	0.00999	0.001	2029/1650

[a] NVAR refers to the number of variants in all 32 individuals from the 8 families when compared to the reference genome (hg19). [b] *P*-value is based on permutations, the empirical significance. [c] I-value indicates the proportion of null replicates for which the best test statistic was tied. [d] DESC contains the number of variants in cases/controls.

Table S9. The polygenic risk score and summary of results in the comparison of probands and healthy parents

Family ID	Father	Mother	Mean [a]	Proband	Proband ***vs*** mother increase (%) [b]	Proband ***vs*** mean increase (%) [c]
B5	5.1847	5.05077	5.117735	5.42641	7.44	6.03
B6	6.05225	5.41625	5.73425	5.57301	2.89	-2.81
B8	5.30071	5.3839	5.342305	5.90051	9.60	10.45
B9	6.02009	5.61828	5.819185	6.52813	16.19	12.18
B10	5.53655	4.69908	5.117815	5.41896	15.32	5.88
B11	5.16369	5.95777	5.56073	5.58196	-6.31	0.386
B12	6.15814	5.31027	5.734205	6.0097	13.17	4.80
LI	5.75368	4.65429	5.203985	5.22316	12.22	0.37
p-value	0.3528	0.0083	0.0211	-	-	-
R^2	0.02164	0.5834	0.4678	-	-	-

[a] The mean score is calculated as the average of mother's score and father's score. [b] The increase of risk score is calculated as (the proband score – mother's score)/mother's score x 100%. [c] The increase of risk score is calculated as (the proband score – the mean score of mother and father)/mean score x 100%.

Table S10. The genotypes of the SCZ-risk alleles in MZ twins

Family ID	dbSNP ID	MAF[a]	Genotype F [b]	Genotype M [b]	Genotype T [b]	Gene	Transcript ID	Function	***p***-value [c]
B8	rs2007044	G=0.4964	A/G	A/G	G/G	*CACNA1C*	NM_000719	intron	3.22E-18
B8	rs2973155	T=0.3728	T/C	T/C	C/C	Intergenic			1.11E-10
B8	rs6434928	G=0.2821	G/A	G/A	G/G	Intergenic			2.06E-11
B8	rs1023500	C=0.2969	T/C	T/C	T/T	*CENPM*	NM_001002876	intron	3.43E-08
B10	rs1023500	C=0.2969	T/C	T/C	T/T	*CENPM*	NM_001002876	intron	3.43E-08
B10	rs10503253	A=0.2270	C/A	C/A	A/A	*CSMD1*	NM_033225	intron	1.06E-08
B10	rs397805810	NA	A/ACT	A/ACT	ACT/ACT	*MAD1L1*	NM_001013836.1.	intron	8.2E-15
B10	rs59979824	A=0.267	C/A	C/A	C/C	*AC013401.4*	ENST00000434525	intron	8.41E-09
B10	rs8042374	G=0.4495	A/G	A/G	A/A	*CHRNA3*	CCDS10305.1	intron	2.44E-13
B10	rs1501357	C=0.3992	T/C	T/C	C/C	*HCN1*	CCDS3952.1	intron	5.05E-09
B10	rs2535627	T=0.4665	T/C	T/C	T/T	*ITIH4*	NM_001166449	3downstream	4.26E-11
B5	rs2535627	T=0.4665	T/C	T/C	T/T	*ITIH4*	NM_001166449	3downstream	4.26E-11
B5	rs1501357	C=0.3992	T/C	T/C	C/C	*HCN1*	CCDS3952.1	intron	5.05E-09
B5	rs11139497	T=0.4467	T/A	T/A	A/A	*RP11-388B24.3*	ENST00000417796	intron	3.61E-09
B5	rs2905426	T=0.4097	G/T	G/T	G/G	Intergenic			3.63E-10
B5	rs4702	G=0.3538	G/A	G/A	G/G	*FES*	NM_001143783	5upstream	8.30E-14
B5	rs55661361	A=0.4058	G/A	G/A	G/G	*NRGN*	NM_001126181	intron	2.80E-12
B5	rs6704768	G=0.4619	G/A	G/A	G/G	*GIGYF2*	NM_001103146	intron	2.32E-12
B12	rs11685299	A=0.2534	C/A	C/A	C/C	*CUL3*	NM_001257197.1	intron	1.12E-08
B12	rs5825114	G=0.3948	GA/G	GA/G	GA/GA	Intergenic			8.03E-11
B12	rs6704641	G=0.3327	G/A	G/A	A/A	*SATB2*	CCDS2327.1	intron	8.33E-09
B6	rs6704641	G=0.3327	G/A	G/A	A/A	*SATB2*	CCDS2327.1	intron	8.33E-09

B6	rs12148337	C=0.4331	T/C	T/C	T/T	Intergenic			1.79E-08
B6	rs56873913	G=0.3472	T/G	T/G	T/T	*PRRG2*	NM_000951	intron	4.69E-08
B6	rs679087	A=0.1877	C/A	C/A	C/C	*TMTC1*	NM_001193451	intron	3.91E-08
B6	rs9420	A=0.247	A/G	A/G	A/A	*TMX2*	NM_001144012	3downstream	2.24E-09
B6	rs11682175	C=0.3508	T/C	T/C	C/C	*SNORD78*	ENST00000390866	intron	1.47E-11
B9	rs11682175	C=0.3508	T/C	T/C	C/C	*SNORD78*	ENST00000390866	intron	1.47E-11
B9	rs2909457	G=0.3117	G/A	G/A	G/G	Intergenic			4.62E-08
B9	rs397805810	NA	A/ACT	A/ACT	ACT/ACT	*MAD1L1*	NM_001013836.1	intron	8.2E-15
B9	rs59979824	A=0.267	C/A	C/A	C/C	*AC013401.4*	ENST00000434525	intron	8.41E-09
LI	rs11682175	C=0.3508	T/C	T/C	C/C	*SNORD78*	ENST00000390866	intron	1.47E-11
LI	rs11210892	A=0.4455	G/A	G/A	G/G	Intergenic			3.39E-10
LI	rs2973155	T=0.3728	T/C	T/C	C/C	Intergenic			1.11E-10
LI	rs4523957	G=0.4770	G/T	G/T	T/T	*SRR*	NM_021947	intron	2.86E-10
B11	rs11191419	A=0.3906	T/A	T/A	T/T	*C10orf32*	NM_001136200	5upstream	6.20E-19
B11	rs1498232	C=0.4667	T/C	T/C	T/T	*AL645944.1*	ENST00000410855	intron	2.86E-09
B11	rs4129585	A=0.2388	A/C	A/C	A/A	*TSNARE1*	CCDS6384.1	intron	1.74E-15

[a] The minor allele frequency (MAF) from the 1000 Genomes Project (Sudmant et al., 2015). [b] F-father; M-mother; T-twins. [c] *p*-value from the original paper (Schizophrenia Working Group of the Psychiatric Genomics, 2014).

Table S11. The number of rare inherited loss-of-function and damaging missense variants in 8 families with MZ twins

Sample	Damaging missense [a]		Frameshift [b]		Nonsense [b]		Total	
	MAF <0.05 [c]	MAF<0.01 [c]	MAF<0.05	MAF<0.01	MAF<0.05	MAF<0.01	MAF<0.05	MAF<0.01
B10_father	322	236	48	44	7	6	377	286
B10_mother	273	199	49	47	10	6	332	252
B11_father	305	222	47	41	13	12	365	275
B11_mother	320	225	52	48	10	9	382	282
B12_father	325	226	44	42	13	11	382	279
B12_mother	255	192	50	47	7	5	312	244
B5_father	350	255	48	45	16	12	414	312
B5_mother	324	241	56	55	11	10	391	306
B6_mother	329	244	44	40	8	7	381	291
B6_father	357	263	47	43	7	4	411	310
B8_father	353	258	57	53	7	5	417	316
B8_mother	366	270	42	41	6	6	414	317
B9_father	351	250	42	40	15	12	408	302
B9_mother	328	229	48	45	6	5	382	279
LI_father	347	258	47	45	14	10	408	313
LI_mother	321	240	56	51	15	13	392	304
Average	326.63	238.00	48.56	45.44	10.31	8.31	385.50	291.75
B10_twin	357	264	51	47	9	6	417	317
B11_twin	316	229	53	49	14	14	383	292
B12_twin	316	222	49	48	10	7	375	277
B5_twin	352	265	56	54	18	14	426	333
B6_twin	343	261	39	37	5	3	387	301
B8_twin	359	256	54	53	6	4	419	313
B9_twin	320	229	42	40	12	11	374	280
LI_twin	332	235	51	48	11	10	394	293
Average	336.88	245.13	49.38	47.00	10.63	8.63	396.88	300.75
***p*-value** [d]								
twin vs mother	0.1034	0.1316	0.9064	0.8957	0.3358	0.3419	0.0329	0.1017
twin vs father	0.7992	0.8855	0.3483	0.1418	0.3042	0.5983	0.9209	0.82
twin vs parents	0.2644	0.3365	0.5715	0.2663	0.7429	0.7173	0.262	0.2598

[a] Missense variants are rated as damaging when at least two of five prediction algorithms (SIFT (Ng, 2003; Kumar et al., 2009), PolyPhen2 HumDiv, PolyPhen2 HumVar (Adzhubei et al., 2010, 2013), LRT (Chun and Fay, 2009) and MutationTaster (Schwarz JM et al., 2014)) suggesting a potential deleterious effect.

[b] Frameshift and nonsense variants are grouped as loss-of-function variants. [c] MAF- minor allele frequency. The cut-off of the MAF was based on the 1000 Genomes Project (Sudmant et al., 2015) . [d] *p*-value of the Student's t-test.

Table S12. The rare damaging variants in *LAMC1*

Family ID	Mutant	Type [b]	MAF	SIFT [a]	Polyphen2 HDIV [b]	Polyphen2 HVAR [c]	LRT [d]	MutationTaster [e]
B5	p.G170R	PI	NA	D	D	D	D	D
B10	p.D325N	PI	NA	D	D	D	D	D
B8	p.F386Y	PI	0.0002	T	B	B	D	D
LI	p.D532E	PI	0.003395	T	B	B	D	D
B10	p.R557W	MI	0.005391	T	P	B	D	D
B6	p.R557W	PI	0.005391	T	P	B	D	D

We used five programs to predict the potential deleterious effect of the variant. [a] SIFT - D: Deleterious (sift <= 0.05); T: tolerated (sift > 0.05). [b] PolyPhen 2 HDIV - D: Probably damaging (>= 0.957), P: possibly damaging (0.453 ~ 0.956); B: benign (<= 0.452). [c] PolyPhen 2 HVar - D: Probably damaging (>= 0.909), P: possibly damaging (0.447 ~ 0.909); B: benign (<= 0.446). [d] LRT - D: Deleterious; N: Neutral; U: Unknown. [e] MutationTaster - A: disease causing automatic; D: disease causing; N: polymorphism; P: polymorphism automatic..

Table S13. Summary of the rare inherited loss-of-function or damaging variants in SCZ-associated genes in 8 pairs of MZ twins

Family ID	Gene	Mutant	RVIS (%ile) [a]	Type [b]	Class [c]	1000genome MAF	ExAC_EAS MAF
B11	RELN	p.M927V	-2.15(1.46)	MI	I	0.0002	0.00155
B6	MTHFR	p.R46W	0.09(60.71)	MI	I	0.000399	0.00242
B5	PLXNA2	p.P618H	-1.48(3.64)	PI	I	0.001997	0.00827
LI	FEZ1	p.D180V	-0.01(53.51)	PI	I	NA	0.00013
B5	GAD1	p.T27K	-0.51(21.56)	MI	I	0.001797	0.00915
B8	PTPRF	p.R216H	-3.92(0.20)	MI	II	NA	NA
B9	ATP2B4	p.G288V	-0.83(11.51)	PI	II	0.003994	0.01072
B5	DLG2	p.P395S	-1.60(3.04)	MI	II	NA	NA
B5	PTPN11	p.K131R	-0.43(25.15)	PI	II	NA	0.00038
B8	PGAM5	p.R118P	0.17(65.76)	PI	II	NA	NA
B5	MAP1A	p.P2127R	1.16(92.61)	MI	II	NA	NA
B11	MYO5A	p.R609C	-1.41(4.15)	PI	II	0.000599	0.0014
B8	ABR	p.N431S	-1.04(7.80)	MI	II	0.0002	0.00165
B10	DNM2	p.A253V	-1.33(4.71)	MI	II	NA	NA
B9	SYNJ1	p.A512V	0.50(79.67)	MI	II	0.000599	0.00571
B8	ATP5O	p.S91R	-0.05(50.01)	MI	II	NA	0.00051
B5	ITPR1	p.I2030M	-3.35(0.40)	MI	II	0.005591	0.0512
LI	ITPR1	p.I1421V	-3.35(0.40)	MI	II	NA	
B11	IQSEC1	p.R457Q	-1.26(5.34)	MI	II	0.002596	0.0187
B5	SDHA	p.Y55H	-0.86(10.95)	MI	II	0.003195	0.02124
B8	GSN	p.R144Q	0.61(82.94)	MI	II	0.0002	0.00242
B12	GSN	p.K546R	0.61(82.94)	MI	II	0.0002	0.00051
B6	LRRIQ3	p.R525C	1.38(94.60)	MI	III	0.004393	0.00013
B9	LRRIQ3	p.R180*	1.38(94.60)	PI	III	0.000399	0.00013
LI	VPS45	p.Y395C	-0.54(20.54)	MI	III	NA	NA
B5	CLP1	p.R143C	-0.05(50.01)	PI	III	NA	NA
B9	LRP1	p.S2934L	-7.28(0.02)	PI	III	0.000799	0.00064
B6	SHMT2	p.R437H	-0.11(45.26)	MI	III	0.002396	0.02708
B9	BCL11B	p.R319Q	NA	PI	III	NA	NA
LI	PLCB2	p.V356M	-0.70(14.81)	PI	III	0.0002	NA
B12	MAN2A2	p.D891G	-1.45(3.93)	MI	III	0.004193	0.01996
LI	MAN2A2	p.D891G	-1.45(3.93)	PI	III	0.004193	0.01996
B5	GRIN2A	p.R856G	-1.46(3.89)	MI	III	0.000399	0.00013
B5	SEZ6L2	p.A242V	-1.12(6.58)	PI	III	NA	NA
B5	CNOT1	p.N2204S	-1.90(1.97)	MI	III	0.001198	0.00264
B11	THAP11	p.L157V	-0.30(32.62)	MI	III	0.000799	0.00191
B6	CTRL	p.H75Y	0.60(82.66)	MI	III	0.003794	0.01375
B12	NFATC3	p.P297S	0.16(64.96)	MI	III	NA	0.00102
B5	SGSM2	p.G533E	-0.74(13.80)	MI	III	0.001597	0.01329

B10	RAI1	p.P1432L	-3.68(0.25)	PI	III	0.000998	0.00581
B6	SREBF1	p.V610M	-0.79(12.60)	MI	III	0.007388	0.00013
LI	TOM1L2	p.R65T	-1.16(6.17)	MI	III	NA	NA
B6	MYO15A	p.A2248V	-1.26(5.35)	PI	III	0.004393	NA
B12	GATAD2A	p.L207V	-0.95(9.27)	MI	III	0.0002	0.00293
B10	PRRG2	p.S73C	0.66(84.35)	MI	III	0.002396	0.01398
B11	CYP26B1	p.W172R	0.00(54.03)	PI	III	NA	NA
B12	EPC2	p.I461V	-0.29(33.20)	MI	III	NA	NA
B5	PLCL1	p.A809T	-0.24(36.28)	PI	III	NA	NA
B8	C2orf82	p.I98M	NA	PI	III	NA	NA
B9	ACTR5	p.P580L	0.27(70.58)	MI	III	0.001997	0.01659
LI	CYP2D6	p.G169R	1.77(96.77)	PI	III	0.001997	0.01531
B9	TCF20	p.P1240A	-2.55(0.85)	MI	III	NA	0.00102
B9	TRANK1	p.A1422T	-3.56(0.31)	MI	III	0.002196	0.00242
LI	TRANK1	p.V601L	-3.56(0.31)	MI	III	0.005591	0.02346
LI	NISCH	p.R930H	-2.31(1.20)	PI	III	0.00619	0.02747
B8	NISCH	p.Q1463H	-2.31(1.20)	PI	III	0.004593	0.02175
B12	STAB1	p.R2071H	-4.12(0.16)	MI	III	NA	NA
B6	MSL2	p.P25L	-0.53(20.70)	PI	III	0.0002	0.00102
B12	MAN2A1	p.I459M	-1.57(3.20)	PI	III	0.0002	
B6	WDR55	p.S246N	0.46(78.59)	MI	III	0.0002	0.00432
B11	PCDHA5	frameshift	0.10(60.76)	PI	III	NA	NA
B8	PCDHA6	p.N403T	-0.76(13.18)	PI	III	0.00599	0.02365
B12	PCDHA6	p.N403T	-0.76(13.18)	PI	III	0.00599	0.02365
LI	PCDHA10	p.G627V	1.61(95.93)	PI	III	0.000399	0.0028
B5	GALNT10	p.K433R	-1.20(5.79)	PI	III	0.002596	0.00878
B10	RIMS1	p.L1314P	-1.54(3.32)	PI	III	0.001997	0.00106
B10	MPP6	p.L343M	-0.09(46.74)	MI	III	NA	NA

[a] From a distribution of 16,956 genes with pre-calculated respective residual variation intolerance score (RVIS) (Petrovski et al., 2013). [b] MI – inherited from mother; PI – inherited from father. [c] Class I - 38 genes in the SZGR data set (Jia et al., 2010); Class II - 206 genes in SCZ-related pathways or complexes, including PSD-95, ARC, NMDAR network, mGluR5, which were reported in Kirov et al. (2012); class III - 352 genes targeted by the108 SCZ susceptibility loci showing a genome-wide significance (Schizophrenia Working Group of the Psychiatric Genomics, 2014). [d] The minor allele frequency (MAF) from the 1000 Genomes Project (Sudmant et al., 2015). [e] MAF from Exome Aggregation Consortium (ExAC)'s East Asian Studies (EAS) (Lek et al., 2016).

Table S14. Polygenic burden analysis of rare damaging and loss of function variants in schizophrenia-related gene sets

Gene set	Number of genes	Number of mutations		*p*-value [i]	Odd ratio (OR)
		Twins	Parents		
Class I gene					
SZGR core gene [a]	38	10	8	0.647	**1.25287**
Class II gene					
PSD human core [b]	685	148	147	0.953	1.00926
NMDAR network	61	14	10	0.423	**1.40497**
mGluR5	39	12	11	0.838	1.09342
PSD-95	65	14	18	0.596	0.77794
Pre-synapse [b]	431	68	54	0.203	**1.26922**
Pre-synaptic active zone	173	26	21	0.469	**1.24304**
Synaptic vesicle	344	48	38	0.281	**1.27093**
Class III gene					
108 schizophrenia-risk loci [c]	352	92	72	0.115	**1.29131**
Other gene sets					
schizophrenia *de novo* mutations [d]	613	282	282	1.000	1.00212
FMR1 targets [e]	1589	608	584	0.439	1.04926
Axon guidance pathways [f]	432	128	128	1.000	1.00198
Nerve impulse [f]	237	30	37	0.462	0.80996
Nervous system development [f]	1874	506	497	0.712	1.02534
Neuronal cell body [f]	309	66	71	0.731	0.92938
Neuron projection [f]	699	160	150	0.564	1.07355
Cancer pathway [g]	398	104	95	0.520	1.09893
CFG analysis identification [h]	42	16	15	0.860	1.06900
Linkage and association study [h]	223	54	52	0.846	1.04100
Pascal Gene-based test identification [h]	343	54	49	0.622	1.10500
Sherlock analysis identification [h]	12	2	3	1.000	0.66778

[a] Gene set of the SZGR database (Jia et al., 2010). [b] Gene sets in Kirov et al. (2012). [c] Gene set of 108 schizophrenia-associated loci showing a genome-wide significance (Schizophrenia Working Group of the Psychiatric Genomics, 2014). [d] Gene set of nonsynonymous *de novo* mutations reported by Fromer et al. (2014). [e] Genes listed as targets of FMRP (fragile X mental retardation protein) by Darnell et al. (2011) and Ascano et al. (2012). [f] Gene sets reported by Yuen et al. (2015). [g] Gene set of the KEGG database (2000). [h] Gene sets of the SZDB database (http://www.szdb.org/) (Wu et al., 2017). [i] *p*-value of the enrichment of rare variants in each gene set was estimated by the Fisher's exact test.

Table S15. The number of CNVs (> 1 Kb) in 8 families with MZ twins

Family	Father	Mother	Proband	Mean	Total
B10	337	287	292	305.333	916
B11	202	306	248	252	756
B12	198	216	253	222.333	667
B5	307	328	352	329	987
B6	269	266	260	265	795
B8	322	318	273	304.333	913
B9	290	240	280	270	810
LI	212	282	281	258.333	775
Mean	267.125	280.375	279.875	275.792	-
Total	2137	2243	2239	-	6619

Table S16. Summary of the inherited CNV in schizophrenia-associated genes in 8 pairs of MZ twins

Family	CNVs	Gene	Type [a]	Prediction [b]	Reference
B5	chr16:16,934,363-16,940414; Deletion	intergenic	MI or PI	Delly; Lumpy; CNVnator	(Ingason et al., 2011)
B6	chr3:196934573-196939255; Deletion	DLG1 intron	MI	Delly; Lumpy; CNVnator	(Levinson et al., 2011; Walsh et al., 2008)
B11	chr16: 15273991-18185846; Deletion	*MPV17L*, *C16orf45*, *KIAA0430*, *NDE1*, *MYH11*, *C16orf63*, *ABCC1*, *ABCC6*, *NOMO3* and *XYLT1*	PI	Delly; Lumpy; BreakDancer	(Ingason et al., 2011)
LI	chr3:195198880-195223975; Deletion	intergenic	MI	Lumpy; CNVnator	(Levinson et al., 2011; Walsh et al., 2008)

[a] MI – inherited from mother; PI – inherited from father. [b] The CNVs (length > 1000 bp) was calculated by combining the results from four algorithms (Breakdancer (Chen et al., 2009), CNVnator (Abyzov et al., 2011), Delly (Rausch et al., 2012) and Lumpy (Layer et al., 2014)).

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Supplemental Table S4. The distribution of DNMs in 8 pairs of MZ twins

Sample	Chromosome	chromPosition	Allele1	Allele2	Band	dbSNP	Gene	Transcript	Predicted function
B103 B104	chr3	118923152	T	C	q13.32		UPK1B	NM_006952	3utr
B103 B104	chr9	17578171	A	C	p22.2		SH3GL2	NM_003026	5upstream
B103 B104	chr10	71169085	T	G	q22.1		TACR2	NM_001057	intronic
B103 B104	chr11	126855973	C	A	q24.2	rs11220689	KIRREL3	NM_001161707	intronic
B103 B104	chr12	11507171	G	A	p13.2		PRB1	NM_005039	intronic
B103 B104	chr12	104900233	A	G	q23.3		CHST11	NM_001173982	intronic
B103 B104	chr12	133397352	G	A	q24.33		GOLGA3	NM_001172557	intronic
B103 B104	chr13	43170170	G	A	q14.11		TNFSF11	NM_003701	intronic
B103 B104	chr13	74635919	C	T	q22.1	rs114949718	KLF12	NM_007249	intronic
B103 B104	chr15	56398367	T	C	q21.3		RFX7	NM_022841	intronic
B103 B104	chr15	78321335	T	G	q25.1		TBC1D2B	NM_015079	intronic
B103 B104	chr16	19266444	C	T	p12.3		SYT17	NM_016524	intronic
B103 B104	chr17	20145251	C	T	p11.2		SPECC1	NM_001033553	intronic
B103 B104	chr17	66580685	C	T	q24.2		FAM20A	NM_001243746	intronic
B103 B104	chr18	64205104	G	A	q22.1		CDH19	NM_001271028	intronic
B103 B104	chr2	185753314	C	A	q32.1		ZNF804A	NM_194250	intronic
B103 B104	chr2	231640380	T	G	q37.1		CAB39	NM_001130849	intronic
B103 B104	chr20	19700562	G	A	p11.23		SLC24A3	NM_020689	intronic
B103 B104	chr20	34319685	T	G	q11.22		RBM39	NM_001242599	intronic
B103 B104	chr20	34752676	C	T	q11.23		EPB41L1	NM_001258329	intronic
B103 B104	chr20	37200837	G	T	q11.23		RALGAPB	NM_001282917	intronic
B103 B104	chr3	68974336	T	C	p14.1		FAM19A4	NM_001005527	intronic
B103 B104	chr3	89209126	T	G	p11.1		EPHA3	NM_005233	intronic
B103 B104	chr3	178375930	C	T	q26.32		KCNMB2	NM_001278911	intronic
B103 B104	chr5	63649074	T	A	q12.3		RNF180	NM_001113561	intronic
B103 B104	chr8	674731	A	G	p23.3		ERICH1	NM_207332	intronic
B103 B104	chr8	118537055	G	A	q24.11		MED30	NM_001282986	intronic
B103 B104	chr8	121565361	A	G	q24.12		SNTB1	NM_021021	intronic
B103 B104	chr10	128776253	G	T	q26.2		DOCK1	NM_001290223	intronic (splice_site) IVS3+1G>T
B103 B104	chr13	101552351	C	A	q32.3		NALCN_AS1	NR_047687	non-coding intronic
B103 B104	chr14	104320775	G	A	q32.33		LINC00637	NR_038436	non-coding intronic
B103 B104	chr11	94241750	C	A	q21		intergenic		
B103 B104	chr12	42352274	A	C	q12		intergenic		
B103 B104	chr12	58842837	C	G	q14.1		intergenic		
B103 B104	chr12	79083015	G	A	q21.2		intergenic		
B103 B104	chr13	72813354	G	T	q21.33		intergenic		
B103 B104	chr14	78571385	T	C	q24.3	rs177181	intergenic		
B103 B104	chr14	85591083	T	C	q31.3		intergenic		
B103 B104	chr16	63843879	C	T	q21		intergenic		
B103 B104	chr16	69210370	A	C	q22.1		intergenic		
B103 B104	chr16	69210371	C	A	q22.1		intergenic		
B103 B104	chr17	2421858	C	T	p13.3		intergenic		
B103 B104	chr17	41633492	A	C	q21.31	rs112683677	intergenic		
B103 B104	chr18	57721409	A	G	q21.32		intergenic		
B103 B104	chr2	21646773	C	T	p24.1		intergenic		
B103 B104	chr2	82569412	T	C	p12		intergenic		
B103 B104	chr2	88639215	C	T	p11.2		intergenic		
B103 B104	chr3	95967254	T	G	q11.2		intergenic		
B103 B104	chr3	140390293	C	T	q23		intergenic		
B103 B104	chr4	53162451	C	T	q12		intergenic		
B103 B104	chr4	125864692	C	T	q28.1		intergenic		
B103 B104	chr4	162128420	C	T	q32.2		intergenic		
B103 B104	chr5	172158355	G	A	q35.1		intergenic		
B103 B104	chr7	21283412	G	A	p15.3	rs116816019	intergenic		
B103 B104	chr7	152835331	A	G	q36.2		intergenic		
B103 B104	chr8	5214640	G	A	p23.2		intergenic		
B103 B104	chr8	25459305	G	T	p21.2		intergenic		
B103 B104	chr8	47649158	A	G	q11.1		intergenic		
B103 B104	chr8	66834673	A	G	q13.1		intergenic		
B103 B104	chr8	106280832	T	G	q23.1		intergenic		
B103 B104	chr9	11417777	A	T	p23		intergenic		
B103 B104	chr9	12145048	G	A	p23		intergenic		
B103 B104	chrX	20489848	T	C	p22.12		intergenic		
B103 B104	chrX	42471130	T	G	p11.3		intergenic		
B103 B104	chrX	81626563	A	G	q21.1		intergenic		
B103 B104	chr6	78082285	TA	T			intergenic		
B104	chr2	153664053	G	A	q23.3	rs142683553	intergenic		
B113	chr12	26742696	A	G	p11.23	rs11048597	ITPR2	NM_002223	intronic
B113	chr9	127918364	G	A	q33.3		PPP6C	NM_001123355	intronic
B113	chr20	52177664	A	G	q13.2		RP4_724E16.2	NR_110051	non-coding intronic
B113	chr15	96155017	C	A	q26.2		intergenic		
B113	chr4	122576011	T	C	q27		intergenic		
B113 B114	chr19	53997821	C	T	q13.42	rs10423700	ZNF813	NM_001004301	3downstream

B113 B114	chr20	34539772	A	G	q11.23		SCAND1	NM_016558	3downstream
B113 B114	chr3	98242563	T	C	q11.2		CLDND1	NM_001040181	5upstream
B113 B114	chr10	117458502	T	A	q25.3		ATRNL1	NM_207303	intronic
B113 B114	chr11	16923047	T	A	p15.1		PLEKHA7	NM_175058	intronic
B113 B114	chr12	80731159	G	A	q21.31		OTOGL	NM_173591	intronic
B113 B114	chr13	28557037	C	T	q12.2		URAD	NM_001105577	intronic
B113 B114	chr15	62958974	T	C	q22.2		TLN2	NM_015059	intronic
B113 B114	chr16	89961140	C	A	q24.3	rs35518096	TCF25	NM_014972	intronic
B113 B114	chr18	6348505	T	C	p11.31		L3MBTL4	NM_173464	intronic
B113 B114	chr18	32683856	G	C	q12.1		MAPRE2	NM_001143826	intronic
B113 B114	chr2	10036253	C	T	p25.1		TAF1B	NM_005680	intronic
B113 B114	chr3	171864901	G	C	q26.31		FNDC3B	NM_001135095	intronic
B113 B114	chr4	89304876	G	A	q22.1		HERC6	NM_001165136	intronic
B113 B114	chr5	127441266	A	G	q23.3		SLC12A2	NM_001046	intronic
B113 B114	chr9	124373251	T	C	q33.2		DAB2IP	NM_032552	intronic
B113 B114	chr1	101012039	G	T	p21.2		intergenic		
B113 B114	chr10	123074966	T	C	q26.12		intergenic		
B113 B114	chr11	27959616	C	G	p14.1		intergenic		
B113 B114	chr12	103506236	C	T	q23.2		intergenic		
B113 B114	chr13	65852160	C	A	q21.32		intergenic		
B113 B114	chr16	24377252	A	G	p12.1		intergenic		
B113 B114	chr16	86024734	T	C	q24.1	rs12596402	intergenic		
B113 B114	chr17	26144055	A	G	q11.2		intergenic		
B113 B114	chr2	207501019	T	C	q33.3		intergenic		
B113 B114	chr2	220870844	T	C	q35		intergenic		
B113 B114	chr22	49740309	G	C	q13.33		intergenic		
B113 B114	chr3	174392522	T	C	q26.31		intergenic		
B113 B114	chr5	31616208	A	G	p13.3		intergenic		
B113 B114	chr5	118782822	G	A	q23.1	rs56372726	intergenic		
B113 B114	chr6	34401949	G	A	p21.31		intergenic		
B113 B114	chr6	142436831	C	G	q24.1		intergenic		
B113 B114	chr6	145886359	G	C	q24.3		intergenic		
B113 B114	chr7	108858722	C	A	q31.1		intergenic		
B113 B114	chr7	108858732	G	T	q31.1		intergenic		
B113 B114	chr11	33429549	G	GT			intergenic		
B114	chr12	67837996	G	A	q15	rs35359961	intergenic		
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B123 B124	chr19	35449194	T	C	q13.11		ZNF792	NM_175872	nonsyn p.N522S
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B123 B124	chr3	60760326	C	G	p14.2		FHIT	NM_001166243	intronic
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B53 B54	chr17	39595049	G	T	q21.2		KRT38	NM_006771	nonsyn p.P265H	
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B83 B84	chr4	132378202	C	T	q28.3		intergenic		
B83 B84	chr5	5609554	A	C	p15.32		intergenic		
B83 B84	chr5	54356988	C	G	q11.2		intergenic		
B83 B84	chr5	116550935	G	A	q23.1		intergenic		
B83 B84	chr7	138001305	T	C	q33		intergenic		
B83 B84	chr8	138597674	G	C	q24.23		intergenic		
B83 B84	chr9	82567879	T	C	q21.31		intergenic		
B83 B84	chr19	58613970	CAA	C			ZSCAN18	CCDS46214.1	intron
B84	chr13	45830708	C	T	q14.13		GTF2F2	NM_004128	intronic
B84	chr2	167878685	A	G	q24.3		XIRP2	NM_001079810	intronic
B84	chr6	149959476	T	A	q25.1		KATNA1	NM_001204076	intronic
B84	chr7	8681356	T	C	p21.3		NXPH1	NM_152745	intronic
B84	chr9	94497890	T	C	q22.31		ROR2	NM_004560	intronic
B84	chr4	36920104	A	C	p14		intergenic		
B84	chr17	41382052	T	C	q21.31	rs76863987	LINC00854	NR_047479	5upstream
B93 B94	chr7	135414085	A	G	q33		SLC13A4	NM_012450	5upstream
B93 B94	chr10	94297199	C	A	q23.33		IDE	NM_004969	syn
B93 B94	chr16	2147410	G	A	p13.3	rs374486955	PKD1	NM_000296	nonsyn p.R3438W
B93 B94	chr19	57037004	C	G	q13.43		ZNF471	NM_020813	nonsyn p.A523G
B93 B94	chr1	43646521	C	T	p34.2		WDR65	NM_001167965	intronic
B93 B94	chr1	186835728	A	G	q31.1		PLA2G4A	NM_024420	intronic
B93 B94	chr1	224422130	T	C	q42.11		NVL	NM_001243146	intronic
B93 B94	chr10	21979949	C	G	p12.31		MLLT10	NM_001195626	intronic
B93 B94	chr10	131456911	C	T	q26.3		MGMT	NM_002412	intronic

B93 B94	chr11	36428450	G	C	p12		PRR5L	NM_001160167	intronic
B93 B94	chr11	36428499	C	A	p12		PRR5L	NM_001160167	intronic
B93 B94	chr11	83191270	C	A	q14.1		DLG2	NM_001142699	intronic
B93 B94	chr11	131293441	T	C	q25		NTM	NM_001048209	intronic
B93 B94	chr15	39934086	G	A	q14		FSIP1	NM_152597	intronic
B93 B94	chr15	39986654	C	G	q14		FSIP1	NM_152597	intronic
B93 B94	chr16	81098864	G	A	q23.2		C16orf46	NM_001100873	intronic
B93 B94	chr17	49168444	T	C	q21.33		SPAG9	NM_001130527	intronic
B93 B94	chr17	54433363	A	G	q22		ANKFN1	NM_153228	intronic
B93 B94	chr17	54433372	T	C	q22		ANKFN1	NM_153228	intronic
B93 B94	chr17	58787185	G	A	q23.2		BCAS3	NM_001099432	intronic
B93 B94	chr17	65581782	G	A	q24.2		PITPNC1	NM_012417	intronic
B93 B94	chr18	72619462	C	T	q22.3		ZNF407	NM_001146189	intronic
B93 B94	chr2	32706159	A	G	p22.3		BIRC6	NM_016252	intronic
B93 B94	chr2	73229215	T	A	p13.2		SFXN5	NM_144579	intronic
B93 B94	chr2	182975989	A	T	q31.3		PPP1R1C	NM_001080545	intronic
B93 B94	chr2	241957475	G	A	q37.3		SNED1	NM_001080437	intronic
B93 B94	chr3	113657145	G	A	q13.31		GRAMD1C	NM_001172105	intronic
B93 B94	chr4	66240782	A	G	q13.1		EPHA5	NM_001281765	intronic
B93 B94	chr5	65445453	C	G	q12.3		SREK1	NM_001077199	intronic
B93 B94	chr5	65445884	C	G	q12.3		SREK1	NM_001077199	intronic
B93 B94	chr6	139472949	A	G	q24.1		HECA	NM_016217	intronic
B93 B94	chr6	146456876	G	A	q24.3		GRM1	NM_001278064	intronic
B93 B94	chr6	163212623	C	A	q26		PACRG	NM_001080378	intronic
B93 B94	chr7	17914832	C	A	p21.1		SNX13	NM_015132	intronic
B93 B94	chr7	97860074	G	A	q21.3		TECPR1	NM_015395	intronic
B93 B94	chr7	132653469	G	A	q33		CHCHD3	NM_017812	intronic
B93 B94	chr8	22898794	T	C	p21.3		TNFRSF10B	NM_003842	intronic
B93 B94	chr9	77343121	T	A	q21.13		TRPM6	NM_001177310	intronic
B93 B94	chr6	21747135	C	T	p22.3		CASC15	NR_015410	non-coding intronic
B93 B94	chr1	64191122	G	A	p31.3		intergenic		
B93 B94	chr1	151829054	G	C	q21.3		intergenic		
B93 B94	chr10	72842217	A	G	q22.1		intergenic		
B93 B94	chr11	90849241	A	G	q14.3		intergenic		
B93 B94	chr11	116421503	A	T	q23.3		intergenic		
B93 B94	chr12	20304160	A	G	p12.2		intergenic		
B93 B94	chr13	79350867	A	G	q31.1		intergenic		
B93 B94	chr14	64047343	A	G	q23.2		intergenic		
B93 B94	chr15	50440752	G	T	q21.2		intergenic		
B93 B94	chr15	70747621	C	G	q23	rs10220813	intergenic		
B93 B94	chr18	508117	A	C	p11.32		intergenic		
B93 B94	chr18	10325164	C	T	p11.22		intergenic		
B93 B94	chr18	12057323	A	C	p11.21		intergenic		
B93 B94	chr18	73917867	G	A	q23		intergenic		
B93 B94	chr19	21062165	C	T	p12		intergenic		
B93 B94	chr2	6263593	C	T	p25.2		intergenic		
B93 B94	chr2	16519005	G	C	p24.3		intergenic		
B93 B94	chr2	79194279	T	A	p12		intergenic		
B93 B94	chr2	138486744	A	G	q22.1		intergenic		
B93 B94	chr2	156799239	A	G	q24.1		intergenic		
B93 B94	chr2	164338918	G	A	q24.3		intergenic		
B93 B94	chr2	212168526	A	G	q34		intergenic		
B93 B94	chr3	30351562	T	C	p24.1		intergenic		
B93 B94	chr3	46151596	A	G	p21.31		intergenic		
B93 B94	chr3	50574244	T	C	p21.31		intergenic		
B93 B94	chr3	70268930	A	C	p13		intergenic		
B93 B94	chr3	88598299	A	G	p11.1		intergenic		
B93 B94	chr4	5962283	A	G	p16.2		intergenic		
B93 B94	chr4	103002276	C	T	q24		intergenic		
B93 B94	chr4	154787026	A	G	q31.3		intergenic		
B93 B94	chr5	3908422	C	T	p15.33		intergenic		
B93 B94	chr6	165600454	T	C	q27		intergenic		
B93 B94	chr7	19081637	C	A	p21.1		intergenic		
B93 B94	chr7	85163682	G	A	q21.11		intergenic		
B93 B94	chr7	103763484	C	T	q22.1		intergenic		
B93 B94	chr8	41046737	A	T	p11.21		intergenic		
B93 B94	chr9	25091094	G	C	p21.3		intergenic		
B93 B94	chr9	89456759	C	T	q21.33		intergenic		
B93 B94	chrX	55365659	C	A	p11.21		intergenic		
B93 B94	chrX	55595693	C	A	p11.21		intergenic		
B93 B94	chr11	72159268	CT	C		rs141487620	intergenic		
B93 B94	chr16	81278499	TG	T			BCMO1	CCDS10934.1	intron
LJ	chr6	12749681	C	G	p24.1		PHACTR1	NM_001242648	intronic
LJ	chr19	3354318	T	C	p13.3		intergenic		
LJ LS	chr1	7523207	G	A	p36.23		CAMTA1	NM_015215	intronic
LJ LS	chr1	118539438	T	C	p12		SPAG17	NM_206996	intronic

LJ LS	chr10	24822488	T	C	p12.1		KIAA1217	NM_001098500	intronic
LJ LS	chr10	53069364	T	C	q21.1		PRKG1	NM_001098512	intronic
LJ LS	chr10	64995156	A	G	q21.3		JMJD1C	NM_001282948	intronic
LJ LS	chr10	118484331	C	T	q25.3		HSPA12A	NM_025015	intronic
LJ LS	chr11	84078617	G	T	q14.1		DLG2	NM_001142699	intronic
LJ LS	chr11	100687399	G	A	q22.1		ARHGAP42	NM_152432	intronic
LJ LS	chr16	71264288	C	T	q22.2		HYDIN	NM_001198542	intronic
LJ LS	chr19	8468273	C	T	p13.2		RAB11B	NM_004218	intronic
LJ LS	chr19	57113990	G	T	q13.43		ZNF71	NM_021216	intronic
LJ LS	chr2	167170722	C	T	q24.3		SCN9A	NM_002977	intronic
LJ LS	chr3	120315393	G	A	q13.33		NDUFB4	NM_001168331	intronic
LJ LS	chr4	39119053	G	T	p14		KLHL5	NM_001007075	intronic
LJ LS	chr4	115909880	T	A	q26		NDST4	NM_022569	intronic
LJ LS	chr4	115909881	C	A	q26		NDST4	NM_022569	intronic
LJ LS	chr6	37921491	G	A	p21.2		ZFAND3	NM_021943	intronic
LJ LS	chr6	124510527	C	T	q22.31		NKAIN2	NM_001040214	intronic
LJ LS	chr6	124515065	A	T	q22.31		NKAIN2	NM_001040214	intronic
LJ LS	chr7	20427755	C	T	p21.1		ITGB8	NM_002214	intronic
LJ LS	chr7	50434049	G	A	p12.2		IKZF1	NM_001220765	intronic
LJ LS	chr8	3381974	C	A	p23.2		CSMD1	NM_033225	intronic
LJ LS	chr9	105381829	C	T	q31.1		LINC00587	NR_103830	non-coding intronic
LJ LS	chr1	79265388	C	T	p31.1		intergenic		
LJ LS	chr10	73143664	G	A	q22.1		intergenic		
LJ LS	chr11	97265884	G	C	q22.1		intergenic		
LJ LS	chr12	105193487	A	G	q23.3	rs77594953	intergenic		
LJ LS	chr13	73798380	G	A	q22.1		intergenic		
LJ LS	chr14	39073589	G	C	q21.1		intergenic		
LJ LS	chr16	47797029	T	C	q12.1		intergenic		
LJ LS	chr16	76707882	C	T	q23.1		intergenic		
LJ LS	chr17	13733612	C	T	p12		intergenic		
LJ LS	chr18	38732423	A	G	q12.3		intergenic		
LJ LS	chr2	146609689	G	A	q22.3		intergenic		
LJ LS	chr2	184856690	T	C	q32.1		intergenic		
LJ LS	chr2	186167711	T	G	q32.1		intergenic		
LJ LS	chr2	186167712	A	T	q32.1		intergenic		
LJ LS	chr2	218660590	C	G	q35		intergenic		
LJ LS	chr2	238513142	C	T	q37.3	rs59237394	intergenic		
LJ LS	chr20	50688780	G	T	q13.2		intergenic		
LJ LS	chr3	36418949	C	T	p22.3		intergenic		
LJ LS	chr3	155766601	G	A	q25.31		intergenic		
LJ LS	chr4	27144101	G	A	p15.2		intergenic		
LJ LS	chr4	70243771	G	C	q13.2		intergenic		
LJ LS	chr4	129175380	T	G	q28.2		intergenic		
LJ LS	chr7	36863766	C	T	p14.2		intergenic		
LJ LS	chr7	54008241	C	T	p11.2		intergenic		
LJ LS	chr9	15398550	G	C	p22.3		intergenic		
LJ LS	chr9	134281048	C	T	q34.13		intergenic		
LJ LS	chrY	17459241	A	G	q11.221		intergenic		
LJ LS	chrX	93019398	CAG	C			intergenic		
LS	chr12	123034737	A	T	q24.31	rs12581072	KNTC1	NM_014708	intronic
LS	chr2	174321128	C	G	q31.1	rs13021431	intergenic		

Supplemental Table S7. The regulatory elements hit by *de novo* SNVs in 8 pairs of MZ twins

Sample	Chromosome	chromPosition	Allele1	Allele2	Transition/Transversion	Gene	Predicted function	CpG_site	CpG_island	Score*	Regulatory elements hits
B93 B94	chr15	50440752	G	T	transversion	intergenic		CpG_site	NA	2a	Motifs|PWM||Stra13, Motifs|Footprinting|AosmcSerumfree|Stra13, Motifs|Footprinting|AosmcSerumfree|USF, Motifs|Footprinting|Mcf7Hypoxlac|Stra13, Motifs|Footprinting|A549|Stra13, Motifs|Footprinting|Mcf7|USF, Motifs|Footprinting|Mcf7Hypoxlac|USF, Motifs|Footprinting|H9es|Stra13, Motifs|Footprinting|A549|, Motifs|Footprinting|SkMC|, Motifs|Footprinting|K562|, Motifs|PWM||CREB3L1, Motifs|PWM||USF, Motifs|Footprinting|Mcf7|Stra13, Motifs|Footprinting|A549|USF, Chromatin_Structure||DNase-seq|Progfib, Chromatin_Structure||DNase-seq|Nb4, Chromatin_Structure|Diffa14d|DNase-seq|H7es, Chromatin_Structure||DNase-seq|Ipsnihi7, Chromatin_Structure||DNase-seq|Htr8, Chromatin_Structure||DNase-seq|Hpaec, Chromatin_Structure||DNase-seq|Hpdlf, Chromatin_Structure|Znfb34a8|DNase-seq|K562, Chromatin_Structure||DNase-seq|Fibropag08395, Chromatin_Structure||DNase-seq|Hmvecdneo, Chromatin_Structure||DNase-seq|Hpde6e6e7, Chromatin_Structure||DNase-seq|Huvec, Chromatin_Structure||DNase-seq|Hrpe, Chromatin_Structure|Znfe103c6|DNase-seq|K562, Chromatin_Structure|Est10nm30m|DNase-seq|Ishikawa, Chromatin_Structure||DNase-seq|Ipsnihi11, Chromatin_Structure||DNase-seq|Hcf, Chromatin_Structure|Ifng4h|FAIRE|Helas3, Chromatin_Structure||DNase-seq|Ag09319, Chromatin_Structure||DNase-seq|Panisd, Chromatin_Structure||DNase-seq|Prec, Chromatin_Structure||DNase-seq|Fibroblgm03348, Chromatin_Structure|Est100nm1h|DNase-seq|Mcf7, Chromatin_Structure|Znf4c50c4|DNase-seq|K562, Chromatin_Structure||DNase-seq|Hcpe, Chromatin_Structure||DNase-seq|Ag10803, Chromatin_Structure|Hypoxlaccon|DNase-seq|Mcf7, Chromatin_Structure||DNase-seq|Rptec, Chromatin_Structure||DNase-seq|Gm12864, Chromatin_Structure||DNase-seq|M059j, Chromatin_Structure||DNase-seq|Lncap, Chromatin_Structure||DNase-seq|Nt2d1, Chromatin_Structure||DNase-seq|K562G2mphase, Chromatin_Structure||DNase-seq|Gm04504, Chromatin_Structure||DNase-seq|Hs5, Chromatin_Structure|Nabut|FAIRE|K562, Chromatin_Structure||FAIRE|Nhek, Chromatin_Structure||DNase-seq|Hcfaa, Chromatin_Structure||DNase-seq|Hmveclbl, Chromatin_Structure||DNase-seq|Hah, Chromatin_Structure||DNase-seq|Hpf, Chromatin_Structure||DNase-seq|Wi38, Chromatin_Structure|Ohurea|FAIRE|K562, Chromatin_Structure||DNase-seq|Hvmf, Chromatin_Structure||DNase-seq|Hrgec, Chromatin_Structure||DNase-seq|Stellate, Chromatin_Structure||DNase-seq|Hffmyc, Chromatin_Structure||DNase-seq|Th17, Chromatin_Structure||DNase-seq|Hmvecdlyneo, Chromatin_Structure||DNase-

B93 B94	chr16	2147410	G	A	transition		PKD1	nonsyn p.R3438W	CpG_site	NA	2b	Motifs|Footprinting|Huh75|TLX1::NFIC, Motifs|Footprinting|AosmcSerumfree|TLX1::NFIC, Motifs|Footprinting|Melano|CTF1, Motifs|Footprinting|Myometr|myogenin/NF-1, Motifs|Footprinting|H1hesc|TLX1::NFIC, Motifs|Footprinting|Htr8|CTF1, Motifs|Footprinting|Panislets|CTF1, Motifs|Footprinting|Myometr|CTF1, Motifs|Footprinting|Gm19239|CTF1, Motifs|Footprinting|Osteobl|CTF1, Motifs|Footprinting|Phte|TLX1::NFIC, Motifs|PWM||myogenin/NF-1, Motifs|Footprinting|Mcf7|TLX1::NFIC, Motifs|Footprinting|Gm12891|myogenin/NF-1, Motifs|Footprinting|Chorion|CTF1, Motifs|Footprinting|Gm18507|TLX1::NFIC, Motifs|Footprinting|K562|TLX1::NFIC, Motifs|Footprinting|Panislets|myogenin/NF-1, Motifs|Footprinting|Gm12892|myogenin/NF-1, Motifs|Footprinting|Hmec|TLX1::NFIC, Motifs|Footprinting|Fibrop|TLX1::NFIC, Motifs|Footprinting|A549|CTF1, Motifs|Footprinting|H1hesc|CTF1, Motifs|Footprinting|Fibrobl|TLX1::NFIC, Motifs|Footprinting|Gm18507|CTF1, Motifs|Footprinting|Panislets|TLX1::NFIC, Motifs|Footprinting|Mcf7Hypoxlac|TLX1::NFIC, Motifs|Footprinting|Huvec|CTF1, Motifs|Footprinting|Gm12892|TLX1::NFIC, Motifs|Footprinting|Huh7|TLX1::NFIC, Motifs|Footprinting|Lncap|myogenin/NF-1, Motifs|Footprinting|Huh75|CTF1, Motifs|Footprinting|Mcf7|CTF1, Motifs|Footprinting|Hepatocytes|CTF1, Motifs|Footprinting|Hsmm|myogenin/NF-1, Motifs|Footprinting|Helas3|TLX1::NFIC, Motifs|Footprinting|Chorion|TLX1::NFIC, Motifs|Footprinting|Gliobla|CTF1, Motifs|Footprinting|AosmcSerumfree|CTF1, Motifs|Footprinting|Hpde6e6e7|CTF1, Motifs|Footprinting|Hepg2|TLX1::NFIC, Motifs|PWM||NFIX, Motifs|Footprinting|LncapAndro|myogenin/NF-1, Motifs|Footprinting|Gm19239|TLX1::NFIC, Motifs|Footprinting|Medullo|TLX1::NFIC, Motifs|Footprinting|Gm12892|CTF1, Motifs|Footprinting|Lncap|CTF1, Motifs|Footprinting|Cll|TLX1::NFIC, Motifs|Footprinting|Hsmmt|myogenin/NF-1, Motifs|Footprinting|Hsmm|TLX1::NFIC, Motifs|Footprinting|A549|TLX1::NFIC, Motifs|Footprinting|Myometr|TLX1::NFIC, Motifs|Footprinting|Mcf7Hypoxlac|CTF1, Motifs|Footprinting|Hmec|CTF1, Motifs|PWM||CTF1, Motifs|Footprinting|8988t|myogenin/NF-1, Motifs|PWM||NFIB, Motifs|Footprinting|Hepg2|CTF1, Motifs|Footprinting|Phte|CTF1, Motifs|Footprinting|Huh7|CTF1,

B83 B84	chr19	4796824	G	A	transition	FEM1A	3downstream	CpG_site	NA	2b	Motifs|Footprinting|Medullo|Olf-1, Motifs|PWM||CAC-bindingprotein, Motifs|Footprinting|H1hesc|Olf-1, Motifs|Footprinting|Gm12891|Olf-1, Motifs|PWM||Olf-1, Motifs|Footprinting|Gm12892|Olf-1, Chromatin_Structure||DNase-seq|Hrpe, Chromatin_Structure||DNase-seq|Medullod341, Chromatin_Structure||DNase-seq|Skmc, Chromatin_Structure||DNase-seq|Osteobl, Chromatin_Structure|Diff4d|DNase-seq|Lhcnm2, Chromatin_Structure||DNase-seq|Lhcnm2, Chromatin_Structure||DNase-seq|Medullo, Protein_Binding||ChIP-seq|GM12878|POLR2A, Protein_Binding||ChIP-seq|K562|POLR2A, Protein_Binding||ChIP-seq|GM12892|POLR2A, Protein_Binding||ChIP-
B83 B84	chr20	9966603	G	A	transition	LOC101929371	5upstream	NA	NA	2b	Motifs|PWM||IRF-7, Motifs|Footprinting|Hpde6e6e7|IRF-7, Chromatin_Structure||DNase-seq|Saec, Chromatin_Structure|Estctrl0h|DNase-seq|Mcf7, Chromatin_Structure||DNase-seq|Mcf7, Chromatin_Structure||DNase-seq|Hcf, Chromatin_Structure||DNase-seq|Hmec, Chromatin_Structure|Est100nm1h|DNase-seq|Mcf7, Chromatin_Structure||DNase-seq|Ag10803, Chromatin_Structure||DNase-seq|Hvmf, Chromatin_Structure||DNase-seq|Hcm, Chromatin_Structure||DNase-seq|Hsmm, Chromatin_Structure||DNase-seq|Hpdlf, Chromatin_Structure||DNase-seq|Hee, Chromatin_Structure||DNase-seq|Hmf, Chromatin_Structure||DNase-seq|Hae, Chromatin_Structure||DNase-seq|Skmc, Chromatin_Structure||DNase-seq|Hgf, Protein_Binding|01pct|ChIP-seq|MCF10A-Er-Src|STAT3, Protein_Binding|01pct|ChIP-seq|MCF10A-Er-Src|MYC, Protein_Binding|4ohtam_1um_4hr|ChIP-seq|MCF10A-Er-Src|FOS, Protein_Binding|01pct_4hr|ChIP-seq|MCF10A-Er-Src|STAT3, Protein_Binding|4ohtam_1um_36hr|ChIP-seq|MCF10A-Er-Src|STAT3
B62	chr7	27657582	G	A	transition	HIBADH	intronic	CpG_site	NA	2b	Motifs|Footprinting|Hepg2|STAT5A(homotetramer), Motifs|PWM||STAT5A(homotetramer), Chromatin_Structure|Ifng4h|FAIRE|Helas3, Chromatin_Structure||DNase-seq|Huh7, Chromatin_Structure||DNase-seq|Hepg2, Chromatin_Structure||FAIRE|Hepg2, Protein_Binding|forskolin|ChIP-seq|HepG2|POLR2A, Protein_Binding||ChIP-seq|HepG2|HDAC2, Protein_Binding||ChIP-seq|HepG2|SP1, Protein_Binding||ChIP-seq|HepG2|HNF4A, Protein_Binding||ChIP-seq|HepG2|YY1, Protein_Binding|forskolin|ChIP-seq|HepG2|HNF4A, Protein_Binding||ChIP-seq|NB4|MYC, Protein_Binding||ChIP-seq|HepG2|HNF4G, Protein_Binding||ChIP-seq|HepG2|POLR2A,

B53 B54	chr17	41382052	T	C	transition		LINC00854	5upstream	NA	NA	2b	Motifs|Footprinting|K562|, Motifs|PWM||EBF1, Chromatin_Structure||DNase-seq|Progfib, Chromatin_Structure||DNase-seq|Ipsnihi7, Chromatin_Structure||DNase-seq|Htr8, Chromatin_Structure||DNase-seq|Hpdlf, Chromatin_Structure||DNase-seq|Fibropag08395, Chromatin_Structure||DNase-seq|Hpde6e6e7, Chromatin_Structure||DNase-seq|Huvec, Chromatin_Structure||FAIRE|UrotsaUt189, Chromatin_Structure|Est10nm30m|DNase-seq|Ishikawa, Chromatin_Structure||DNase-seq|Cd20ro01794, Chromatin_Structure||DNase-seq|Heartoc, Chromatin_Structure||DNase-seq|Cd4naivewb11970640, Chromatin_Structure||DNase-seq|Hcf, Chromatin_Structure|Ifng4h|FAIRE|Helas3, Chromatin_Structure||DNase-seq|UrotsaUt189, Chromatin_Structure||DNase-seq|Panisd, Chromatin_Structure||DNase-seq|Fibroblgm03348, Chromatin_Structure||DNase-seq|Sknsh, Chromatin_Structure|Hypoxlaccon|DNase-seq|Mcf7, Chromatin_Structure||FAIRE|Gm12878, Chromatin_Structure||FAIRE|Nhbe, Chromatin_Structure||DNase-seq|Lncap, Chromatin_Structure||FAIRE|Htr8, Chromatin_Structure||DNase-seq|K562G2mphase, Chromatin_Structure||DNase-seq|Cerebrumfrontaloc, Chromatin_Structure||DNase-seq|Gm04504, Chromatin_Structure|Nabut|FAIRE|K562, Chromatin_Structure||FAIRE|Nhek, Chromatin_Structure||DNase-seq|Hcfaa, Chromatin_Structure||DNase-seq|Gm12891, Chromatin_Structure||DNase-seq|8988t, Chromatin_Structure||DNase-seq|Gm19240, Chromatin_Structure||DNase-seq|Melano, Chromatin_Structure||DNase-seq|Wi38, Chromatin_Structure|Ohurea|FAIRE|K562, Chromatin_Structure||DNase-seq|Hrgec, Chromatin_Structure||DNase-seq|Olfneurosphere, Chromatin_Structure||DNase-seq|Stellate, Chromatin_Structure|Ctcfshrna|DNase-seq|Mcf7, Chromatin_Structure||DNase-seq|Gm10248, Chromatin_Structure||FAIRE|Medullo, Chromatin_Structure||DNase-seq|Hae, Chromatin_Structure||DNase-seq|Ipscwru1, Chromatin_Structure||DNase-seq|Th2, Chromatin_Structure||FAIRE|Gm12892, Chromatin_Structure||DNase-seq|Medullod341, Chromatin_Structure||DNase-seq|Gm20000, Chromatin_Structure||DNase-seq|H7es, Chromatin_Structure||DNase-seq|Hsmm, Chromatin_Structure||DNase-seq|Fibropag08396, Chromatin_Structure||DNase-seq|Monocd14, Chromatin_Structure||DNase-seq|Tregwb78495824, Chromatin_Structure||DNase-seq|Cd4naivewb78495824, Chromatin_Structure||DNase-seq|Hasp, Chromatin_Structure||DNase-seq|Panislets, Chromatin_Structure||DNase-seq|Gm13976, Chromatin_Structure||DNase-seq|Saec,

B84	chr17	41382052	T	C	transition		LINC00854	5upstream	NA	NA	2b	Motifs|Footprinting|K562|, Motifs|PWM||EBF1, Chromatin_Structure||DNase-seq|Progfib, Chromatin_Structure||DNase-seq|Ipsnihi7, Chromatin_Structure||DNase-seq|Htr8, Chromatin_Structure||DNase-seq|Hpdlf, Chromatin_Structure||DNase-seq|Fibropag08395, Chromatin_Structure||DNase-seq|Hpde6e6e7, Chromatin_Structure||DNase-seq|Huvec, Chromatin_Structure||FAIRE|UrotsaUt189, Chromatin_Structure|Est10nm30m|DNase-seq|Ishikawa, Chromatin_Structure||DNase-seq|Cd20ro01794, Chromatin_Structure||DNase-seq|Heartoc, Chromatin_Structure||DNase-seq|Cd4naivewb11970640, Chromatin_Structure||DNase-seq|Hcf, Chromatin_Structure|Ifng4h|FAIRE|Helas3, Chromatin_Structure||DNase-seq|UrotsaUt189, Chromatin_Structure||DNase-seq|Panisd, Chromatin_Structure||DNase-seq|Fibroblgm03348, Chromatin_Structure||DNase-seq|Sknsh, Chromatin_Structure|Hypoxlaccon|DNase-seq|Mcf7, Chromatin_Structure||FAIRE|Gm12878, Chromatin_Structure||FAIRE|Nhbe, Chromatin_Structure||DNase-seq|Lncap, Chromatin_Structure||FAIRE|Htr8, Chromatin_Structure||DNase-seq|K562G2mphase, Chromatin_Structure||DNase-seq|Cerebrumfrontaloc, Chromatin_Structure||DNase-seq|Gm04504, Chromatin_Structure|Nabut|FAIRE|K562, Chromatin_Structure||FAIRE|Nhek, Chromatin_Structure||DNase-seq|Hcfaa, Chromatin_Structure||DNase-seq|Gm12891, Chromatin_Structure||DNase-seq|8988t, Chromatin_Structure||DNase-seq|Gm19240, Chromatin_Structure||DNase-seq|Melano, Chromatin_Structure||DNase-seq|Wi38, Chromatin_Structure|Ohurea|FAIRE|K562, Chromatin_Structure||DNase-seq|Hrgec, Chromatin_Structure||DNase-seq|Olfneurosphere, Chromatin_Structure||DNase-seq|Stellate, Chromatin_Structure|Ctcfshrna|DNase-seq|Mcf7, Chromatin_Structure||DNase-seq|Gm10248, Chromatin_Structure||FAIRE|Medullo, Chromatin_Structure||DNase-seq|Hae, Chromatin_Structure||DNase-seq|Ipscwru1, Chromatin_Structure||DNase-seq|Th2, Chromatin_Structure||FAIRE|Gm12892, Chromatin_Structure||DNase-seq|Medullod341, Chromatin_Structure||DNase-seq|Gm20000, Chromatin_Structure||DNase-seq|H7es, Chromatin_Structure||DNase-seq|Hsmm, Chromatin_Structure||DNase-seq|Fibropag08396, Chromatin_Structure||DNase-seq|Monocd14, Chromatin_Structure||DNase-seq|Tregwb78495824, Chromatin_Structure||DNase-seq|Cd4naivewb78495824, Chromatin_Structure||DNase-seq|Hasp, Chromatin_Structure||DNase-seq|Panislets, Chromatin_Structure||DNase-seq|Gm13976, Chromatin_Structure||DNase-seq|Saec,

B103 B104	chr18	57721409	A	G	transition	intergenic	NA	NA	2b	Motifs|Footprinting|Hpde6e6e7|Foxq1, Motifs|Footprinting|AosmcSerumfree|Foxq1, Motifs|Footprinting|Phte|Foxq1, Motifs|PWM|Foxq1, Motifs|Footprinting|Helas3Ifna4h|Foxq1, Motifs|Footprinting|Helas3|Foxq1, Motifs|Footprinting|Panisd|Foxq1, Chromatin_Structure|DNase-seq|Hpde6e6e7, Chromatin_Structure|Ifng4h|FAIRE|Helas3, Chromatin_Structure|FAIRE|Nhek, Chromatin_Structure|DNase-seq|Hah, Chromatin_Structure|DNase-seq|Wi38, Chromatin_Structure|Ctcfshrna|DNase-seq|Mcf7, Chromatin_Structure|FAIRE|Medullo, Chromatin_Structure|DNase-seq|Hsmm, Chromatin_Structure|FAIRE|Gliobla, Chromatin_Structure|DNase-seq|K562, Chromatin_Structure|FAIRE|Helas3, Chromatin_Structure|DNase-seq|Mcf7, Chromatin_Structure|DNase-seq|Fibrop, Chromatin_Structure|DNase-seq|A549, Chromatin_Structure|DNase-seq|Helas3, Chromatin_Structure|Ifna4h|DNase-seq|Helas3, Chromatin_Structure|DNase-seq|Nha, Chromatin_Structure|FAIRE|K562, Chromatin_Structure|DNase-seq|Phte, Chromatin_Structure|DNase-seq|Lhcnm2, Chromatin_Structure|DNase-seq|Hsmmemb, Chromatin_Structure|DNase-seq|Fibropag20443, Chromatin_Structure|DNase-seq|Nhlf, Chromatin_Structure|Znfg54a11|DNase-seq|K562, Chromatin_Structure|DNase-seq|Aoaf, Protein_Binding|ChIP-seq|HeLa-

B53 B54	chr15	68647297	T	A	transversion	ITGA11	intronic	NA	NA	2b	Motifs|Footprinting|Helas3Ifna4h|Ik-1, Motifs|Footprinting|Fibrop|Ik-1, Motifs|Footprinting|Hpde6e6e7|Ik-1, Motifs|Footprinting|AosmcSerumfree|Ik-1, Motifs|PWM|Ik-1, Motifs|Footprinting|Htr8|Ik-1, Motifs|Footprinting|Hsmm|Ik-1, Motifs|Footprinting|Fibrobl|Ik-1, Motifs|Footprinting|Hmec|Ik-1, Motifs|Footprinting|Panisd|Ik-1, Motifs|Footprinting|8988t|Ik-1, Motifs|Footprinting|Gliobla|Ik-1, Motifs|Footprinting|A549|Ik-1, Motifs|Footprinting|Myometr|Ik-1, Motifs|Footprinting|Huvec|Ik-1, Motifs|Footprinting|Helas3|Ik-1, Chromatin_Structure|DNase-seq|Progfib, Chromatin_Structure|DNase-seq|Ipsnihi7, Chromatin_Structure|DNase-seq|Htr8, Chromatin_Structure|DNase-seq|Fibropag08395, Chromatin_Structure|DNase-seq|Monocd14ro1746, Chromatin_Structure|DNase-seq|Hpde6e6e7, Chromatin_Structure|DNase-seq|Huvec, Chromatin_Structure|FAIRE|UrotsaUt189, Chromatin_Structure|Znfe103c6|DNase-seq|K562, Chromatin_Structure|Ifng4h|FAIRE|Helas3, Chromatin_Structure|DNase-seq|UrotsaUt189, Chromatin_Structure|DNase-seq|Panisd, Chromatin_Structure|DNase-seq|Fibroblgm03348, Chromatin_Structure|Est100nm1h|DNase-seq|Mcf7, Chromatin_Structure|FAIRE|Nhbe, Chromatin_Structure|DNase-seq|Hs5, Chromatin_Structure|DNase-seq|Hmvecdad, Chromatin_Structure|DNase-seq|Olfneurosphere, Chromatin_Structure|DNase-seq|Stellate, Chromatin_Structure|Serumfree|DNase-seq|Aosmc, Chromatin_Structure|DNase-seq|Hsmm, Chromatin_Structure|DNase-seq|Fibropag08396, Chromatin_Structure|DNase-seq|Monocd14, Chromatin_Structure|Lenticon|DNase-seq|Fibroblgm03348, Chromatin_Structure|Ifna4h|FAIRE|Helas3, Chromatin_Structure|DNase-seq|Fibrobl, Chromatin_Structure|DNase-seq|K562, Chromatin_Structure|FAIRE|Urotsa, Chromatin_Structure|DNase-seq|Hl60, Chromatin_Structure|DNase-seq|Ips, Chromatin_Structure|DNase-seq|Mcf7, Chromatin_Structure|DNase-seq|Fibrop, Chromatin_Structure|DNase-seq|Nhek, Chromatin_Structure|DNase-seq|Gliobla, Chromatin_Structure|DNase-seq|A549, Chromatin_Structure|DNase-seq|Helas3, Chromatin_Structure|Ifna4h|DNase-seq|Helas3, Chromatin_Structure|DNase-seq|Urotsa, Chromatin_Structure|DNase-seq|Rwpe1, Chromatin_Structure|DNase-seq|Hmec, Chromatin_Structure|DNase-seq|Imr90, Chromatin_Structure|Lentimyod|DNase-seq|Fibroblgm03348, Chromatin_Structure|FAIRE|Astrocy, Chromatin_Structure|DNase-seq|Hsmmfshd, Chromatin_Structure|DNase-seq|Hsmmemb, Chromatin_Structure|DNase-
B103 B104	chr16	69210370	A	C	transversion	intergenic		NA	NA	2b	Motifs|Footprinting|SkMC|, Motifs|PWM|RREB1, Motifs|Footprinting|AosmcSerumfree|RREB1, Chromatin_Structure|DNase-seq|Fibropag08395, Chromatin_Structure|DNase-seq|Ag09319, Chromatin_Structure|DNase-seq|Rptec, Chromatin_Structure|DNase-seq|Hffmyc, Chromatin_Structure|DNase-seq|Hff, Chromatin_Structure|DNase-seq|Fibrop, Chromatin_Structure|DNase-seq|Hgf, Protein_Binding|ChIP-seq|AG04450|CTCF, Protein_Binding|4ohtam_1um_12hr|ChIP-seq|MCF10A-Er-Src|STAT3,

B103 B104	chr16	69210371	C	A	transversion	intergenic		NA	NA	2b	Motifs|Footprinting|SkMC|, Motifs|PWM||RREB1, Motifs|Footprinting|AosmcSerumfree|RREB1, Chromatin_Structure||DNase-seq|Fibropag08395, Chromatin_Structure||DNase-seq|Ag09319, Chromatin_Structure||DNase-seq|Rptec, Chromatin_Structure||DNase-seq|Hffmyc, Chromatin_Structure||DNase-seq|Hff, Chromatin_Structure||DNase-seq|Fibrop, Chromatin_Structure||DNase-seq|Hgf, Protein_Binding||ChIP-seq|AG04450|CTCF, Protein_Binding|4ohtam_1um_12hr|ChIP-seq|MCF10A-Er-Src|STAT3,
B83 B84	chr8	125660326	T	C	transition	MTSS1	intronic	NA	NA	2b	Motifs|Footprinting|Mcf7|IPF1, Motifs|Footprinting|Htr8|IPF1, Motifs|PWM||IPF1, Motifs|Footprinting|Gm12878|IPF1, Chromatin_Structure||DNase-seq|Htr8, Chromatin_Structure||DNase-seq|Huvec, Chromatin_Structure||FAIRE|Htr8, Protein_Binding||ChIP-seq|HFF-Myc|CTCF, Protein_Binding||ChIP-seq|Osteobl|CTCF, Protein_Binding||ChIP-seq|K562|CTCF

B123 B124	chr12	129314669	A	G	transition	intergenic	NA	NA	2b	Motifs|Footprinting|Gm18507|Pax-8, Motifs|Footprinting|Cll|Pax-8, Motifs|Footprinting|Gm12878|Pax-8, Motifs|PWM|Pax-8, Motifs|Footprinting|Medullo|Pax-8, Motifs|PWM|Duxl, Motifs|Footprinting|Gm19238|Pax-8, Motifs|Footprinting|Gm12892|Pax-8, Motifs|Footprinting|Gm12891|Pax-8, Motifs|Footprinting|Hsmm|Pax-8, Motifs|PWM|FOXJ2, Motifs|Footprinting|Hsmmt|Pax-8, Motifs|Footprinting|Gm19239|Pax-8, Chromatin_Structure|DNase-seq|Nb4, Chromatin_Structure|DNase-seq|Monocd14ro1746, Chromatin_Structure|DNase-seq|Cd20ro01794, Chromatin_Structure|DNase-seq|Prec, Chromatin_Structure|DNase-seq|Fibroblgm03348, Chromatin_Structure|FAIRE|Gm12878, Chromatin_Structure|DNase-seq|Cd34mobilized, Chromatin_Structure|DNase-seq|Gm12891, Chromatin_Structure|DNase-seq|Gm19240, Chromatin_Structure|DNase-seq|Wi38, Chromatin_Structure|DNase-seq|Th17, Chromatin_Structure|DNase-seq|Gm10248, Chromatin_Structure|FAIRE|Gm18507, Chromatin_Structure|DNase-seq|Th2, Chromatin_Structure|FAIRE|Gm12892, Chromatin_Structure|DNase-seq|Medullod341, Chromatin_Structure|DNase-seq|Gm20000, Chromatin_Structure|DNase-seq|Hsmm, Chromatin_Structure|DNase-seq|Monocd14, Chromatin_Structure|DNase-seq|Hmf, Chromatin_Structure|DNase-seq|Skmc, Chromatin_Structure|DNase-seq|Th2wb54553204, Chromatin_Structure|DNase-seq|Gm13976, Chromatin_Structure|DNase-seq|Adultcd4th0, Chromatin_Structure|DNase-seq|Fibrobl, Chromatin_Structure|DNase-seq|Th1wb33676984, Chromatin_Structure|FAIRE|Gm19239, Chromatin_Structure|DNase-seq|K562, Chromatin_Structure|DNase-seq|Adultcd4th1, Chromatin_Structure|DNase-seq|Gcbcell, Chromatin_Structure|DNase-seq|Werirb1, Chromatin_Structure|DNase-seq|Cll, Chromatin_Structure|DNase-seq|Hl60, Chromatin_Structure|DNase-seq|Gm10266, Chromatin_Structure|DNase-seq|T47d, Chromatin_Structure|DNase-seq|A549, Chromatin_Structure|DNase-seq|Gm19238, Chromatin_Structure|DNase-seq|Hsmmt, Chromatin_Structure|DNase-seq|Urotsa, Chromatin_Structure|DNase-seq|Gm19239, Chromatin_Structure|DNase-seq|Naivebcell, Chromatin_Structure|DNase-seq|Hee, Chromatin_Structure|Ohtam|DNase-seq|Wi38, Chromatin_Structure|Diff4d|DNase-seq|Lhcnm2, Chromatin_Structure|DNase-seq|Gm13977, Chromatin_Structure|Lentimyod|DNase-seq|Fibroblgm03348, Chromatin_Structure|DNase-seq|Lhcnm2, Chromatin_Structure|DNase-

B93 B94	chr10	131456911	C	T	transition	MGMT	intronic	NA	NA	2b	Motifs|Footprinting|Huh7|Gfi1, Motifs|Footprinting|Mcf7|Gfi1, Motifs|Footprinting|Huh7|Gfi1b, Motifs|PWM||Gfi1b, Motifs|Footprinting|Mcf7|Gfi-1, Motifs|PWM||Gfi1, Motifs|PWM||Gfi, Motifs|Footprinting|Huh7|Gfi, Motifs|Footprinting|Cll|Gfi1, Motifs|Footprinting|Cll|Gfi1b, Motifs|PWM||Gfi-1, Motifs|Footprinting|Mcf7|Gfi, Motifs|Footprinting|Mcf7|Gfi1b, Motifs|Footprinting|Huh7|Gfi-1, Chromatin_Structure||DNase-seq|Ips, Chromatin_Structure||DNase-seq|Mcf7, Chromatin_Structure||FAIRE|H1hesc, Chromatin_Structure||DNase-seq|Ipsnihi7, Chromatin_Structure||DNase-seq|Ag09319, Chromatin_Structure||DNase-seq|Huh7, Chromatin_Structure||DNase-seq|Nhdfad, Chromatin_Structure||DNase-seq|Hgf,
B124	chr1	230530315	T	C	transition	PGBD5	intronic	NA	NA	2b	Motifs|PWM||SP4, Motifs|Footprinting|A549|SP1, Motifs|PWM||SP1, Motifs|PWM||Zfp410, Motifs|PWM||Bcl6b, Motifs|PWM||ZNF219, Motifs|Footprinting|Lncap|ZNF219, Motifs|PWM||Sp1, Motifs|Footprinting|Lncap|CAC-bindingprotein, Motifs|PWM||Zfp740, Motifs|PWM||CAC-bindingprotein, Motifs|PWM||Zfp281, Chromatin_Structure|Est10nm30m|DNase-seq|T47d, Chromatin_Structure||DNase-seq|Gm12891, Chromatin_Structure||DNase-seq|Ipsnihi11, Chromatin_Structure||DNase-seq|Ips, Chromatin_Structure||DNase-seq|Medullo, Chromatin_Structure||DNase-seq|Medullod341, Chromatin_Structure||DNase-seq|Ipsnihi7, Chromatin_Structure||DNase-seq|Gm12892, Chromatin_Structure||DNase-seq|H7es, Chromatin_Structure||DNase-seq|K562, Chromatin_Structure||DNase-seq|T47d, Chromatin_Structure||DNase-seq|Hsmm, Chromatin_Structure|Andro|DNase-seq|Lncap, Chromatin_Structure||DNase-seq|Huh7, Chromatin_Structure||DNase-seq|Monocd14, Chromatin_Structure||DNase-seq|Gm10248, Chromatin_Structure||DNase-seq|Lncap, Chromatin_Structure|Sahactrl|DNase-seq|K562, Chromatin_Structure||DNase-seq|Ipscwru1, Chromatin_Structure||DNase-seq|Hsmmt, Chromatin_Structure||DNase-seq|Gm19238, Chromatin_Structure||DNase-seq|Gm13976, Chromatin_Structure||DNase-
B103 B104	chr4	125864692	C	T	transition	intergenic		CpG_site	NA	2c	Motifs|PWM||IRF-1, Motifs|PWM||IRF1, Motifs|PWM||ICSBP, Motifs|PWM||IRF-2, Motifs|PWM||IRF2, Motifs|PWM||IRF, Motifs|PWM||ISGF-3, Motifs|PWM||STAT1, Chromatin_Structure||DNase-seq|Rwpe1, Chromatin_Structure|Hypoxlac|FAIRE|Mcf7, Chromatin_Structure||FAIRE|Gliobla, Protein_Binding||ChIP-seq|MCF-7|ZNF217, Protein_Binding||ChIP-seq|HeLa-S3|CHD2, Protein_Binding||ChIP-seq|HeLa-S3|JUN, Protein_Binding||ChIP-seq|HeLa-S3|STAT3, Protein_Binding||ChIP-seq|HeLa-S3|MAX, Protein_Binding||ChIP-seq|SK-N-SH|SIN3A, Protein_Binding||ChIP-seq|A549|CEBPB, Protein_Binding||ChIP-seq|HeLa-S3|MYC, Protein_Binding||ChIP-seq|HeLa-S3|RFX5, Protein_Binding|ifng30|ChIP-

LJ	chr6	12749681	C	G	transversion	PHACTR1	intronic	NA	NA	3a	Motifs|PWM||Irf3, Chromatin_Structure||DNase-seq|Ipsnihi7, Chromatin_Structure||DNase-seq|Cd20ro01794, Chromatin_Structure||DNase-seq|Ipsnihi11, Chromatin_Structure||DNase-seq|Fibroblgm03348, Chromatin_Structure||DNase-seq|Sknsh, Chromatin_Structure||DNase-seq|Nt2d1, Chromatin_Structure||DNase-seq|Cerebrumfrontaloc, Chromatin_Structure||DNase-seq|8988t, Chromatin_Structure||DNase-seq|Hah, Chromatin_Structure||DNase-seq|Ipscwru1, Chromatin_Structure||DNase-seq|Gm20000, Chromatin_Structure||DNase-seq|H7es, Chromatin_Structure||DNase-seq|Hsmm, Chromatin_Structure||DNase-seq|Monocd14, Chromatin_Structure||DNase-seq|Gm13976, Chromatin_Structure||DNase-seq|H1hesc, Chromatin_Structure|Lenticon|DNase-seq|Fibroblgm03348, Chromatin_Structure||FAIRE|H1hesc, Chromatin_Structure||DNase-seq|Fibrobl, Chromatin_Structure|Diffa5d|DNase-seq|H7es, Chromatin_Structure||DNase-seq|Hff, Chromatin_Structure|Diffa9d|DNase-seq|H7es, Chromatin_Structure||DNase-seq|Werirb1, Chromatin_Structure||DNase-seq|Ips, Chromatin_Structure||DNase-seq|Hepatocytes, Chromatin_Structure||DNase-seq|Cerebellumoc, Chromatin_Structure||DNase-seq|Gm10266, Chromatin_Structure||DNase-seq|Frontalcortexoc, Chromatin_Structure||DNase-seq|Hsmmt, Chromatin_Structure||DNase-seq|H9es, Chromatin_Structure|Diff4d|DNase-seq|Lhcnm2, Chromatin_Structure|Lentimyod|DNase-seq|Fibroblgm03348, Chromatin_Structure||DNase-seq|Lhcnm2, Chromatin_Structure||DNase-seq|Hsmmfshd, Chromatin_Structure||DNase-seq|Medullo, Protein_Binding||ChIP-seq|H1-hESC|TAF1, Protein_Binding||ChIP-seq|H1-hESC|REST, Protein_Binding||ChIP-seq|H1-hESC|RFX5, Protein_Binding||ChIP-seq|H1-hESC|SP4, Protein_Binding||ChIP-seq|H1-hESC|YY1, Protein_Binding||ChIP-seq|H1-hESC|SIN3A, Protein_Binding||ChIP-seq|H1-hESC|BCL11A
LJ LS	chr7	20427755	C	T	transition	ITGB8	intronic	NA	NA	3a	Motifs|PWM||Zfp281, Chromatin_Structure||DNase-seq|Nhdfneo, Chromatin_Structure||DNase-seq|Sknmc, Protein_Binding||ChIP-seq|SK-N-MC|POLR2A
B103 B104	chr2	21646773	C	T	transition	intergenic		NA	NA	3a	Motifs|PWM||MYF, Motifs|PWM||Myf, Motifs|PWM||NeuroD, Motifs|PWM||Ascl2, Chromatin_Structure||DNase-seq|Th17, Chromatin_Structure||DNase-seq|Mel2183, Chromatin_Structure||DNase-seq|Th1, Chromatin_Structure||DNase-seq|UrotsaUt189, Chromatin_Structure||DNase-seq|Urotsa, Protein_Binding||ChIP-seq|HepG2|MAFK

B123 B124	chr8	23211573	T	G	transversion	LOXL2	intronic	NA	NA	3a	Motifs|PWM||Pou6f1, Motifs|PWM||Sox15, Chromatin_Structure||DNase-seq|Progfib, Chromatin_Structure|Diffa14d|DNase-seq|H7es, Chromatin_Structure||DNase-seq|Ipsnihi7, Chromatin_Structure||DNase-seq|Htr8, Chromatin_Structure||DNase-seq|Hpaec, Chromatin_Structure||DNase-seq|Hpdlf, Chromatin_Structure||DNase-seq|Hmvecdneo, Chromatin_Structure||DNase-seq|Fibropag08395, Chromatin_Structure||DNase-seq|Hpde6e6e7, Chromatin_Structure||DNase-seq|Huvec, Chromatin_Structure||DNase-seq|Hrpe, Chromatin_Structure|Est10nm30m|DNase-seq|Ishikawa, Chromatin_Structure||DNase-seq|Ipsnihi11, Chromatin_Structure||DNase-seq|Heartoc, Chromatin_Structure||DNase-seq|Hcf, Chromatin_Structure|Ifng4h|FAIRE|Helas3, Chromatin_Structure||DNase-seq|Ag09319, Chromatin_Structure||DNase-seq|UrotsaUt189, Chromatin_Structure||DNase-seq|Panisd, Chromatin_Structure||DNase-seq|Prec, Chromatin_Structure||DNase-seq|Fibroblgm03348, Chromatin_Structure|Est100nm1h|DNase-seq|Mcf7, Chromatin_Structure||DNase-seq|Sknsh, Chromatin_Structure||DNase-seq|Ag10803, Chromatin_Structure||DNase-seq|Hcpe, Chromatin_Structure|Hypoxlaccon|DNase-seq|Mcf7, Chromatin_Structure||FAIRE|Nhbe, Chromatin_Structure||DNase-seq|M059j, Chromatin_Structure||DNase-seq|Nt2d1, Chromatin_Structure||DNase-seq|Cerebrumfrontaloc, Chromatin_Structure||DNase-seq|Gm04504, Chromatin_Structure||DNase-seq|Hs5, Chromatin_Structure||DNase-seq|Hmvecdad, Chromatin_Structure||FAIRE|Nhek, Chromatin_Structure||DNase-seq|Hcfaa, Chromatin_Structure||DNase-seq|Hmveclbl, Chromatin_Structure||DNase-seq|8988t, Chromatin_Structure||DNase-seq|Hah, Chromatin_Structure||DNase-seq|Hpf, Chromatin_Structure||DNase-seq|Melano, Chromatin_Structure||DNase-seq|Wi38, Chromatin_Structure|ra|DNase-seq|Sknsh, Chromatin_Structure||DNase-seq|Hvmf, Chromatin_Structure||DNase-seq|Olfneurosphere, Chromatin_Structure||DNase-seq|Hrgec, Chromatin_Structure||DNase-seq|Hffmyc, Chromatin_Structure||DNase-seq|Stellate, Chromatin_Structure|Ctcfshrna|DNase-seq|Mcf7, Chromatin_Structure||DNase-seq|Hmvecdlyneo, Chromatin_Structure||DNase-seq|Hae, Chromatin_Structure||DNase-seq|Ipscwru1, Chromatin_Structure|Estctrl0h|DNase-seq|Mcf7, Chromatin_Structure||DNase-seq|Medullod341, Chromatin_Structure|Serumfree|DNase-seq|Aosmc, Chromatin_Structure||DNase-seq|Ag04450, Chromatin_Structure||DNase-seq|Hmvecdblneo, Chromatin_Structure||DNase-seq|Hct116, Chromatin_Structure||DNase-seq|H7es, Chromatin_Structure||DNase-seq|Fibropag08396, Chromatin_Structure||DNase-seq|Hsmm,
B113 B114	chr11	27959616	C	G	transversion	intergenic		NA	NA	3a	Motifs|PWM||MAFK, Motifs|PWM||Mafb, Motifs|PWM||NRL, Motifs|PWM||POU6F2, Chromatin_Structure|Serumfree|DNase-seq|Aosmc, Protein_Binding|ChIP-seq|HeLa-S3|EP300
B103 B104	chr20	34319685	T	G	transversion	RBM39	intronic	NA	NA	3a	Motifs|PWM||FOXC1, Motifs|PWM||Nkx3-1, Motifs|PWM||Cdx, Chromatin_Structure|Hypoxlac|FAIRE|Mcf7, Chromatin_Structure||DNase-seq|Nhek, Protein_Binding||ChIP-seq|H1-hESC|POLR2A, Protein_Binding||ChIP-seq|HepG2|POLR2A
LJ LS	chr4	39119053	G	T	transversion	KLHL5	intronic	NA	NA	3a	Motifs|PWM||Zfp410, Motifs|PWM||Rxra, Motifs|PWM||MZF1, Chromatin_Structure||DNase-seq|T47d, Protein_Binding|02pct|ChIP-seq|T-47D|GATA3, Protein_Binding|02pct|ChIP-seq|T-47D|FOXA1

B53 B54	chr17	41382051	T	C	transition	LINC00854	5upstream	NA	NA	3a	Motifs|PWM||EBF1, Chromatin_Structure||DNase-seq|Progfib, Chromatin_Structure||DNase-seq|Ipsnihi7, Chromatin_Structure||DNase-seq|Htr8, Chromatin_Structure||DNase-seq|Hpdlf, Chromatin_Structure||DNase-seq|Fibropag08395, Chromatin_Structure||DNase-seq|Hpde6e6e7, Chromatin_Structure||DNase-seq|Huvec, Chromatin_Structure||FAIRE|UrotsaUt189, Chromatin_Structure|Est10nm30m|DNase-seq|Ishikawa, Chromatin_Structure||DNase-seq|Cd20ro01794, Chromatin_Structure||DNase-seq|Heartoc, Chromatin_Structure||DNase-seq|Cd4naivewb11970640, Chromatin_Structure||DNase-seq|Hcf, Chromatin_Structure|Ifng4h|FAIRE|Helas3, Chromatin_Structure||DNase-seq|UrotsaUt189, Chromatin_Structure||DNase-seq|Panisd, Chromatin_Structure||DNase-seq|Fibroblgm03348, Chromatin_Structure||DNase-seq|Sknsh, Chromatin_Structure|Hypoxlaccon|DNase-seq|Mcf7, Chromatin_Structure||FAIRE|Gm12878, Chromatin_Structure||FAIRE|Nhbe, Chromatin_Structure||DNase-seq|Lncap, Chromatin_Structure||FAIRE|Htr8, Chromatin_Structure||DNase-seq|K562G2mphase, Chromatin_Structure||DNase-seq|Cerebrumfrontaloc, Chromatin_Structure||DNase-seq|Gm04504, Chromatin_Structure|Nabut|FAIRE|K562, Chromatin_Structure||FAIRE|Nhek, Chromatin_Structure||DNase-seq|Hcfaa, Chromatin_Structure||DNase-seq|Gm12891, Chromatin_Structure||DNase-seq|8988t, Chromatin_Structure||DNase-seq|Gm19240, Chromatin_Structure||DNase-seq|Melano, Chromatin_Structure||DNase-seq|Wi38, Chromatin_Structure|Ohurea|FAIRE|K562, Chromatin_Structure||DNase-seq|Hrgec, Chromatin_Structure||DNase-seq|Olfneurosphere, Chromatin_Structure||DNase-seq|Stellate, Chromatin_Structure|Ctcfshrna|DNase-seq|Mcf7, Chromatin_Structure||DNase-seq|Gm10248, Chromatin_Structure||FAIRE|Medullo, Chromatin_Structure||DNase-seq|Hae, Chromatin_Structure||DNase-seq|Ipscwru1, Chromatin_Structure||DNase-seq|Th2, Chromatin_Structure||FAIRE|Gm12892, Chromatin_Structure||DNase-seq|Medullod341, Chromatin_Structure||DNase-seq|Gm20000, Chromatin_Structure||DNase-seq|H7es, Chromatin_Structure||DNase-seq|Hsmm, Chromatin_Structure||DNase-seq|Fibropag08396, Chromatin_Structure||DNase-seq|Monocd14, Chromatin_Structure||DNase-seq|Tregwb78495824, Chromatin_Structure||DNase-seq|Cd4naivewb78495824, Chromatin_Structure||DNase-seq|Hasp, Chromatin_Structure||DNase-seq|Panislets, Chromatin_Structure||DNase-seq|Gm13976, Chromatin_Structure||DNase-seq|Saec,
B103 B104	chr17	41633492	A	C	transversion	intergenic		NA	NA	3a	Motifs|PWM||Srf, Motifs|PWM||FOXP1, Motifs|PWM||Zfp105, Motifs|PWM||Mtf1, Chromatin_Structure||DNase-seq|A549, Chromatin_Structure||DNase-seq|Helas3, Chromatin_Structure|Ifna4h|DNase-seq|Helas3, Protein_Binding||ChIP-seq|U2OS|SETDB1, Protein_Binding|02pct|ChIP-seq|A549|YY1

B103 B104	chr10	71169085	T	G	transversion	TACR2	intronic	NA	NA	3a	Motifs|PWM||MAX, Chromatin_Structure||DNase-seq|Th1, Chromatin_Structure||DNase-seq|H1hesc, Chromatin_Structure||DNase-seq|K562G1phase, Chromatin_Structure||DNase-seq|Heartoc, Chromatin_Structure||DNase-seq|Adultcd4th0, Chromatin_Structure||DNase-seq|Cerebellumoc, Chromatin_Structure||DNase-seq|Sknsh, Chromatin_Structure||DNase-seq|Frontalcortexoc, Chromatin_Structure||DNase-seq|H7es, Chromatin_Structure||DNase-seq|K562, Chromatin_Structure||DNase-seq|Hs27a, Chromatin_Structure||DNase-seq|Th1wb54553204, Chromatin_Structure||DNase-seq|Adultcd4th1, Chromatin_Structure||DNase-seq|Psoasmuscleoc, Chromatin_Structure|Andro|DNase-seq|Lncap, Chromatin_Structure||DNase-seq|A549, Chromatin_Structure||DNase-seq|Monocd14ro1746, Chromatin_Structure||DNase-seq|Monocd14, Chromatin_Structure||DNase-seq|Lncap, Chromatin_Structure||DNase-seq|Huvec, Chromatin_Structure||DNase-seq|K562G2mphase, Chromatin_Structure||DNase-seq|Cerebrumfrontaloc, Chromatin_Structure||DNase-seq|Hs5, Chromatin_Structure||DNase-seq|Urotsa, Protein_Binding||ChIP-seq|K562|POLR2A, Protein_Binding|ifng6h|ChIP-seq|K562|MYC, Protein_Binding|ifng30|ChIP-seq|K562|IRF1, Protein_Binding||ChIP
B83 B84	chr2	109609615	G	A	transition	intergenic		NA	NA	3a	Motifs|PWM||Bcl6b, Chromatin_Structure||DNase-seq|Osteobl, Protein_Binding|differential|ChIP-seq|Caco2|HNF4A, Protein_Binding|proliferation|ChIP-seq|Caco2|HNF4A
B53 B54	chr5	118144221	T	C	transition	intergenic		NA	NA	3a	Motifs|PWM||FOXM1, Chromatin_Structure|Nabut|FAIRE|K562, Chromatin_Structure||DNase-seq|H7es, Chromatin_Structure|Diffa9d|DNase-seq|H7es, Protein_Binding|ChIP-seq|HepG2|MAFK
B83 B84	chr4	128589038	T	A	transversion	INTU	intronic	NA	NA	3a	Motifs|PWM||Foxk1, Motifs|PWM||Foxj3, Motifs|PWM||Glis2, Motifs|PWM||TBP, Motifs|PWM||Tbp, Motifs|PWM||FOXC2, Motifs|PWM||Foxl1, Motifs|PWM||FOXP1, Motifs|PWM||Foxa2, Chromatin_Structure||DNase-seq|Sknmc, Protein_Binding||ChIP-seq|SK-N-MC|POLR2A
B62 B63	chr2	183452479	C	T	transition	intergenic		NA	NA	3a	Motifs|PWM||NF-kappaB, Chromatin_Structure||DNase-seq|Hah, Chromatin_Structure||FAIRE|Panislets, Chromatin_Structure||DNase-seq|Sknmc, Chromatin_Structure|FAIRE|Gliobla, Protein_Binding||ChIP-seq|K562|SPI1
B62 B63	chr1	199011594	G	A	transition	intergenic		NA	NA	3a	Motifs|PWM||Zfp187, Motifs|PWM||Nkx3-1, Chromatin_Structure||DNase-seq|Hl60, Protein_Binding||ChIP-seq|IMR90|CEBPB, Protein_Binding||ChIP-seq|HepG2|CEBPB

B63	chr4	1331141	T	C	transition	MAEA	intronic	NA	NA	4	Chromatin_Structure||DNase-seq|Progfib, Chromatin_Structure||DNase-seq|Adultcd4th0, Chromatin_Structure||DNase-seq|Adultcd4th1, Chromatin_Structure||DNase-seq|Hrpe, Chromatin_Structure||DNase-seq|Huvec, Chromatin_Structure||DNase-seq|Heartoc, Chromatin_Structure||DNase-seq|Fibrop, Chromatin_Structure||DNase-seq|Panisd, Chromatin_Structure||DNase-seq|Hepatocytes, Chromatin_Structure||DNase-seq|Gm10266, Chromatin_Structure||DNase-seq|Frontalcortexoc, Chromatin_Structure||DNase-seq|Gliobla, Chromatin_Structure||DNase-seq|Huh7, Chromatin_Structure||DNase-seq|Helas3, Chromatin_Structure||DNase-seq|Hsmmt, Chromatin_Structure||DNase-seq|Gm19238, Chromatin_Structure|Ifna4h|DNase-seq|Helas3, Chromatin_Structure||DNase-seq|Gm12891, Chromatin_Structure||DNase-seq|Hmveclbl, Chromatin_Structure||DNase-seq|8988t, Chromatin_Structure||DNase-seq|Huh75, Chromatin_Structure||DNase-seq|Gm19239, Chromatin_Structure||DNase-seq|Hmec, Chromatin_Structure||DNase-seq|Osteobl, Chromatin_Structure||DNase-seq|Phte, Chromatin_Structure||DNase-seq|Imr90, Chromatin_Structure||DNase-seq|Chorion, Chromatin_Structure||DNase-seq|Gm20000, Chromatin_Structure||DNase-seq|Myometr, Chromatin_Structure||DNase-seq|Hmvecdblneo, Chromatin_Structure||DNase-seq|Fibropag08396, Chromatin_Structure||DNase-seq|Hsmm, Chromatin_Structure||DNase-seq|Psoasmuscleoc, Chromatin_Structure||DNase-seq|Panislets, Protein_Binding||ChIP-seq|HUVEC|FOS, Protein_Binding||ChIP-seq|GM12878|BCL3,
B83 B84	chr5	5609554	A	C	transversion	intergenic		NA	NA	4	Chromatin_Structure||DNase-seq|Th17, Chromatin_Structure||DNase-seq|Th1, Protein_Binding||ChIP-seq|K562|RFX3
B113 B114	chr2	10036253	C	T	transition	TAF1B	intronic	CpG_site	NA	4	Chromatin_Structure||FAIRE|Panislets, Chromatin_Structure||FAIRE|Medullo, Chromatin_Structure||DNase-seq|Mel2183, Chromatin_Structure||DNase-seq|Colo829, Chromatin_Structure||FAIRE|Hepg2, Chromatin_Structure||FAIRE|Nhek, Chromatin_Structure||FAIRE|Gliobla, Protein_Binding|01pct|ChIP-seq|MCF10A-Er-Src|POLR2A
B83	chr8	10745154	T	C	transition	intergenic		NA	NA	4	Chromatin_Structure||DNase-seq|Ipscwru1, Chromatin_Structure||DNase-seq|Medullod341, Chromatin_Structure||DNase-seq|Gcbcell, Chromatin_Structure||DNase-seq|Ips, Chromatin_Structure||DNase-seq|Medullo, Protein_Binding||ChIP-seq|GM12878|SP1
B62 B63	chr6	18908561	G	A	transition	intergenic		NA	NA	4	Chromatin_Structure|Estctrl0h|DNase-seq|Mcf7, Chromatin_Structure||DNase-seq|Mcf7, Chromatin_Structure||DNase-seq|T47d, Protein_Binding|02pct|ChIP-seq|T-47D|GATA3

B103 B104	chrX	20489848	T	C	transition	intergenic		NA	NA	4	Chromatin_Structure||DNase-seq|Melano, Chromatin_Structure||DNase-seq|Mel2183, Chromatin_Structure||DNase-seq|Colo829, Chromatin_Structure||DNase-seq|Huh7, Chromatin_Structure||DNase-seq|Hepg2, Chromatin_Structure||FAIRE|Hepg2, Chromatin_Structure||DNase-seq|Rpmi7951, Protein_Binding||ChIP-seq|HepG2|TCF7L2, Protein_Binding||ChIP-seq|HepG2|MBD4, Protein_Binding||ChIP-seq|HepG2|FOXA1, Protein_Binding||ChIP-seq|HepG2|HDAC2, Protein_Binding||ChIP-seq|HepG2|EP300, Protein_Binding||ChIP-seq|HepG2|RXRA, Protein_Binding||ChIP-seq|HepG2|FOXA2, Protein_Binding||ChIP-seq|HepG2|NFIC, Protein_Binding||ChIP-seq|HepG2|SP1, Protein_Binding||ChIP-seq|HepG2|TEAD4, Protein_Binding||ChIP-seq|HepG2|HNF4A, Protein_Binding||ChIP-seq|VCaP|AR, Protein_Binding||ChIP-seq|HepG2|ARID3A, Protein_Binding||ChIP-seq|HepG2|CEBPB, Protein_Binding||ChIP-seq|HepG2|HNF4G, Protein_Binding||ChIP-seq|HepG2|SMC3.
B114	chr13	21816875	A	G	transition	intergenic		NA	NA	4	Chromatin_Structure||DNase-seq|T47d, Chromatin_Structure||FAIRE|Nhbe, Chromatin_Structure|Hypoxlac|DNase-seq|Mcf7, Chromatin_Structure||DNase-seq|Rwpe1, Chromatin_Structure||DNase-seq|Mcf7, Chromatin_Structure||FAIRE|H1hesc, Chromatin_Structure||DNase-seq|Hmec, Protein_Binding|EWS-ERG-fusion|ChIP-seq|CADO-ES1|ERG, Protein_Binding|EWS-ERG-fusion|ChIP-seq|CADO-ES1|EWSR1
B114	chr13	21816893	T	A	transversion	intergenic		NA	NA	4	Chromatin_Structure||FAIRE|Nhbe, Chromatin_Structure|Hypoxlac|DNase-seq|Mcf7, Chromatin_Structure||DNase-seq|Rwpe1, Chromatin_Structure||DNase-seq|Mcf7, Chromatin_Structure||FAIRE|H1hesc, Chromatin_Structure||DNase-seq|Hmec, Protein_Binding|EWS-ERG-fusion|ChIP-seq|CADO-ES1|ERG, Protein_Binding|EWS-ERG-fusion|ChIP-seq|CADO-ES1|EWSR1
B114	chr13	21816900	G	A	transition	intergenic		CpG_site	NA	4	Chromatin_Structure||FAIRE|Nhbe, Chromatin_Structure|Hypoxlac|DNase-seq|Mcf7, Chromatin_Structure||DNase-seq|Rwpe1, Chromatin_Structure||DNase-seq|Mcf7, Chromatin_Structure||FAIRE|H1hesc, Chromatin_Structure||DNase-seq|Hmec, Protein_Binding|EWS-ERG-fusion|ChIP-seq|CADO-ES1|ERG, Protein_Binding|EWS-ERG-fusion|ChIP-seq|CADO-ES1|EWSR1
B114	chr13	21816903	A	G	transition	intergenic		NA	NA	4	Chromatin_Structure||FAIRE|Nhbe, Chromatin_Structure|Hypoxlac|DNase-seq|Mcf7, Chromatin_Structure||DNase-seq|Rwpe1, Chromatin_Structure||DNase-seq|Mcf7, Chromatin_Structure||FAIRE|H1hesc, Chromatin_Structure||DNase-seq|Hmec, Protein_Binding|EWS-ERG-fusion|ChIP-seq|CADO-ES1|ERG, Protein_Binding|EWS-ERG-fusion|ChIP-seq|CADO-ES1|EWSR1
B62 B63	chr20	30972541	G	C	transversion	ASXL1	intronic	NA	NA	4	Chromatin_Structure||DNase-seq|Hbvp, Chromatin_Structure||DNase-seq|Th1wb33676984, Chromatin_Structure||DNase-seq|Th17, Chromatin_Structure||DNase-seq|Hepg2, Chromatin_Structure||DNase-seq|Mel2183, Chromatin_Structure||DNase-seq|Th1, Chromatin_Structure||DNase-seq|Colo829, Chromatin_Structure||DNase-seq|Be2c, Protein_Binding||ChIP-seq|HepG2|HNF4G
B53 B54	chr13	32042443	G	A	transition	intergenic		NA	NA	4	Chromatin_Structure|Est10nm30m|DNase-seq|Ecc1, Chromatin_Structure|Dm002p1h|DNase-seq|Ecc1, Protein_Binding||ChIP-seq|HSPC|IKZF1
B113 B114	chr20	34539772	A	G	transition	SCAND1	3downstream	NA	NA	4	Chromatin_Structure||DNase-seq|Tregwb83319432, Protein_Binding||ChIP-seq|U87|POLR2A, Protein_Binding||ChIP-seq|HepG2|POLR2A, Protein_Binding||ChIP-seq|H1-hESC|POLR2A

B123 B124	chr19	35449194	T	C	transition	ZNF792	nonsyn p.N522S	NA	NA	4	Chromatin_Structure|Znf4c50c4|DNase-seq|K562, Chromatin_Structure||DNase-seq|H7es, Chromatin_Structure||DNase-seq|K562, Chromatin_Structure|Znfp5|DNase-seq|K562, Chromatin_Structure||DNase-seq|Be2c, Protein_Binding||ChIP-seq|K562|SETDB1, Protein_Binding||ChIP-seq|U2OS|SETDB1, Protein_Binding||ChIP-seq|K562|TRIM28, Protein_Binding||ChIP-seq|K562|RFX3, Protein_Binding||ChIP-seq|K562|CBX3,
B93 B94	chr15	39934086	G	A	transition	FSIP1	intronic	NA	NA	4	Chromatin_Structure||DNase-seq|Hpde6e6e7, Chromatin_Structure||DNase-seq|Huvec, Chromatin_Structure||DNase-seq|Tregwb83319432, Chromatin_Structure|Ctcfshrna|DNase-seq|Mcf7, Chromatin_Structure||DNase-seq|Mcf7, Chromatin_Structure||DNase-seq|Helas3, Chromatin_Structure|Ifna4h|DNase-seq|Helas3, Chromatin_Structure||DNase-seq|Rwpe1, Chromatin_Structure|Hypoxlac|DNase-seq|Mcf7,
B62 B63	chr2	47527796	G	A	transition	AC073283.4	non-coding intronic	NA	NA	4	Chromatin_Structure||FAIRE|Gm19239, Chromatin_Structure||DNase-seq|Naivebcell, Chromatin_Structure||DNase-seq|Cd20ro01778,
B93 B94	chr17	49168444	T	C	transition	SPAG9	intronic	NA	NA	4	Chromatin_Structure||DNase-seq|Hmec, Chromatin_Structure||FAIRE|K562, Chromatin_Structure||FAIRE|Hepg2, Protein_Binding||ChIP-seq|A549|POLR2A
B62 B63	chr20	52707477	A	G	transition	intergenic		NA	NA	4	Chromatin_Structure||DNase-seq|Gm12878, Chromatin_Structure||DNase-seq|Gm06990, Chromatin_Structure||DNase-seq|Gm12865, Protein_Binding||ChIP-seq|GM12878|EBF1
B93 B94	chr19	57037004	C	G	transversion	ZNF471	nonsyn p.A523G	NA	NA	4	Chromatin_Structure||DNase-seq|Hrgec, Protein_Binding||ChIP-seq|GM12878|ZNF274, Protein_Binding||ChIP-seq|U2OS|SETDB1, Protein_Binding||ChIP-seq|NT2-D1|ZNF274, Protein_Binding||ChIP-seq|HEK293|TRIM28, Protein_Binding|02pct|ChIP-seq|A549|TCF12, Protein_Binding||ChIP-seq|K562|ZNF274, Protein_Binding||ChIP-seq|U2OS|TRIM28, Protein_Binding||ChIP-seq|HepG2|ZNF274,

B123 B124	chr5	64235781	C	A	transversion	CWC27	intronic	NA	NA	4	Motifs|Footprinting|A549|, Chromatin_Structure|Diffa14d|DNase-seq|H7es, Chromatin_Structure||DNase-seq|Ipsnihi7, Chromatin_Structure|Znfb34a8|DNase-seq|K562, Chromatin_Structure||DNase-seq|Hpdlf, Chromatin_Structure||DNase-seq|Huvec, Chromatin_Structure||DNase-seq|Hrpe, Chromatin_Structure|Est10nm30m|DNase-seq|Ishikawa, Chromatin_Structure||DNase-seq|Ipsnihi11, Chromatin_Structure||DNase-seq|Hcf, Chromatin_Structure||DNase-seq|Ag09319, Chromatin_Structure||DNase-seq|Prec, Chromatin_Structure|Est100nm1h|DNase-seq|Mcf7, Chromatin_Structure|Znf4c50c4|DNase-seq|K562, Chromatin_Structure||DNase-seq|Hcpe, Chromatin_Structure||DNase-seq|Rptec, Chromatin_Structure|Hypoxlaccon|DNase-seq|Mcf7, Chromatin_Structure||DNase-seq|Nt2d1, Chromatin_Structure||DNase-seq|Hcfaa, Chromatin_Structure||DNase-seq|Hmveclbl, Chromatin_Structure||DNase-seq|Hpf, Chromatin_Structure||DNase-seq|Wi38, Chromatin_Structure||DNase-seq|Hvmf, Chromatin_Structure||DNase-seq|Hffmyc, Chromatin_Structure|Ctcfshrna|DNase-seq|Mcf7, Chromatin_Structure||DNase-seq|Hae, Chromatin_Structure|Estctrl0h|DNase-seq|Mcf7, Chromatin_Structure||DNase-seq|Ag04450, Chromatin_Structure||DNase-seq|H7es, Chromatin_Structure||DNase-seq|Fibropag08396, Chromatin_Structure||DNase-seq|Hmf, Chromatin_Structure||DNase-seq|Hpaf, Chromatin_Structure||DNase-seq|Hasp, Chromatin_Structure||DNase-seq|Skmc, Chromatin_Structure|Znfa41c6|DNase-seq|K562, Chromatin_Structure|Znff41b2|DNase-seq|K562, Chromatin_Structure||DNase-seq|Saec, Chromatin_Structure||DNase-seq|H1hesc, Chromatin_Structure||DNase-seq|Hbvp, Chromatin_Structure||DNase-seq|Nhbera, Chromatin_Structure||DNase-seq|Hipe, Chromatin_Structure|Diffa5d|DNase-seq|H7es, Chromatin_Structure||DNase-seq|Hff, Chromatin_Structure||DNase-seq|K562, Chromatin_Structure|Diffa9d|DNase-seq|H7es, Chromatin_Structure|Tam10030|DNase-seq|Ishikawa, Chromatin_Structure||DNase-seq|Hrce, Chromatin_Structure||DNase-seq|Hnpce, Chromatin_Structure|Diffa2d|DNase-seq|H7es, Chromatin_Structure||DNase-seq|Ips, Chromatin_Structure||DNase-seq|Mcf7, Chromatin_Structure||DNase-seq|Fibrop, Chromatin_Structure||DNase-seq|T47d, Chromatin_Structure||DNase-seq|A549, Chromatin_Structure||DNase-seq|Huh7, Chromatin_Structure||DNase-seq|Hgf, Chromatin_Structure||DNase-seq|Nha, Chromatin_Structure|Znfp5|DNase-seq|K562, Chromatin_Structure|Est10nm30m|DNase-seq|T47d, Chromatin_Structure||DNase-seq|Rwpe1, Chromatin_Structure||DNase-seq|Hconf, Chromatin_Structure||DNase-seq|Hre, Chromatin_Structure||DNase-seq|Nhdfneo, Chromatin_Structure||DNase-seq|Hs5, Chromatin_Structure||DNase-seq|Th1wb33676984, Chromatin_Structure|Hypoxlac|FAIRE|Mcf7, Chromatin_Structure||DNase-seq|Th1, Chromatin_Structure||DNase-seq|Msc, Protein_Binding|ChIP-seq|HEK293|TRIM28
B93 B94	chr5	65445884	C	G	transversion	SREK1	intronic	NA	NA	4	

B83 B84	chr12	66449758	T	C	transition	intergenic		NA	NA	4	Chromatin_Structure||DNase-seq|Gm10248, Chromatin_Structure||FAIRE|Gm18507, Chromatin_Structure||DNase-seq|Gm20000, Chromatin_Structure||DNase-seq|Gm13976, Chromatin_Structure||FAIRE|Gm19239, Chromatin_Structure||DNase-seq|Gm10266, Chromatin_Structure||FAIRE|Gm12891, Chromatin_Structure||DNase-seq|Gm18507, Protein_Binding|tnfa|ChIP-seq|GM12878|NFKB1, Protein_Binding||ChIP-seq|GM12878|ATF2, Protein_Binding||ChIP-seq|GM12878|TBL1XR1, Protein_Binding|tnfa|ChIP-seq|GM12892|NFKB1, Protein_Binding|tnfa|ChIP-seq|GM19193|NFKB1, Protein_Binding||ChIP-seq|GM12878|NFIC, Protein_Binding||ChIP-seq|GM12878|EP300, Protein_Binding||ChIP-seq|GM12878|NFATC1, Protein_Binding||ChIP-seq|GM12878|POLR2A, Protein_Binding||ChIP-seq|GM12878|BHLHE40, Protein_Binding||ChIP-seq|GM12891|POU2F2, Protein_Binding||ChIP-seq|VCaP|AR, Protein_Binding||ChIP-
LJ LS	chr16	71264288	C	T	transition	HYDIN	intronic	NA	NA	4	Chromatin_Structure||DNase-seq|Ipsnihi7, Chromatin_Structure||DNase-seq|Hpde6e6e7, Chromatin_Structure||DNase-seq|Ipsnihi11, Chromatin_Structure||DNase-seq|UrotsaUt189, Chromatin_Structure||DNase-seq|Sknsh, Chromatin_Structure||DNase-seq|Lncap, Chromatin_Structure||DNase-seq|Nt2d1, Chromatin_Structure||DNase-seq|Ipscwru1, Chromatin_Structure|Estctrl0h|DNase-seq|Mcf7, Chromatin_Structure||DNase-seq|Medullod341, Chromatin_Structure||DNase-seq|H7es, Chromatin_Structure||DNase-seq|Panislets, Chromatin_Structure||DNase-seq|H1hesc, Chromatin_Structure|Tam10030|DNase-seq|Ishikawa, Chromatin_Structure|Andro|DNase-seq|Lncap, Chromatin_Structure||DNase-seq|Ips, Chromatin_Structure||DNase-seq|Mcf7, Chromatin_Structure||DNase-seq|T47d, Chromatin_Structure||DNase-seq|A549, Chromatin_Structure||DNase-seq|Hek293t, Chromatin_Structure||DNase-seq|Urotsa, Chromatin_Structure|Est10nm30m|DNase-seq|T47d, Chromatin_Structure||DNase-seq|Rwpe1, Chromatin_Structure|Est10nm30m|DNase-seq|Ecc1, Chromatin_Structure||DNase-seq|H9es, Chromatin_Structure||DNase-seq|Medullo, Chromatin_Structure||DNase-seq|Chorion, Protein_Binding||ChIP-seq|K562|REX3
B54	chr12	76731688	G	T	transversion	intergenic		NA	NA	4	Chromatin_Structure||DNase-seq|Fibroblgm03348, Chromatin_Structure|Serumfree|DNase-seq|Aosmc, Chromatin_Structure|Lenticon|DNase-seq|Fibroblgm03348, Chromatin_Structure||DNase-seq|Fibrobl, Chromatin_Structure||DNase-seq|Hipe, Chromatin_Structure||DNase-seq|Fibrop, Chromatin_Structure||DNase-seq|Hbvsmc, Chromatin_Structure||DNase-seq|Gm04503, Chromatin_Structure||DNase-seq|Bj, Chromatin_Structure||DNase-seq|Fibropag20443, Protein_Binding||ChIP-seq|HUVEC|GATA2, Protein_Binding||ChIP-seq|HeLa-S3|JUN, Protein_Binding||ChIP-seq|HeLa-S3|EP300, Protein_Binding||ChIP-seq|IMR90|CEBPB, Protein_Binding||ChIP-seq|HeLa-S3|JUND, Protein_Binding||ChIP-seq|HUVEC|FOS, Protein_Binding||ChIP-

B113 B114	chr16	86024734	T	C	transition	intergenic	NA	NA	4	Chromatin_Structure||DNase-seq|Cd20ro01794, Chromatin_Structure||DNase-seq|Gm12891, Chromatin_Structure||DNase-seq|8988t, Chromatin_Structure||DNase-seq|Gm19240, Chromatin_Structure||DNase-seq|Gm10248, Chromatin_Structure||FAIRE|Gm18507, Chromatin_Structure||FAIRE|Gm12892, Chromatin_Structure||DNase-seq|Gm13976, Chromatin_Structure||FAIRE|Gm19239, Chromatin_Structure||DNase-seq|K562, Chromatin_Structure||DNase-seq|Gcbcell, Chromatin_Structure||DNase-seq|Cll, Chromatin_Structure||DNase-seq|Gm10266, Chromatin_Structure||DNase-seq|Gm19238, Chromatin_Structure||DNase-seq|Gm19239, Chromatin_Structure||DNase-seq|Naivebcell, Chromatin_Structure||DNase-seq|Th1, Chromatin_Structure||FAIRE|Gm12891, Chromatin_Structure||DNase-seq|Gm12892, Chromatin_Structure||DNase-seq|Gm18507, Chromatin_Structure||DNase-seq|Gm12878, Protein_Binding||ChIP-seq|GM12878|PML, Protein_Binding||ChIP-seq|GM12878|ATF2, Protein_Binding||ChIP-seq|GM12878|TAF1, Protein_Binding||ChIP-seq|GM18505|POLR2A, Protein_Binding||ChIP-seq|GM12878|YY1
B123 B124	chr14	104390533	C	T	transition	intergenic	NA	NA	4	Chromatin_Structure||DNase-seq|Huvec, Chromatin_Structure||DNase-seq|Heartoc, Chromatin_Structure||DNase-seq|Panisd, Chromatin_Structure|Znf4c50c4|DNase-seq|K562, Chromatin_Structure||DNase-seq|Sknsh, Chromatin_Structure|Znff41b2|DNase-seq|K562, Chromatin_Structure|Znfa41c6|DNase-seq|K562, Chromatin_Structure||DNase-seq|H1hesc, Chromatin_Structure||DNase-seq|K562, Chromatin_Structure|Saha1u72hr|DNase-seq|K562, Chromatin_Structure||DNase-seq|Hepatocytes, Chromatin_Structure||DNase-seq|T47d, Chromatin_Structure|Sahactrl|DNase-seq|K562, Chromatin_Structure|Nabut|DNase-seq|K562, Chromatin_Structure||DNase-seq|Osteobl, Protein_Binding||ChIP-seq|SK-N-SH|EP300, Protein_Binding||ChIP-seq|K562|ZBTB7A, Protein_Binding||ChIP-seq|SH-SY5Y|GATA2, Protein_Binding||ChIP-seq|HUVEC|GATA2, Protein_Binding|shbrg1|ChIP-seq|CD36|GATA1, Protein_Binding||ChIP-seq|K562|TEAD4, Protein_Binding||ChIP-seq|Jurkat|CREBBP, Protein_Binding||ChIP-seq|SK-N-SH|YY1, Protein_Binding|shLuc|ChIP-seq|CD36|GATA1, Protein_Binding||ChIP-

LJ LS	chr3	120315393	G	A	transition		NDUFB4	intronic	NA	CpG_island	4	Chromatin_Structure||DNase-seq|H1hesc, Chromatin_Structure||DNase-seq|Progfib, Chromatin_Structure|Lenticon|DNase-seq|Fibroblgm03348, Chromatin_Structure|Ifna4h|FAIRE|Helas3, Chromatin_Structure||DNase-seq|Adultcd4th0, Chromatin_Structure||DNase-seq|Fibrobl, Chromatin_Structure||DNase-seq|Ipsnihi7, Chromatin_Structure||DNase-seq|Htr8, Chromatin_Structure||DNase-seq|Hpaec, Chromatin_Structure||FAIRE|Gm19239, Chromatin_Structure||DNase-seq|K562, Chromatin_Structure|Tam10030|DNase-seq|Ishikawa, Chromatin_Structure||DNase-seq|Fibropag08395, Chromatin_Structure|Andro|DNase-seq|Lncap, Chromatin_Structure||DNase-seq|Adultcd4th1, Chromatin_Structure||DNase-seq|Hpde6e6e7, Chromatin_Structure||DNase-seq|Gcbcell, Chromatin_Structure||DNase-seq|Cll, Chromatin_Structure||DNase-seq|Mel2183, Chromatin_Structure||DNase-seq|Huvec, Chromatin_Structure|Est10nm30m|DNase-seq|Ishikawa, Chromatin_Structure|Saha1u72hr|DNase-seq|K562, Chromatin_Structure||DNase-seq|Ipsnihi11, Chromatin_Structure||DNase-seq|Cd20ro01794, Chromatin_Structure||DNase-seq|Ips, Chromatin_Structure||DNase-seq|Mcf7, Chromatin_Structure||DNase-seq|Heartoc, Chromatin_Structure||DNase-seq|Colo829, Chromatin_Structure|Ifng4h|FAIRE|Helas3, Chromatin_Structure||DNase-seq|Fibrop, Chromatin_Structure||DNase-seq|UrotsaUt189, Chromatin_Structure||DNase-seq|Panisd, Chromatin_Structure||DNase-seq|Fibroblgm03348, Chromatin_Structure||DNase-seq|Cerebellumoc, Chromatin_Structure||DNase-seq|Gm10266, Chromatin_Structure||DNase-seq|Nhek, Chromatin_Structure||DNase-seq|Frontalcortexoc, Chromatin_Structure||DNase-seq|Sknsh, Chromatin_Structure|Hypoxlaccon|DNase-seq|Mcf7, Chromatin_Structure||DNase-seq|Gliobla, Chromatin_Structure||DNase-seq|T47d, Chromatin_Structure||DNase-seq|A549, Chromatin_Structure||DNase-seq|Huh7, Chromatin_Structure||DNase-seq|Lncap, Chromatin_Structure|Sahactrl|DNase-seq|K562, Chromatin_Structure|Nabut|DNase-seq|K562, Chromatin_Structure||DNase-seq|Helas3, Chromatin_Structure||DNase-seq|K562G2mphase, Chromatin_Structure||DNase-seq|Gm19238, Chromatin_Structure||DNase-seq|Hsmmt, Chromatin_Structure||DNase-seq|Cerebrumfrontaloc, Chromatin_Structure|Ifna4h|DNase-seq|Helas3, Chromatin_Structure||FAIRE|Nhek, Chromatin_Structure||DNase-seq|Hek293t, Chromatin_Structure||DNase-seq|Urotsa, Chromatin_Structure||DNase-seq|Gm12891, Chromatin_Structure|Est10nm30m|DNase-seq|T47d, Chromatin_Structure||DNase-seq|Rwpe1, Chromatin_Structure||DNase-seq|Huh75, Chromatin_Structure||DNase-
B113	chr4	122576011	T	C	transition		intergenic		NA	NA	4	Chromatin_Structure||FAIRE|Gm19239, Chromatin_Structure||DNase-seq|Gcbcell, Chromatin_Structure||FAIRE|Gm12878, Chromatin_Structure||DNase-seq|Gm12864, Chromatin_Structure||DNase-seq|Gm12865, Chromatin_Structure||DNase-seq|Gm12891, Chromatin_Structure||DNase-seq|Gm19240, Chromatin_Structure||DNase-seq|Gm12878, Protein_Binding||ChIP-seq|GM12891|SPI1, Protein_Binding||ChIP-seq|GM12878|RUNX3,

B103 B104	chr12	133397352	G	A	transition	GOLGA3	intronic	NA	NA	4	Chromatin_Structure||DNase-seq|Gm10248, Chromatin_Structure||FAIRE|Gm18507, Chromatin_Structure||DNase-seq|Gm13976, Chromatin_Structure||DNase-seq|Gcbcell, Chromatin_Structure||DNase-seq|Gm12865, Chromatin_Structure||DNase-seq|Gm12892, Chromatin_Structure||DNase-seq|Gm18507, Chromatin_Structure||DNase-seq|Gm12878_Protein_Binding|ChIP-
LJ LS	chr9	134281048	C	T	transition	intergenic		CpG_site	NA	4	Motifs|Footprinting|SkMC|, Chromatin_Structure||DNase-seq|Progfib, Chromatin_Structure|Diffa14d|DNase-seq|H7es, Chromatin_Structure||DNase-seq|Ipsnihi7, Chromatin_Structure||DNase-seq|Hpdlf, Chromatin_Structure||DNase-seq|Hmvecdneo, Chromatin_Structure||DNase-seq|Fibropag08395, Chromatin_Structure||DNase-seq|Hrpe, Chromatin_Structure|Est10nm30m|DNase-seq|Ishikawa, Chromatin_Structure||DNase-seq|Ipsnihi11, Chromatin_Structure||DNase-seq|Hcf, Chromatin_Structure|Ifng4h|FAIRE|Helas3, Chromatin_Structure||DNase-seq|Ag09319, Chromatin_Structure||DNase-seq|Panisd, Chromatin_Structure||DNase-seq|Fibroblgm03348, Chromatin_Structure||DNase-seq|Hcpe, Chromatin_Structure||DNase-seq|Ag10803, Chromatin_Structure||DNase-seq|Rptec, Chromatin_Structure||DNase-seq|M059j, Chromatin_Structure||DNase-seq|Nt2d1, Chromatin_Structure||DNase-seq|K562G2mphase, Chromatin_Structure||DNase-seq|Gm04504, Chromatin_Structure||DNase-seq|Hcfaa, Chromatin_Structure||DNase-seq|Hpf, Chromatin_Structure||DNase-seq|Hah, Chromatin_Structure||DNase-seq|Melano, Chromatin_Structure||DNase-seq|Hvmf, Chromatin_Structure||DNase-seq|Olfneurosphere, Chromatin_Structure||DNase-seq|Hffmyc, Chromatin_Structure||DNase-seq|Hae, Chromatin_Structure||DNase-seq|Ipscwru1, Chromatin_Structure|Serumfree|DNase-seq|Aosmc, Chromatin_Structure||DNase-seq|Ag04450, Chromatin_Structure||DNase-seq|H7es, Chromatin_Structure||DNase-seq|Fibropag08396, Chromatin_Structure||DNase-seq|Hsmm, Chromatin_Structure||DNase-seq|Hac, Chromatin_Structure||DNase-seq|Ag09309, Chromatin_Structure||DNase-seq|Hmf, Chromatin_Structure||DNase-seq|Hpaf, Chromatin_Structure||DNase-seq|Skmc, Chromatin_Structure||DNase-seq|Hasp, Chromatin_Structure||DNase-seq|H1hesc, Chromatin_Structure|Lenticon|DNase-seq|Fibroblgm03348, Chromatin_Structure|Ifna4h|FAIRE|Helas3, Chromatin_Structure||DNase-seq|Fibrobl, Chromatin_Structure||DNase-seq|Hipe, Chromatin_Structure|Diffa5d|DNase-seq|H7es, Chromatin_Structure||DNase-seq|Hff, Chromatin_Structure||DNase-seq|K562, Chromatin_Structure|Diffa9d|DNase-seq|H7es, Chromatin_Structure|Tam10030|DNase-seq|Ishikawa, Chromatin_Structure||DNase-seq|Hrce, Chromatin_Structure||DNase-seq|Hnpce, Chromatin_Structure||DNase-seq|Hl60, Chromatin_Structure||DNase-seq|Ips, Chromatin_Structure||DNase-seq|Fibrop, Chromatin_Structure||DNase-seq|Nhek, Chromatin_Structure||DNase-seq|T47d, Chromatin_Structure||DNase-seq|Hgf, Chromatin_Structure||DNase-seq|Hsmmt, Chromatin_Structure||DNase-

B93 B94	chr7	135414085	A	G	transition	SLC13A4	5upstream	NA	NA	4	Chromatin_Structure|Znfb34a8|DNase-seq|K562, Chromatin_Structure||DNase-seq|Fibropag08395, Chromatin_Structure|Znfe103c6|DNase-seq|K562, Chromatin_Structure||DNase-seq|Fibroblgm03348, Chromatin_Structure||DNase-seq|Hs5, Chromatin_Structure|Ohurea|FAIRE|K562, Chromatin_Structure||DNase-seq|Hsmm, Chromatin_Structure|Znff41b2|DNase-seq|K562, Chromatin_Structure|Znfa41c6|DNase-seq|K562, Chromatin_Structure|Lenticon|DNase-seq|Fibroblgm03348, Chromatin_Structure||DNase-seq|Fibrobl, Chromatin_Structure||DNase-seq|K562, Chromatin_Structure||DNase-seq|Fibrop, Chromatin_Structure||DNase-seq|Hgf, Chromatin_Structure||DNase-seq|Hsmmt, Chromatin_Structure||FAIRE|K562, Chromatin_Structure||DNase-seq|Hsmmfshd, Chromatin_Structure|Znfg54a11|DNase-seq|K562, Chromatin_Structure|Znf4g7d3|DNase-seq|K562, Protein_Binding|ChIP-Motifs|Footprinting|HepG2|, Chromatin_Structure||DNase-seq|Rptec, Chromatin_Structure||DNase-seq|Gm10248, Chromatin_Structure||FAIRE|Gm18507, Chromatin_Structure||DNase-seq|H7es, Chromatin_Structure||FAIRE|Gm19239, Chromatin_Structure|Diffa5d|DNase-seq|H7es, Chromatin_Structure||DNase-seq|Hrce, Chromatin_Structure|Diffa2d|DNase-seq|H7es, Chromatin_Structure||DNase-seq|A549, Chromatin_Structure||DNase-seq|Huh7, Chromatin_Structure||DNase-seq|Caco2, Chromatin_Structure||DNase-seq|Gm19238, Chromatin_Structure||DNase-seq|Huh75, Chromatin_Structure||DNase-seq|Gm19239, Chromatin_Structure||DNase-seq|Hre, Chromatin_Structure||DNase-seq|Hepg2, Chromatin_Structure||DNase-seq|Gm18507, Chromatin_Structure||DNase-seq|Gm12878, Chromatin_Structure||FAIRE|Hepg2, Chromatin_Structure||FAIRE|A549, Protein_Binding||ChIP-seq|HepG2|EP300, Protein_Binding||ChIP-seq|HepG2|RAD21, Protein_Binding||ChIP-seq|HepG2|NFIC, Protein_Binding||ChIP-seq|HepG2|SP1, Protein_Binding|02pct|ChIP-seq|A549|EP300, Protein_Binding|differential|ChIP-
B83 B84	chr7	138001305	T	C	transition	intergenic		NA	NA	4	
B83 B84	chr1	150338972	A	G	transition	RPRD2	intronic	NA	NA	4	Chromatin_Structure||DNase-seq|Saec, Chromatin_Structure||DNase-seq|Th1, Chromatin_Structure||DNase-seq|Wi38, Chromatin_Structure||DNase-seq|Hipe, Chromatin_Structure||DNase-seq|Nhek, Chromatin_Structure||DNase-seq|Ag10803, Chromatin_Structure||DNase-seq|Hbmec, Chromatin_Structure||DNase-seq|Hsmm, Chromatin_Structure||DNase-seq|Th17, Chromatin_Structure|Ohtam|DNase-seq|Wi38, Chromatin_Structure||DNase-seq|A549, Chromatin_Structure||DNase-seq|Msc, Chromatin_Structure||DNase-seq|Hmf, Chromatin_Structure||DNase-seq|Nhdfad, Chromatin_Structure|Hypoxlac|FAIRE|Mcf7, Chromatin_Structure||DNase-seq|Hsmmt, Chromatin_Structure||DNase-seq|Panislets, Chromatin_Structure||DNase-seq|Hs5, Chromatin_Structure||DNase-seq|Hnpce, Chromatin_Structure||DNase-seq|Gm04504, Chromatin_Structure||DNase-seq|Nha, Chromatin_Structure||DNase-seq|Nhlf, Protein_Binding|02pct|ChIP-seq|A549|FOSL2, Protein_Binding||ChIP-seq|GM19099|POLR2A, Protein_Binding||ChIP-seq|K562|ARID3A,

B53 B54	chr4	155058899	C	A	transversion	intergenic	NA	NA	4	Chromatin_Structure||DNase-seq|H1hesc, Chromatin_Structure||FAIRE|H1hesc, Chromatin_Structure||DNase-seq|Ipsnihi7, Chromatin_Structure|Diffa5d|DNase-seq|H7es, Chromatin_Structure|Tam10030|DNase-seq|Ishikawa, Chromatin_Structure||DNase-seq|Hrce, Chromatin_Structure|Est10nm30m|DNase-seq|Ishikawa, Chromatin_Structure||DNase-seq|Ipsnihi11, Chromatin_Structure||DNase-seq|Ips, Chromatin_Structure||DNase-seq|Hepatocytes, Chromatin_Structure||DNase-seq|Nt2d1, Chromatin_Structure||DNase-seq|Gm19239, Chromatin_Structure||DNase-seq|Phte, Chromatin_Structure||DNase-seq|H9es, Chromatin_Structure||DNase-seq|Ipscwru1, Chromatin_Structure||DNase-seq|Chorion, Chromatin_Structure||DNase-seq|H7es, Chromatin_Structure||DNase-seq|Psoasmuscleoc, Chromatin_Structure||DNase-
B103 B104	chr5	172158355	G	A	transition	intergenic	CpG_site	NA	4	Chromatin_Structure||DNase-seq|Hpde6e6e7, Chromatin_Structure|Ifng4h|FAIRE|Helas3, Chromatin_Structure|Hypoxlaccon|DNase-seq|Mcf7, Chromatin_Structure||DNase-seq|Lncap, Chromatin_Structure||DNase-seq|K562G2mphase, Chromatin_Structure|Ctcfshrna|DNase-seq|Mcf7, Chromatin_Structure||FAIRE|Gm12892, Chromatin_Structure||DNase-seq|Panislets, Chromatin_Structure|Ifna4h|FAIRE|Helas3, Chromatin_Structure||DNase-seq|K562, Chromatin_Structure|Andro|DNase-seq|Lncap, Chromatin_Structure||DNase-seq|Mcf7, Chromatin_Structure||DNase-seq|T47d, Chromatin_Structure||DNase-seq|A549, Chromatin_Structure||DNase-seq|Huh7, Chromatin_Structure||DNase-seq|Helas3, Chromatin_Structure|Ifna4h|DNase-seq|Helas3, Chromatin_Structure||DNase-seq|Hek293t, Chromatin_Structure||DNase-seq|Urotsa, Chromatin_Structure|Est10nm30m|DNase-seq|T47d, Chromatin_Structure||DNase-seq|Rwpe1, Chromatin_Structure||DNase-seq|Huh75, Chromatin_Structure||DNase-seq|Hmec, Chromatin_Structure|Est10nm30m|DNase-seq|Ecc1, Chromatin_Structure||DNase-seq|Phte, Chromatin_Structure|Hypoxlac|DNase-seq|Mcf7, Chromatin_Structure|Hypoxlac|FAIRE|Mcf7, Chromatin_Structure||DNase-seq|Th1, Chromatin_Structure||DNase-seq|K562G1phase, Chromatin_Structure||DNase-seq|Chorion, Chromatin_Structure|Dm002p1h|DNase-seq|Ecc1, Chromatin_Structure||DNase-seq|Gm12892, Chromatin_Structure|Randshrna|DNase-seq|Mcf7, Protein_Binding||ChIP-seq|HeLa-S3|CHD2, Protein_Binding||ChIP-seq|HepG2|MAX, Protein_Binding||ChIP-seq|K562|JUND, Protein_Binding||ChIP-seq|HepG2|SIN3A, Protein_Binding||ChIP-seq|HeLa-S3|SMARCC1, Protein_Binding||ChIP-seq|HeLa-S3|MYC, Protein_Binding|differential|ChIP-seq|Caco2|CDX2, Protein_Binding||ChIP-seq|HeLa-S3|CEBPB, Protein_Binding||ChIP-seq|K562|MAX, Protein_Binding||ChIP-seq|K562|RCOR1

B83 B84	chr2	209224680	C	T	transition	PIKFYVE	3downstream	NA	NA	4	Chromatin_Structure||DNase-seq|H1hesc, Chromatin_Structure||DNase-seq|K562, Chromatin_Structure||DNase-seq|Olfneurosphere, Chromatin_Structure||DNase-seq|H9es, Chromatin_Structure||DNase-seq|Ipscwru1, Chromatin_Structure||DNase-seq|Cd20ro01794, Chromatin_Structure||DNase-seq|Ipsnihi11, Chromatin_Structure||DNase-seq|Ips, Chromatin_Structure||DNase-seq|Chorion, Chromatin_Structure||DNase-seq|Myometr, Chromatin_Structure||DNase-seq|Sknsh, Chromatin_Structure||DNase-seq|H7es, Chromatin_Structure||DNase-seq|Monocd14, Chromatin_Structure||DNase-seq|Huh7, Chromatin_Structure||DNase-seq|Hmf, Chromatin_Structure||DNase-seq|Cerebrumfrontaloc, Protein_Binding||ChIP-seq|GM12878|POLR2A,
B103 B104	chr8	674731	A	G	transition	ERICH1	intronic	NA	NA	5	Chromatin_Structure||DNase-seq|Ipsnihi7, Chromatin_Structure||DNase-seq|Cerebrumfrontaloc, Chromatin_Structure||DNase-seq|Ipscwru1, Chromatin_Structure||DNase-seq|H7es, Chromatin_Structure||DNase-seq|H1hesc, Chromatin_Structure||DNase-seq|Ips, Chromatin_Structure||DNase-seq|H9es, Chromatin_Structure||DNase-seq|Chorion
B123 B124	chr8	1779112	G	A	transition	ARHGEF10	intronic	CpG_site	NA	5	Chromatin_Structure||DNase-seq|Medullod341, Chromatin_Structure||DNase-seq|H7es, Chromatin_Structure||DNase-seq|Monocd14, Chromatin_Structure||DNase-seq|H1hesc, Chromatin_Structure|Ifna4h|FAIRE|Helas3, Chromatin_Structure||DNase-seq|K562, Chromatin_Structure||DNase-seq|Ips, Chromatin_Structure||DNase-seq|Hepatocytes, Chromatin_Structure||DNase-seq|Frontalcortexoc, Chromatin_Structure||DNase-seq|Gm19239, Chromatin_Structure||DNase-seq|Osteobl, Chromatin_Structure|Est10nm30m|DNase-seq|Ecc1, Chromatin_Structure||DNase-seq|Th1, Chromatin_Structure||DNase-seq|Medullo,
B62 B63	chr4	1883359	A	G	transition	WHSC1	intronic	NA	NA	5	Chromatin_Structure||DNase-seq|Hpdlf, Chromatin_Structure||DNase-seq|Ag10803, Chromatin_Structure||DNase-seq|Hgf, Chromatin_Structure||DNase-seq|Nhdfneo, Chromatin_Structure||DNase-seq|Nhdfad
B123 B124	chr4	2463556	C	T	transition	RP11_503N18.	intronic	NA	NA	5	Chromatin_Structure||DNase-seq|Progfib, Chromatin_Structure||DNase-seq|H1hesc, Chromatin_Structure||DNase-seq|Nb4, Chromatin_Structure||DNase-seq|Adultcd4th0, Chromatin_Structure||FAIRE|Gm19239, Chromatin_Structure|Diffa5d|DNase-seq|H7es, Chromatin_Structure||DNase-seq|K562, Chromatin_Structure||DNase-seq|Adultcd4th1, Chromatin_Structure|Andro|DNase-seq|Lncap, Chromatin_Structure||DNase-seq|Monocd14ro1746, Chromatin_Structure||DNase-seq|Hepatocytes, Chromatin_Structure||DNase-seq|Gm10266, Chromatin_Structure||DNase-seq|Frontalcortexoc, Chromatin_Structure||DNase-seq|Huh7, Chromatin_Structure||DNase-seq|Lncap, Chromatin_Structure||DNase-seq|Gm19238, Chromatin_Structure|Znfp5|DNase-seq|K562, Chromatin_Structure||DNase-seq|Gm12891, Chromatin_Structure||DNase-seq|8988t, Chromatin_Structure||DNase-seq|Huh75, Chromatin_Structure||DNase-seq|Hmec, Chromatin_Structure||DNase-seq|Osteobl, Chromatin_Structure||DNase-seq|Gm13977, Chromatin_Structure||DNase-seq|Medullo, Chromatin_Structure||DNase-seq|Chorion, Chromatin_Structure||DNase-seq|Gm20000, Chromatin_Structure||DNase-seq|Gm12892,

B93 B94	chr5	3908422	C	T	transition	intergenic		NA	NA	5	Motifs|PWM||SPIB, Motifs|PWM||PU.1, Motifs|PWM||Elf5, Motifs|PWM||SPIC, Motifs|PWM||SOX, Motifs|PWM||Sfpi1, Motifs|PWM||PU1, Motifs|PWM||SPI1, Motifs|PWM||Spic, Motifs|PWM||ELF5, Motifs|PWM||SPI-B, Chromatin_Structure||DNase-seq|T47d
B93 B94	chr2	6263593	C	T	transition	intergenic		NA	NA	5	Chromatin_Structure||DNase-seq|Werirb1
LJ LS	chr1	7523207	G	A	transition	CAMTA1	intronic	CpG_site	NA	5	Motifs|PWM||Dax1, Motifs|PWM||RARG, Motifs|PWM||Zic3, Motifs|Footprinting|Gm12892|PPARG::RXRA, Motifs|PWM||PPARG::RXRA, Chromatin_Structure||DNase-seq|Sknsh, Chromatin_Structure||DNase-seq|H7es, Chromatin_Structure||DNase-seq|Werirb1
LJ LS	chr19	8468273	C	T	transition	RAB11B	intronic	CpG_site	NA	5	Motifs|Footprinting|H1hesc|SREBP-1, Motifs|PWM||VDR, Motifs|Footprinting|Hsmmt|SREBP-1, Motifs|Footprinting|Gm12892|SREBP, Motifs|Footprinting|8988t|SREBP, Motifs|Footprinting|Lncap|SREBP-1, Motifs|Footprinting|Huh7|SREBP-1, Motifs|Footprinting|Gm12878|SREBP-1, Motifs|Footprinting|Gm12891|SREBP, Motifs|Footprinting|Huh7|SREBP, Motifs|Footprinting|Gm12891|SREBP-1, Motifs|Footprinting|Gm12878|SREBP, Motifs|PWM||SREBP, Motifs|Footprinting|Hpde6e6e7|SREBP, Motifs|Footprinting|LncapAndro|SREBP, Motifs|Footprinting|Chorion|SREBP, Motifs|Footprinting|LncapAndro|SREBP-1, Motifs|Footprinting|Osteobl|SREBP, Motifs|Footprinting|Gm12892|SREBP-1, Motifs|PWM||SREBP1, Motifs|Footprinting|Myometr|SREBP, Motifs|Footprinting|Hpde6e6e7|SREBP-1, Motifs|Footprinting|Hepatocytes|SREBP, Motifs|Footprinting|H1hesc|SREBP, Motifs|Footprinting|A549|SREBP-1, Motifs|Footprinting|Hsmm|SREBP, Motifs|Footprinting|Cll|SREBP, Motifs|PWM||SREBP-1, Motifs|Footprinting|Osteobl|SREBP-1, Motifs|Footprinting|Lncap|SREBP, Motifs|Footprinting|Chorion|SREBP-1, Chromatin_Structure||DNase-seq|Gm12891, Chromatin_Structure||DNase-seq|H1hesc, Chromatin_Structure||DNase-seq|Osteobl, Chromatin_Structure||DNase-seq|K562, Chromatin_Structure||DNase-seq|Imr90, Chromatin_Structure||DNase-seq|Hepg2, Chromatin_Structure||DNase-seq|Th1, Chromatin_Structure||DNase-seq|Cd20ro01794, Chromatin_Structure||DNase-seq|Ips, Chromatin_Structure||DNase-seq|Chorion, Chromatin_Structure||DNase-seq|Gm12892, Chromatin_Structure||DNase-seq|Frontalcortexoc, Chromatin_Structure||DNase-seq|H7es, Chromatin_Structure||DNase-
B93 B94	chr18	12057323	A	C	transversion	intergenic		NA	NA	5	Motifs|PWM||Pit-1, Motifs|Footprinting|H1hesc|Pit-1, Motifs|Footprinting|Gm12891|Pit-1, Motifs|Footprinting|Huh7|Pit-1, Motifs|Footprinting|LncapAndro|Pit-1, Motifs|Footprinting|Chorion|Pit-1, Chromatin_Structure||DNase-seq|Medullod341, Chromatin_Structure||DNase-seq|Huh7, Chromatin_Structure||DNase-seq|Huh75, Chromatin_Structure||DNase-seq|Osteobl, Chromatin_Structure||DNase-seq|Medullo,
B124	chr17	12638660	T	C	transition	MYOCD	intronic	NA	NA	5	Chromatin_Structure||DNase-seq|Hcf, Chromatin_Structure||DNase-seq|Osteobl
B124	chr17	12638661	A	G	transition	MYOCD	intronic	NA	NA	5	Chromatin_Structure||DNase-seq|Hcf, Chromatin_Structure||DNase-seq|Osteobl

B123 B124	chr3	13085170	C	T	transition	IQSEC1	intronic	CpG_site	NA	5	Chromatin_Structure||DNase-seq|K562, Chromatin_Structure||DNase-seq|Adultcd4th1, Chromatin_Structure||DNase-seq|Hepatocytes, Chromatin_Structure||DNase-seq|T47d, Chromatin_Structure||DNase-seq|Huh7, Chromatin_Structure|Nabut|DNase-seq|K562, Chromatin_Structure||DNase-seq|8988t, Chromatin_Structure||DNase-seq|Huh75, Chromatin_Structure||DNase-seq|Th1, Chromatin_Structure||DNase-seq|Medullo, Chromatin_Structure||DNase-seq|Chorion, Chromatin_Structure||DNase-seq|Psoasmuscleoc,
B53 B54	chr9	16205585	C	T	transition	C9orf92	intronic	NA	NA	5	Motifs|PWM||EWSR1-FLI1, Chromatin_Structure||DNase-seq|Hcf, Chromatin_Structure||DNase-seq|Fibroblgm03348, Chromatin_Structure||DNase-seq|Hmf, Chromatin_Structure||DNase-seq|Hpaf, Chromatin_Structure||DNase-seq|Fibrobl, Chromatin_Structure||DNase-seq|Hcm, Chromatin_Structure||DNase-seq|Nhdfad, Chromatin_Structure||DNase-seq|Aoaf
B53 B54	chr5	16385105	T	C	transition	RP1_167G20.1	non-coding intronic	NA	NA	5	Chromatin_Structure|Znfa41c6|DNase-seq|K562, Chromatin_Structure||DNase-seq|K562
B113 B114	chr11	16923047	T	A	transversion	PLEKHA7	intronic	NA	NA	5	Motifs|PWM||Nanog, Chromatin_Structure||DNase-seq|Melano, Chromatin_Structure||FAIRE|Gm12892
B103 B104	chr9	17578171	A	C	transversion	SH3GL2	5upstream	NA	NA	5	Chromatin_Structure||DNase-seq|Hepatocytes, Chromatin_Structure||DNase-seq|Helas3, Chromatin_Structure|Ifna4h|DNase-seq|Helas3
B103 B104	chr16	19266444	C	T	transition	SYT17	intronic	NA	NA	5	Chromatin_Structure||DNase-seq|Ips
B53 B54	chr5	19663794	A	G	transition	CDH18	intronic	NA	NA	5	Motifs|PWM||MEF-2, Protein_Binding||ChIP-seq|HSPC|SPI1
B103 B104	chr20	19700562	G	A	transition	SLC24A3	intronic	NA	NA	5	Chromatin_Structure||DNase-seq|Cll
B103 B104	chr17	20145251	C	T	transition	SPECC1	intronic	NA	NA	5	Chromatin_Structure|Ctcfshrna|DNase-seq|Mcf7, Chromatin_Structure||DNase-seq|Mcf7
B53 B54	chr7	20169410	C	T	transition	AC005062.2	non-coding intronic	CpG_site	NA	5	Chromatin_Structure||DNase-seq|H7es, Chromatin_Structure|Diffa5d|DNase-seq|H7es
B93 B94	chr12	20304160	A	G	transition	intergenic		NA	NA	5	Chromatin_Structure|Est10nm30m|DNase-seq|Ishikawa, Chromatin_Structure||DNase-seq|Nt2d1, Chromatin_Structure|Tam10030|DNase-seq|Ishikawa, Chromatin_Structure|Est10nm30m|DNase-seq|Ecc1, Chromatin_Structure||FAIRE|Gm12891, Chromatin_Structure|Dm002p1h|DNase-seq|Ecc1
B103 B104	chr7	21283412	G	A	transition	intergenic		CpG_site	NA	5	Chromatin_Structure||DNase-seq|Th17, Chromatin_Structure||DNase-seq|Th1
B83 B84	chr12	21661853	A	C	transversion	GOLT1B	intronic	NA	NA	5	Chromatin_Structure||FAIRE|Gm18507, Chromatin_Structure||DNase-seq|Hl60
B93 B94	chr6	21747135	C	T	transition	CASC15	non-coding intronic	NA	NA	5	Chromatin_Structure||DNase-seq|H7es

B62 B63	chr22	22003746	T	A	transversion	intergenic	NA	NA	5	Motifs|Footprinting|Helas3|HNF3beta, Motifs|PWM||HFH8(FOXF1A), Motifs|Footprinting|H1hesc|FOXD3, Motifs|Footprinting|Gm12892|HNF3beta, Motifs|Footprinting|Gm12892|HFH4(FOXJ1), Motifs|Footprinting|Gm12892|HFH3(FOXI1), Motifs|PWM||HNF3beta, Motifs|PWM||HNF3, Motifs|PWM||HFH4(FOXJ1), Motifs|Footprinting|Gm12878|HNF3beta, Motifs|PWM||Foxj3, Motifs|Footprinting|Helas3|HFH8(FOXF1A), Motifs|Footprinting|Chorion|HNF3beta, Motifs|Footprinting|Chorion|Foxd3, Motifs|PWM||FOXC1, Motifs|Footprinting|Helas3|FOXfactors, Motifs|Footprinting|Helas3|FOXO3A, Motifs|Footprinting|Chorion|Freac-7, Motifs|Footprinting|Gm12878|FOXI1, Motifs|Footprinting|K562|FOXO3A, Motifs|Footprinting|Gm12892|Freac-7, Motifs|PWM||Dbx1, Motifs|PWM||Freac-7, Motifs|Footprinting|Gm12878|HFH4(FOXJ1), Motifs|Footprinting|Gm12891|Cdx, Motifs|Footprinting|Gm12892|FOXO3A, Motifs|Footprinting|Helas3|FOXI1, Motifs|PWM||Cdx, Motifs|Footprinting|Gm12878|FOXO3A, Motifs|Footprinting|K562|HFH4(FOXJ1), Motifs|PWM||ONECUT3, Motifs|PWM||Tlx2, Motifs|Footprinting|K562|HNF3, Motifs|Footprinting|Gm12892|HFH8(FOXF1A), Motifs|Footprinting|Gm12878|FOXfactors, Motifs|Footprinting|Gm12878|Foxd3, Motifs|Footprinting|Huh7|HFH3(FOXI1), Motifs|Footprinting|Gm12892|Cdx, Motifs|Footprinting|H1hesc|HNF3, Motifs|PWM||Tbp, Motifs|PWM||FOXC2, Motifs|PWM||FOXO3A, Motifs|Footprinting|H1hesc|HFH3(FOXI1), Motifs|Footprinting|Helas3|HFH4(FOXJ1), Motifs|PWM||FOXfactors, Motifs|PWM||Foxl1, Motifs|Footprinting|K562|Cdx, Motifs|Footprinting|Chorion|HFH4(FOXJ1), Motifs|Footprinting|H1hesc|Freac-7, Motifs|Footprinting|Helas3|Cdx, Motifs|PWM||FOXP1, Motifs|Footprinting|Chorion|HFH3(FOXI1), Motifs|Footprinting|H1hesc|Foxd3, Motifs|Footprinting|Helas3|HFH3(FOXI1), Motifs|Footprinting|Gm12878|Freac-7, Motifs|Footprinting|Gm12878|Cdx, Motifs|PWM||FOXJ2, Motifs|Footprinting|Gm12878|HFH8(FOXF1A), Motifs|Footprinting|Chorion|Cdx, Motifs|Footprinting|K562|HFH3(FOXI1), Motifs|Footprinting|Gm12892|FOXI1, Motifs|PWM||Hoxd8, Motifs|Footprinting|Chorion|HFH8(FOXF1A), Motifs|Footprinting|Chorion|FOXO3A, Motifs|Footprinting|Gm12878|HNF3, Motifs|Footprinting|Chorion|FOXfactors, Motifs|Footprinting|Helas3|Freac-7, Motifs|Footprinting|Chorion|HNF3, Motifs|Footprinting|Gm12892|FOXfactors, Motifs|Footprinting|H1hesc|HFH4(FOXJ1), Motifs|PWM||FOXL1, Motifs|PWM||FOXD3, Motifs|PWM||FOXI1, Motifs|PWM||Foxd3,

B62 B63	chr1	23120852	C	T	transition	EPHB2	intronic	CpG_site	NA	5	Motifs|Footprinting|Gm12892|HSF, Motifs|Footprinting|Gm12892|HSF1, Motifs|PWM||HSF1, Motifs|Footprinting|Gm12891|HSF1, Motifs|Footprinting|Hsmm|HSF1, Motifs|Footprinting|Hsmmt|HSF1, Motifs|PWM||HSF, Motifs|Footprinting|Osteobl|HSF1, Motifs|Footprinting|Hsmmt|HSF, Motifs|Footprinting|Gm12891|HSF, Chromatin_Structure||DNase-seq|Gm12891, Chromatin_Structure||DNase-seq|Cd20ro01794, Chromatin_Structure||DNase-seq|Osteobl, Chromatin_Structure||DNase-seq|Gm12892, Chromatin_Structure||DNase-seq|K562, Chromatin_Structure||DNase-seq|Hsmm, Chromatin_Structure||DNase-seq|Monocd14, Chromatin_Structure|Nabut|DNase-seq|K562, Chromatin_Structure||DNase-seq|K562G2mphase, Chromatin_Structure||DNase-
LJ LS	chr10	24822488	T	C	transition	KIAA1217	intronic	NA	NA	5	Motifs|PWM||Pitx1, Chromatin_Structure||DNase-seq|Hbvp
B103 B104	chr8	25459305	G	T	transversion	intergenic		NA	NA	5	Motifs|PWM||FOXG1, Chromatin_Structure||DNase-seq|Hmveclbl, Chromatin_Structure||DNase-seq|Hmvecdblad, Chromatin_Structure||DNase-seq|Mel2183, Chromatin_Structure||DNase-seq|Colo829
B113 B114	chr17	26144055	A	G	transition	intergenic		NA	NA	5	Chromatin_Structure||DNase-seq|Werirb1
LJ LS	chr4	27144101	G	A	transition	intergenic		CpG_site	NA	5	Chromatin_Structure|Est10nm30m|DNase-seq|Ishikawa, Chromatin_Structure||DNase-seq|H7es, Chromatin_Structure|Tam10030|DNase-seq|Ishikawa, Chromatin_Structure||DNase-seq|H9es
B113 B114	chr13	28557037	C	T	transition	URAD	intronic	NA	NA	5	Motifs|Footprinting|Hepatocytes|STAT5A(homotetramer), Motifs|PWM||STAT5A(homotetramer), Chromatin_Structure||DNase-seq|Hepatocytes, Chromatin_Structure||DNase-seq|Hmec, Chromatin_Structure||DNase-seq|Myometr
B83 B84	chr4	29361176	T	A	transversion	intergenic		NA	NA	5	Motifs|PWM||Cdx, Motifs|PWM||Sox4, Motifs|PWM||FOXP1, Chromatin_Structure||DNase-seq|Monocd14ro1746, Chromatin_Structure||DNase-seq|Monocd14
B93 B94	chr3	30351562	T	C	transition	intergenic		NA	NA	5	Chromatin_Structure||DNase-seq|H7es
B62 B63	chr11	30391709	A	C	transversion	intergenic		NA	NA	5	Motifs|PWM||NR2E1, Motifs|PWM||FOXC2, Protein_Binding||ChIP-seq|VCaP|AR
B83 B84	chr14	35915225	C	T	transition	intergenic		NA	NA	5	Chromatin_Structure||DNase-seq|Jurkat
LJ LS	chr3	36418949	C	T	transition	intergenic		NA	NA	5	Chromatin_Structure||DNase-seq|Rptec
B93 B94	chr11	36428450	G	C	transversion	PRR5L	intronic	NA	NA	5	Chromatin_Structure||DNase-seq|Hgf
B93 B94	chr11	36428499	C	A	transversion	PRR5L	intronic	NA	NA	5	Chromatin_Structure||DNase-seq|Hgf
LJ LS	chr7	36863766	C	T	transition	intergenic		NA	NA	5	Chromatin_Structure||DNase-seq|Werirb1
LJ LS	chr6	37921491	G	A	transition	ZFAND3	intronic	NA	NA	5	Chromatin_Structure||DNase-seq|M059j, Chromatin_Structure||DNase-seq|Hae, Chromatin_Structure||DNase-seq|Hmf, Chromatin_Structure||DNase-seq|Hipe, Chromatin_Structure||DNase-seq|Werirb1, Chromatin_Structure||DNase-seq|Nhdfad, Chromatin_Structure||DNase-seq|Be2c
B53 B54	chr17	39595049	G	T	transversion	KRT38	nonsyn p.P265H	NA	NA	5	Motifs|PWM||HNF4A, Chromatin_Structure||DNase-seq|Gm10248, Chromatin_Structure||DNase-seq|Medullod341, Chromatin_Structure||DNase-seq|Gm13976, Chromatin_Structure||DNase-seq|Adultcd4th1, Chromatin_Structure||DNase-seq|Mcf7, Chromatin_Structure|Hypoxlac|DNase-seq|Mcf7, Chromatin_Structure||DNase-seq|Medullo, Chromatin_Structure||DNase-seq|Gm12892
B83 B84	chr6	40413089	G	A	transition	LRFN2	intronic	CpG_site	NA	5	Chromatin_Structure||DNase-seq|Nhek
B123 B124	chr21	40959364	T	C	transition	intergenic		NA	NA	5	Motifs|PWM||Srf, Protein_Binding||ChIP-seq|K562|EP300, Protein_Binding||ChIP-seq|K562|TAL1
B93 B94	chr8	41046737	A	T	transversion	intergenic		NA	NA	5	Motifs|PWM||NF-Y, Motifs|PWM||alpha-CP1, Chromatin_Structure||DNase-seq|M059j, Chromatin_Structure||DNase-seq|H7es, Chromatin_Structure|Diffa5d|DNase-seq|H7es

B53 B54	chr13	42917019	C	G	transversion	intergenic		NA	NA	5	Chromatin_Structure||DNase-seq|Monocd14ro1746, Chromatin_Structure||DNase-seq|Monocd14
B53 B54	chr22	43077904	T	C	transition	intergenic		NA	NA	5	Protein_Binding|proliferation|ChIP-seq|Caco2|HNF4A
B83 B84	chr14	43204114	A	G	transition	intergenic		NA	NA	5	Motifs|PWM||Freac-7, Motifs|PWM||FOXJ3, Motifs|PWM||TEF, Motifs|PWM||FOXJ2, Chromatin_Structure||DNase-seq|Hffmyc, Chromatin_Structure||DNase-seq|Hff
B93 B94	chr1	43646521	C	T	transition	WDR65	intronic	NA	NA	5	Motifs|PWM||VDR, Chromatin_Structure||DNase-seq|Th1, Chromatin_Structure||DNase-seq|Th17
B62 B63	chr22	45313501	A	G	transition	PHF21B	intronic	NA	NA	5	Motifs|PWM||GATA-6, Motifs|Footprinting|Cll|GATA-6, Motifs|Footprinting|H1hesc|GATA-6, Motifs|PWM||Gata1, Motifs|Footprinting|Fibrop|GATA-6, Motifs|Footprinting|Hsmm|GATA-6, Motifs|Footprinting|Hsmm|Gata1, Motifs|Footprinting|Gm12892|Gata1, Motifs|Footprinting|Chorion|Gata1, Motifs|Footprinting|GM12892|Nrf-2, Motifs|Footprinting|Hsmmt|GATA-6, Motifs|Footprinting|8988t|GATA-6, Motifs|Footprinting|Lymphoblastoid|Nrf-2, Motifs|Footprinting|LncapAndro|Gata1, Motifs|Footprinting|Lncap|GATA-6, Motifs|Footprinting|Lymphoblastoid|Eip74EF, Motifs|Footprinting|GM12892|Eip74EF, Motifs|Footprinting|Gm12892|GATA-6, Motifs|Footprinting|Lymphoblastoid|dl, Motifs|Footprinting|Lymphoblastoid|GABPA, Motifs|Footprinting|Lymphoblastoid|ELK4, Motifs|Footprinting|Chorion|GATA-6, Motifs|Footprinting|Lymphoblastoid|dl_1, Motifs|Footprinting|Hepatocytes|GATA-6, Motifs|Footprinting|LncapAndro|GATA-6, Motifs|Footprinting|H1hesc|Gata1, Motifs|Footprinting|GM12892|ELK4, Motifs|Footprinting|GM12892|GABPA, Chromatin_Structure||DNase-seq|Cd20ro01794, Chromatin_Structure||DNase-seq|Ips, Chromatin_Structure||DNase-seq|Chorion, Chromatin_Structure||DNase-seq|Hepatocytes, Chromatin_Structure||DNase-seq|Gm12892, Chromatin_Structure||DNase-seq|H7es, Chromatin_Structure||DNase-seq|Hsmm, Chromatin_Structure||DNase-seq|Psoasmuscleoc, Chromatin_Structure||DNase-seq|Monocd14, Chromatin_Structure||DNase-seq|Lncap, Chromatin_Structure||DNase-seq|Hsmmt, Chromatin_Structure||DNase-
B123 B124	chr11	47691714	A	G	transition	AGBL2	intronic	NA	NA	5	Chromatin_Structure||DNase-seq|Werirb1, Chromatin_Structure||DNase-seq|Cerebellumoc
B113 B114	chr22	49740309	G	C	transversion	intergenic		CpG_site	NA	5	Motifs|PWM||AP-2gamma, Motifs|PWM||TCFAP2A, Motifs|PWM||AP-2alpha, Motifs|PWM||HIC1, Motifs|PWM||Zic3, Motifs|PWM||TFAP2C, Motifs|PWM||TFAP2A, Chromatin_Structure||DNase-seq|Hah, Chromatin_Structure||DNase-seq|Phte, Chromatin_Structure||DNase-
LJ LS	chr7	50434049	G	A	transition	IKZF1	intronic	NA	NA	5	Chromatin_Structure||DNase-seq|Cd4naivewb11970640
B93 B94	chr3	50574244	T	C	transition	intergenic		NA	NA	5	Chromatin_Structure||DNase-seq|Hbvp, Chromatin_Structure||FAIRE|UrotsaUt189, Chromatin_Structure||DNase-seq|Cerebrumfrontaloc, Chromatin_Structure||DNase-seq|8988t
LJ LS	chr20	50688780	G	T	transversion	intergenic		NA	NA	5	Protein_Binding||ChIP-seq|HepG2|MAFK
B123 B124	chr3	51651550	C	T	transition	RAD54L2	intronic	NA	NA	5	Motifs|PWM||Hoxb13, Motifs|PWM||Hoxd10, Motifs|PWM||Hoxd13, Chromatin_Structure||DNase-seq|Monocd14ro1746, Chromatin_Structure||DNase-seq|Medullo, Chromatin_Structure||DNase-seq|Monocd14
B103 B104	chr4	53162451	C	T	transition	intergenic		NA	NA	5	Chromatin_Structure|Tam10030|DNase-seq|Ishikawa, Chromatin_Structure|Est10nm30m|DNase-seq|Ishikawa

B83 B84	chr5	54356988	C	G	transversion	intergenic		CpG_site	NA	5	Chromatin_Structure||DNase-seq|Th17, Chromatin_Structure||DNase-seq|Th2, Chromatin_Structure||DNase-seq|Th2wb54553204, Chromatin_Structure||DNase-seq|Th1
LJ LS	chr19	57113990	G	T	transversion	ZNF71	intronic	NA	NA	5	Chromatin_Structure||DNase-seq|Panislets
B83 B84	chr4	57295012	G	A	transition	PPAT	intronic	NA	NA	5	Chromatin_Structure||DNase-seq|K562, Chromatin_Structure|Znf4c50c4|DNase-seq|K562, Chromatin_Structure||DNase-seq|Hrgec
B62 B63	chr11	60294628	C	T	transition	MS4A13	intronic	NA	NA	5	Chromatin_Structure||DNase-seq|Hs27a
B53 B54	chr5	61635205	C	T	transition	KIF2A	intronic	NA	NA	5	Chromatin_Structure||DNase-seq|Th1wb33676984, Chromatin_Structure||DNase-seq|Th1
B113 B114	chr15	62958974	T	C	transition	TLN2	intronic	NA	NA	5	Motifs|PWM||Nr2f2, Motifs|PWM||RARA, Motifs|PWM||NR2F6, Motifs|PWM||Rara, Motifs|PWM||NUR77, Motifs|PWM||RARG, Motifs|PWM||NR2F1, Motifs|PWM||RARB, Motifs|PWM||PPARgamma:RXRalpha, Chromatin_Structure||DNase-seq|H7es
B93 B94	chr5	65445453	C	G	transversion	SREK1	intronic	NA	NA	5	Motifs|PWM||Oct-1, Motifs|PWM||FOXD3, Motifs|PWM||FOXD2, Chromatin_Structure||DNase-seq|Hpdlf, Chromatin_Structure||DNase-seq|Hcf, Chromatin_Structure||DNase-seq|Ag09319, Chromatin_Structure||DNase-seq|Hcpe, Chromatin_Structure||DNase-seq|Ag10803, Chromatin_Structure||DNase-seq|Gm04504, Chromatin_Structure||DNase-seq|Hcfaa, Chromatin_Structure||DNase-seq|Hah, Chromatin_Structure||DNase-seq|Hpf, Chromatin_Structure||DNase-seq|Wi38, Chromatin_Structure||DNase-seq|Hvmf, Chromatin_Structure||DNase-seq|Hae, Chromatin_Structure||DNase-seq|Ag04450, Chromatin_Structure||DNase-seq|Hsmm, Chromatin_Structure||DNase-seq|Ag09309, Chromatin_Structure||DNase-seq|Hmf, Chromatin_Structure||DNase-seq|Hpaf, Chromatin_Structure||DNase-seq|Skmc, Chromatin_Structure||DNase-seq|Fibrobl, Chromatin_Structure||DNase-seq|Hipe, Chromatin_Structure||DNase-seq|Hnpce, Chromatin_Structure||DNase-seq|Hl60, Chromatin_Structure||DNase-seq|Hgf, Chromatin_Structure||DNase-seq|Nha, Chromatin_Structure||DNase-seq|Gm04503, Chromatin_Structure||DNase-seq|Hconf, Chromatin_Structure||DNase-seq|Nhdfneo, Chromatin_Structure||DNase-seq|Hcm, Chromatin_Structure||DNase-seq|Hee, Chromatin_Structure||DNase-seq|Imr90, Chromatin_Structure||DNase-seq|Ag04449, Chromatin_Structure||DNase-seq|Nhlf
B93 B94	chr17	65581782	G	A	transition	PITPNC1	intronic	NA	NA	5	Motifs|PWM||RP58, Motifs|PWM||ZNF238, Protein_Binding||ChIP-seq|HEK293-T-REx|ZNF263, Protein_Binding||ChIP-seq|U2OS|TRIM28
B123 B124	chr2	67362239	G	A	transition	AC078941.1	non-coding intronic	CpG_site	NA	5	Protein_Binding||ChIP-seq|H1-hESC|YY1
B103 B104	chr3	68974336	T	C	transition	FAM19A4	intronic	NA	NA	5	Motifs|PWM||Cdx, Protein_Binding||ChIP-seq|HepG2|CEBPB, Protein_Binding||ChIP-seq|HepG2|FOXA1
B62 B63	chr11	71038704	G	A	transition	intergenic		NA	NA	5	Motifs|PWM||CTCF, Chromatin_Structure||DNase-seq|8988t, Chromatin_Structure||DNase-seq|Monocd14, Chromatin_Structure||DNase-seq|Huh7, Chromatin_Structure||DNase-seq|Caco2
B93 B94	chr2	73229215	T	A	transversion	SFXN5	intronic	NA	NA	5	Motifs|PWM||IRF7, Motifs|PWM||TCF-4, Chromatin_Structure||DNase-seq|Hepg2
B123 B124	chr12	75146156	T	C	transition	intergenic		NA	NA	5	Protein_Binding||ChIP-seq|HUVEC|GATA2
B123 B124	chr16	77170825	G	A	transition	intergenic		NA	NA	5	Chromatin_Structure||DNase-seq|Mcf7

B53 B54	chr15	77314816	G	A	transition	PSTPIP1	intronic	NA	NA	5	Chromatin_Structure||DNase-seq|Progfib, Chromatin_Structure||DNase-seq|Htr8, Chromatin_Structure||DNase-seq|Fibropag08395, Chromatin_Structure||DNase-seq|Huvec, Chromatin_Structure||DNase-seq|Heartoc, Chromatin_Structure||DNase-seq|UrotsaUt189, Chromatin_Structure||DNase-seq|Panisd, Chromatin_Structure||DNase-seq|Fibroblgm03348, Chromatin_Structure||DNase-seq|Cerebrumfrontaloc, Chromatin_Structure||DNase-seq|Gm12891, Chromatin_Structure||DNase-seq|8988t, Chromatin_Structure||DNase-seq|Gm19240, Chromatin_Structure||DNase-seq|Melano, Chromatin_Structure||DNase-seq|Stellate, Chromatin_Structure||DNase-seq|Gm10248, Chromatin_Structure||DNase-seq|Medullod341, Chromatin_Structure||DNase-seq|Gm20000, Chromatin_Structure||DNase-seq|H7es, Chromatin_Structure||DNase-seq|Fibropag08396, Chromatin_Structure||DNase-seq|Hsmm, Chromatin_Structure||DNase-seq|Gm13976, Chromatin_Structure|Lenticon|DNase-seq|Fibroblgm03348, Chromatin_Structure|Ifna4h|FAIRE|Helas3, Chromatin_Structure||DNase-seq|Adultcd4th0, Chromatin_Structure||DNase-seq|Fibrobl, Chromatin_Structure||DNase-seq|Adultcd4th1, Chromatin_Structure||DNase-seq|Werirb1, Chromatin_Structure||DNase-seq|Fibrop, Chromatin_Structure||DNase-seq|Hepatocytes, Chromatin_Structure||DNase-seq|Gm10266, Chromatin_Structure||DNase-seq|A549, Chromatin_Structure||DNase-seq|Huh7, Chromatin_Structure||DNase-seq|Helas3, Chromatin_Structure||DNase-seq|Hsmmt, Chromatin_Structure||DNase-seq|Gm19238, Chromatin_Structure|Ifna4h|DNase-seq|Helas3, Chromatin_Structure||DNase-seq|Urotsa, Chromatin_Structure||DNase-seq|Gm19239, Chromatin_Structure||DNase-seq|Huh75, Chromatin_Structure||DNase-seq|Phte, Chromatin_Structure||DNase-seq|Gm13977, Chromatin_Structure||DNase-seq|Imr90, Chromatin_Structure|Lentimyod|DNase-seq|Fibroblgm03348, Chromatin_Structure||DNase-seq|Hsmmfshd, Chromatin_Structure||DNase-seq|Hsmmemb, Chromatin_Structure||DNase-seq|Fibropag20443, Chromatin_Structure||DNase-seq|Medullo, Chromatin_Structure||DNase-seq|Chorion, Chromatin_Structure||DNase-
B123 B124	chr9	77915529	G	A	transition	intergenic		CpG_site	NA	5	Motifs|PWM|FAC1, Motifs|PWM|CLOCK, Chromatin_Structure||DNase-seq|M059j, Chromatin_Structure||DNase-seq|Hah, Chromatin_Structure||DNase-seq|Th17, Chromatin_Structure||DNase-seq|Th2, Chromatin_Structure||DNase-seq|Th2wb54553204, Chromatin_Structure||DNase-seq|Th1
B123 B124	chr5	79459636	T	C	transition	SERINC5	intronic	NA	NA	5	Chromatin_Structure||DNase-seq|Hnpce, Chromatin_Structure||DNase-seq|Lhcnm2

B62 B63	chr16	81489957	A	C	transversion	CMIP	intronic	NA	NA	5	Motifs|Footprinting|Gm12891|ARP-1(COUP-TF2), Motifs|Footprinting|Hsmm|ARP-1(COUP-TF2), Motifs|Footprinting|Hmec|ARP-1(COUP-TF2), Motifs|Footprinting|Gm12892|ARP-1(COUP-TF2), Motifs|PWM||RARA, Motifs|Footprinting|Medullo|ARP-1(COUP-TF2), Motifs|Footprinting|Gm19238|ARP-1(COUP-TF2), Motifs|PWM||THRA, Motifs|Footprinting|8988t|ARP-1(COUP-TF2), Motifs|Footprinting|Gm12892|GCNF, Motifs|Footprinting|Lncap|ARP-1(COUP-TF2), Motifs|PWM||ARP-1(COUP-TF2), Motifs|Footprinting|Chorion|ARP-1(COUP-TF2), Motifs|Footprinting|Gm19239|ARP-1(COUP-TF2), Motifs|Footprinting|Osteobl|ARP-1(COUP-TF2), Motifs|Footprinting|A549|GCNF, Motifs|Footprinting|H1hesc|ARP-1(COUP-TF2), Motifs|Footprinting|Huh75|ARP-1(COUP-TF2), Motifs|PWM||TTF-1(Nkx2-1), Motifs|Footprinting|8988t|GCNF, Motifs|Footprinting|8988t|TTF-1(Nkx2-1), Motifs|Footprinting|Gm12892|TTF-1(Nkx2-1), Motifs|PWM||GCNF, Motifs|Footprinting|Hepg2|ARP-1(COUP-TF2), Chromatin_Structure||DNase-seq|Hffmyc, Chromatin_Structure||DNase-seq|H7es, Chromatin_Structure|Znfa41c6|DNase-seq|K562, Chromatin_Structure|Znff41b2|DNase-seq|K562, Chromatin_Structure||DNase-seq|Hff, Chromatin_Structure||DNase-seq|K562, Chromatin_Structure|Znfp5|DNase-seq|K562, Chromatin_Structure||DNase-seq|Osteobl, Chromatin_Structure||DNase-seq|Hepg2, Chromatin_Structure||DNase-
B83 B84	chr9	82567879	T	C	transition	intergenic		NA	NA	5	Chromatin_Structure||DNase-seq|Panisd, Chromatin_Structure||FAIRE|Gliobla, Chromatin_Structure||DNase-seq|Colo829
B93 B94	chr11	83191270	C	A	transversion	DLG2	intronic	NA	NA	5	Motifs|PWM||Gfi1, Chromatin_Structure|Nabut|FAIRE|K562, Chromatin_Structure||DNase-seq|Hah
LJ LS	chr11	84078617	G	T	transversion	DLG2	intronic	NA	NA	5	Motifs|PWM||ATF4, Motifs|PWM||Duxl, Motifs|PWM||Cphx, Motifs|PWM||DUXA, Protein_Binding||ChIP-seq|VCaP|ERG
B103 B104	chr14	85591083	T	C	transition	intergenic		NA	NA	5	Motifs|PWM||LHX8, Motifs|PWM||LHX6, Chromatin_Structure||DNase-seq|H7es, Chromatin_Structure|Diffa5d|DNase-seq|H7es
B62 B63	chr15	90929885	T	C	transition	IQGAP1	5upstream	NA	NA	5	Motifs|PWM||Nkx3-1, Motifs|Footprinting|Hepatocytes|Nkx2-9, Motifs|Footprinting|A549|Nkx2-9, Motifs|PWM||Nkx2-9, Chromatin_Structure||DNase-seq|Hepatocytes, Chromatin_Structure||DNase-seq|Osteobl
B93 B94	chr10	94297199	C	A	transversion	IDE	syn	NA	NA	5	Chromatin_Structure||FAIRE|Gm19239, Chromatin_Structure||DNase-seq|Hae, Chromatin_Structure|Hypoxlac|FAIRE|Mcf7

B93 B94	chr7	97860074	G	A	transition	TECPR1	intronic	NA	NA	5	Chromatin_Structure||DNase-seq|Cd20ro01794, Chromatin_Structure||DNase-seq|Ipsnihi11, Chromatin_Structure||DNase-seq|Sknsh, Chromatin_Structure||DNase-seq|Lncap, Chromatin_Structure||DNase-seq|Cerebrumfrontaloc, Chromatin_Structure||DNase-seq|Gm12891, Chromatin_Structure||DNase-seq|Ipscwru1, Chromatin_Structure||DNase-seq|Medullod341, Chromatin_Structure||DNase-seq|Gm20000, Chromatin_Structure||DNase-seq|H7es, Chromatin_Structure||DNase-seq|Hsmm, Chromatin_Structure||DNase-seq|Monocd14, Chromatin_Structure||DNase-seq|Panislets, Chromatin_Structure||DNase-seq|Gm13976, Chromatin_Structure||DNase-seq|H1hesc, Chromatin_Structure||DNase-seq|K562, Chromatin_Structure||DNase-seq|Adultcd4th1, Chromatin_Structure|Andro|DNase-seq|Lncap, Chromatin_Structure||DNase-seq|Ips, Chromatin_Structure||DNase-seq|Gm10266, Chromatin_Structure||DNase-seq|Frontalcortexoc, Chromatin_Structure||DNase-seq|Huh7, Chromatin_Structure|Nabut|DNase-seq|K562, Chromatin_Structure||DNase-seq|Hsmmt, Chromatin_Structure||DNase-seq|Gm19238, Chromatin_Structure||DNase-seq|Osteobl, Chromatin_Structure||DNase-seq|Gm13977, Chromatin_Structure||DNase-seq|Imr90, Chromatin_Structure||DNase-seq|Hepg2, Chromatin_Structure||DNase-seq|Hsmmfshd, Chromatin_Structure||DNase-seq|Medullo,
B113 B114	chr3	98242563	T	C	transition	CLDND1	5upstream	NA	NA	5	Chromatin_Structure||DNase-seq|Th1wb33676984, Chromatin_Structure||DNase-seq|K562, Chromatin_Structure||DNase-seq|Hepatocytes, Chromatin_Structure||DNase-seq|Gm12865, Chromatin_Structure|Znfp5|DNase-seq|K562, Chromatin_Structure||FAIRE|Gm12892, Chromatin_Structure||DNase-seq|Th1
LJ LS	chr11	100687399	G	A	transition	ARHGAP42	intronic	NA	NA	5	Motifs|PWM||Nkx6-1, Motifs|PWM||GSX2, Motifs|PWM||RAX, Motifs|PWM||HMX2, Motifs|PWM||HMX1, Motifs|PWM||HMX3, Motifs|PWM||REX, Chromatin_Structure||DNase-seq|Hmec
B103 B104	chr13	101552351	C	A	transversion	NALCN_AS1	non-coding intronic	NA	NA	5	Chromatin_Structure||DNase-seq|Th17, Chromatin_Structure||DNase-seq|Th2, Chromatin_Structure||DNase-seq|Th2wb54553204, Chromatin_Structure||DNase-seq|Gm12865, Chromatin_Structure||DNase-seq|Th1
B103 B104	chr14	104320775	G	A	transition	LINC00637	non-coding intronic	CpG_site	NA	5	Chromatin_Structure||DNase-seq|Chorion
B103 B104	chr12	104900233	A	G	transition	CHST11	intronic	NA	NA	5	Protein_Binding||ChIP-seq|Jurkat|CREBBP
LJ LS	chr12	105193487	A	G	transition	intergenic		NA	NA	5	Chromatin_Structure|Ohurea|FAIRE|K562, Chromatin_Structure||DNase-seq|H9es
B93 B94	chr3	113657145	G	A	transition	GRAMD1C	intronic	NA	NA	5	Protein_Binding||ChIP-seq|HepG2|MAFK
B83 B84	chr5	116550935	G	A	transition	intergenic		CpG_site	NA	5	Chromatin_Structure||DNase-seq|Th17, Chromatin_Structure||DNase-seq|Th1
B113 B114	chr10	117458502	T	A	transversion	ATRNL1	intronic	NA	NA	5	Motifs|PWM||Srf, Protein_Binding||ChIP-seq|HepG2|MAFK
B103 B104	chr3	118923152	T	C	transition	UPK1B	3utr	NA	NA	5	Chromatin_Structure||DNase-seq|K562
B103 B104	chr8	121565361	A	G	transition	SNTB1	intronic	NA	NA	5	Chromatin_Structure||DNase-seq|Ipsnihi7, Chromatin_Structure||DNase-seq|Hcpe, Chromatin_Structure||FAIRE|Htr8, Chromatin_Structure||DNase-seq|Nt2d1, Chromatin_Structure||DNase-seq|H7es, Chromatin_Structure||DNase-seq|H1hesc, Chromatin_Structure||DNase-seq|Hbvp, Chromatin_Structure||FAIRE|H1hesc, Chromatin_Structure|Diffa5d|DNase-seq|H7es, Chromatin_Structure||FAIRE|Urotsa, Chromatin_Structure||DNase-seq|Ips,
B113 B114	chr10	123074966	T	C	transition	intergenic		NA	NA	5	Chromatin_Structure||DNase-seq|Fibrop

B113 B114	chr9	124373251	T	C	transition	DAB2IP	intronic	NA	NA	5	Motifs|PWM||HFH8(FOXF1A), Motifs|PWM||HNF3, Motifs|PWM||HFH4(FOXJ1), Motifs|Footprinting|Hmec|FOXJ2, Motifs|PWM||FOXC1, Motifs|PWM||FOXP1, Motifs|PWM||FOXL1, Motifs|PWM||FOXD3, Motifs|PWM||FOXI1, Motifs|PWM||Foxd3, Motifs|Footprinting|LncapAndro|FOXJ2, Motifs|Footprinting|LncapAndro|HNF3, Motifs|Footprinting|H9es|FOXJ2, Motifs|Footprinting|Medullo|HFH4(FOXJ1), Motifs|PWM||FOXC2, Motifs|Footprinting|LncapAndro|HFH8(FOXF1A), Motifs|PWM||FOXJ2, Motifs|Footprinting|Medullo|FOXJ2, Motifs|Footprinting|LncapAndro|HFH4(FOXJ1),
LJ LS	chr6	124510527	C	T	transition	NKAIN2	intronic	NA	NA	5	Chromatin_Structure||DNase-seq|Lhcnm2
LJ LS	chr6	124515065	A	T	transversion	NKAIN2	intronic	NA	NA	5	Motifs|PWM||SREBP, Motifs|PWM||Osr2, Motifs|PWM||SREBP1, Chromatin_Structure||DNase-seq|H7es
B103 B104	chr11	126855973	C	A	transversion	KIRREL3	intronic	NA	NA	5	Chromatin_Structure|Est10nm30m|DNase-seq|T47d, Chromatin_Structure||DNase-seq|T47d
B113 B114	chr5	127441266	A	G	transition	SLC12A2	intronic	NA	NA	5	Chromatin_Structure||FAIRE|Gm19239, Chromatin_Structure||DNase-seq|Nhlf, Chromatin_Structure||DNase-seq|Aoaf
B62 B63	chr7	127503274	G	A	transition	SND1	intronic	NA	NA	5	Chromatin_Structure||DNase-seq|Hcpe, Chromatin_Structure||DNase-seq|Hasp, Chromatin_Structure||DNase-seq|Hbvp, Chromatin_Structure||DNase-seq|Hbvsmc
B123 B124	chr4	128289126	C	T	transition	intergenic		CpG_site	NA	5	Motifs|PWM||HOXA10, Motifs|PWM||Hoxb9, Motifs|PWM||HOXC11, Motifs|PWM||Hoxa9, Motifs|PWM||Hoxa10, Motifs|PWM||hoxa9, Motifs|PWM||HOXA11, Chromatin_Structure||DNase-seq|Osteobl
B123 B124	chr6	138054222	T	C	transition	intergenic		NA	NA	5	Motifs|PWM||Sox4, Protein_Binding||ChIP-seq|HepG2|POLR2A
B93 B94	chr6	139472949	A	G	transition	HECA	intronic	NA	NA	5	Chromatin_Structure||DNase-seq|Hah, Chromatin_Structure||DNase-seq|Hasp, Chromatin_Structure||FAIRE|Gm19239
B83 B84	chr9	139573616	C	T	transition	AGPAT2	intronic	NA	NA	5	Motifs|Footprinting|Chorion|TFII-I, Motifs|Footprinting|H1hesc|TFII-I, Motifs|Footprinting|LncapAndro|TFII-I, Motifs|Footprinting|Lncap|TFII-I, Motifs|Footprinting|Gm12892|TFII-I, Motifs|PWM||TFII-I, Chromatin_Structure||DNase-seq|Cd20ro01794, Chromatin_Structure||DNase-seq|Heartoc, Chromatin_Structure|Ifng4h|FAIRE|Helas3, Chromatin_Structure||DNase-seq|Sknsh, Chromatin_Structure||DNase-seq|Lncap, Chromatin_Structure||DNase-seq|Cerebrumfrontaloc, Chromatin_Structure||DNase-seq|Gm12891, Chromatin_Structure||DNase-seq|8988t, Chromatin_Structure||DNase-seq|Gm20000, Chromatin_Structure||DNase-seq|H7es, Chromatin_Structure||DNase-seq|Monocd14, Chromatin_Structure|Ifna4h|FAIRE|Helas3, Chromatin_Structure|Andro|DNase-seq|Lncap, Chromatin_Structure||DNase-seq|Ips, Chromatin_Structure||DNase-seq|Hepatocytes, Chromatin_Structure||DNase-seq|Gm10266, Chromatin_Structure||DNase-seq|Frontalcortexoc, Chromatin_Structure||DNase-seq|T47d, Chromatin_Structure||DNase-seq|Huh7, Chromatin_Structure||DNase-seq|Helas3, Chromatin_Structure||DNase-seq|Gm19238, Chromatin_Structure|Ifna4h|DNase-seq|Helas3, Chromatin_Structure||DNase-seq|Gm19239, Chromatin_Structure||DNase-seq|Huh75, Chromatin_Structure||DNase-seq|Hmec, Chromatin_Structure||DNase-seq|Osteobl, Chromatin_Structure||DNase-seq|Gm13977, Chromatin_Structure||DNase-

B123 B124	chr9	139787912	G	T	transversion	TRAF2	intronic	NA	NA	5	Chromatin_Structure|Ifng4h|FAIRE|Helas3, Chromatin_Structure||DNase-seq|Lncap, Chromatin_Structure||DNase-seq|Helas3, Chromatin_Structure|Ifna4h|DNase-seq|Helas3, Chromatin_Structure||DNase-seq|Th1
B123 B124	chr6	150229677	G	T	transversion	RAET1E_AS1	non-coding intronic	NA	NA	5	Protein_Binding||ChIP-seq|K562|SETDB1, Protein_Binding||ChIP-seq|K562|TRIM28, Protein_Binding||ChIP-seq|HEK293|TRIM28, Protein_Binding||ChIP-seq|K562|CBX3
B93 B94	chr1	151829054	G	C	transversion	intergenic		NA	NA	5	Chromatin_Structure|Est10nm30m|DNase-seq|T47d, Chromatin_Structure||DNase-seq|Medullod341, Chromatin_Structure||DNase-seq|Medullo, Chromatin_Structure|DNase-seq|T47d, Chromatin_Structure|DNase-seq|Werirb1
B114	chr6	154215825	T	C	transition	intergenic		NA	NA	5	Motifs|Footprinting|Cll|MEF2A, Motifs|Footprinting|Cll|MEF-2, Motifs|PWM||MEF2A, Motifs|PWM||MEF-2, Motifs|Footprinting|Helas3Ifna4h|MEF-2, Motifs|Footprinting|Helas3Ifna4h|MEF2A, Chromatin_Structure||FAIRE|Panislets, Chromatin_Structure|Znf4c50c4|DNase-seq|K562, Chromatin_Structure||FAIRE|Gm12878, Chromatin_Structure|Nabut|FAIRE|K562, Chromatin_Structure||FAIRE|H1hesc, Chromatin_Structure||DNase-seq|Fibrobl, Chromatin_Structure||DNase-seq|Th1wb33676984, Chromatin_Structure||DNase-seq|K562, Chromatin_Structure||DNase-seq|Helas3, Chromatin_Structure|Ifna4h|DNase-seq|Helas3, Chromatin_Structure||FAIRE|K562, Chromatin_Structure|Hypoxlac|FAIRE|Mcf7,
LJ LS	chr2	167170722	C	T	transition	SCN9A	intronic	CpG_site	NA	5	Motifs|PWM||MAFK, Motifs|PWM||Pax-4, Chromatin_Structure||DNase-seq|Gm04503, Chromatin_Structure||DNase-seq|Hah, Chromatin_Structure||DNase-seq|Nhdfneo, Chromatin_Structure||DNase-seq|Hipe, Chromatin_Structure||DNase-seq|Hvmf, Chromatin_Structure||DNase-seq|Hff, Chromatin_Structure||DNase-seq|Hffmyc, Chromatin_Structure||DNase-seq|Ag04449, Chromatin_Structure||FAIRE|Urotsa, Chromatin_Structure||FAIRE|UrotsaUt189, Chromatin_Structure|Serumfree|DNase-seq|Aosmc, Chromatin_Structure||DNase-seq|Ag10803, Chromatin_Structure||DNase-seq|Ag09309,
B113 B114	chr3	171864901	G	C	transversion	FNDC3B	intronic	NA	NA	5	Chromatin_Structure||DNase-seq|K562, Chromatin_Structure|Znfp5|DNase-seq|K562
B123 B124	chr2	172411647	G	C	transversion	CYBRD1	3utr	NA	NA	5	Motifs|PWM||Prrx1, Chromatin_Structure||DNase-seq|Hcm, Chromatin_Structure||DNase-seq|Hmvecdneo, Chromatin_Structure||DNase-seq|Hmveclly, Chromatin_Structure||DNase-seq|Hcf, Chromatin_Structure||DNase-seq|Ag10803, Chromatin_Structure||DNase-seq|Hmf, Chromatin_Structure||DNase-seq|Nhdfad, Chromatin_Structure||DNase-seq|Hpaf, Chromatin_Structure||DNase-seq|Aoaf, Chromatin_Structure|FAIRE|Gliobla
B53 B54	chr5	177563147	C	T	transition	RMND5B	intronic	CpG_site	NA	5	Motifs|Footprinting|Mcf7|Dax1, Motifs|PWM||COUPTF, Motifs|PWM||Dax1, Chromatin_Structure|Est100nm1h|DNase-seq|Mcf7, Chromatin_Structure|Estctrl0h|DNase-seq|Mcf7, Chromatin_Structure||DNase-seq|Mcf7
B123 B124	chr3	181670965	C	T	transition	LINC01206	non-coding intronic	NA	NA	5	Motifs|PWM||Zfp740, Chromatin_Structure||DNase-seq|Nt2d1, Chromatin_Structure||DNase-seq|Hah, Chromatin_Structure||DNase-seq|H7es
LJ LS	chr2	184856690	T	C	transition	intergenic		NA	NA	5	Motifs|PWM||MyoD, Motifs|Footprinting|A549|MyoD, Motifs|PWM||RARG, Protein_Binding||ChIP-seq|HeLa-S3|GTF2B, Protein_Binding||ChIP-seq|HeLa-S3|POLR3A
B62 B63	chr2	188360514	A	G	transition	TFPI	intronic	NA	NA	5	Motifs|PWM||Gata5, Chromatin_Structure||DNase-seq|Hasp

LJ LS	chr2	218660590	C	G	transversion	intergenic		NA	NA	5	Motifs|PWM||Sp1, Chromatin_Structure|Znfa41c6|DNase-seq|K562, Chromatin_Structure||DNase-seq|H1hesc, Chromatin_Structure||DNase-seq|Progfib, Chromatin_Structure|Diffa14d|DNase-seq|H7es, Chromatin_Structure||DNase-seq|Hah, Chromatin_Structure||DNase-seq|Fibrobl, Chromatin_Structure||DNase-seq|Ipsnihi7, Chromatin_Structure||DNase-seq|Hipe, Chromatin_Structure|Est10nm30m|DNase-seq|Ecc1, Chromatin_Structure|Diffa5d|DNase-seq|H7es, Chromatin_Structure||DNase-seq|Hff, Chromatin_Structure||DNase-seq|Hcm, Chromatin_Structure||DNase-seq|K562, Chromatin_Structure|Diffa9d|DNase-seq|H7es, Chromatin_Structure|Tam10030|DNase-seq|Ishikawa, Chromatin_Structure||DNase-seq|Fibropag08395, Chromatin_Structure||DNase-seq|Stellate, Chromatin_Structure|Diff4d|DNase-seq|Lhcnm2, Chromatin_Structure||DNase-seq|Imr90, Chromatin_Structure|Lentimyod|DNase-seq|Fibroblgm03348, Chromatin_Structure||DNase-seq|Hepg2, Chromatin_Structure||DNase-seq|Ipscwru1, Chromatin_Structure||DNase-seq|Lhcnm2, Chromatin_Structure||DNase-seq|Fibropag20443, Chromatin_Structure||DNase-seq|Hnpce, Chromatin_Structure||DNase-seq|Hsmmemb, Chromatin_Structure||DNase-seq|Nhlf, Chromatin_Structure|Est10nm30m|DNase-seq|Ishikawa, Chromatin_Structure||DNase-seq|Ipsnihi11, Chromatin_Structure||DNase-seq|Ips, Chromatin_Structure||DNase-seq|Hcf, Chromatin_Structure||DNase-seq|Fibrop, Chromatin_Structure||DNase-seq|Panisd, Chromatin_Structure||DNase-seq|Cmk, Chromatin_Structure||DNase-seq|Fibroblgm03348, Chromatin_Structure|Dm002p1h|DNase-seq|Ecc1, Chromatin_Structure||DNase-seq|Ag10803, Chromatin_Structure||DNase-seq|H7es, Chromatin_Structure||DNase-seq|Fibropag08396, Chromatin_Structure||DNase-seq|Hvmf
B53 B54	chr2	230096243	G	A	transition	PID1	intronic	NA	NA	5	Motifs|PWM||TCF11:MafG, Protein_Binding||ChIP-seq|PBDE|GATA1
B103 B104	chr2	231640380	T	G	transversion	CAB39	intronic	NA	NA	5	Chromatin_Structure||DNase-seq|Hgf
B53 B54	chr2	241555733	C	A	transversion	GPR35	intronic	CpG_site	NA	5	Motifs|PWM||IRF
B93 B94	chr18	508117	A	C	transversion	intergenic		NA	NA	6	Motifs|PWM||HFH8(FOXF1A), Motifs|Footprinting|A549|HFH8(FOXF1A), Motifs|PWM||HNF3beta, Motifs|PWM||HNF3, Motifs|PWM||HFH4(FOXJ1), Motifs|Footprinting|LncapAndro|Freac-7, Motifs|PWM||FOXC1, Motifs|Footprinting|A549|HFH3(FOXI1), Motifs|Footprinting|LncapAndro|FOXO3A, Motifs|PWM||Dbx1, Motifs|PWM||Freac-7, Motifs|PWM||Cdx, Motifs|Footprinting|Gm12878|FOXO3A, Motifs|Footprinting|A549|Cdx, Motifs|Footprinting|A549|Foxd3, Motifs|PWM||Foxl1, Motifs|Footprinting|Helas3Ifna4h|Cdx, Motifs|PWM||FOXP1, Motifs|Footprinting|Cll|Freac-7, Motifs|Footprinting|A549|Freac-7, Motifs|Footprinting|A549|HFH4(FOXJ1), Motifs|PWM||FOXL1, Motifs|PWM||FOXD3, Motifs|PWM||Foxd3, Motifs|PWM||POU3F1, Motifs|Footprinting|A549|HNF3, Motifs|PWM||Foxj3, Motifs|PWM||ONECUT3, Motifs|PWM||Tlx2, Motifs|PWM||FOXC2, Motifs|PWM||FOXO3A, Motifs|Footprinting|A549|FOXO3A, Motifs|Footprinting|LncapAndro|Cdx, Motifs|Footprinting|Cll|Cdx, Motifs|Footprinting|Gm12878|Cdx, Motifs|PWM||FOXJ2, Motifs|Footprinting|Cll|FOXO3A, Motifs|Footprinting|A549|HNF3beta, Motifs|PWM||Foxa2
B53 B54	chr12	634274	T	G	transversion	B4GALNT3	intronic	NA	NA	6	

B53 B54	chr7	2384889	C	G	transversion	intergenic		NA	NA	6	Motifs|PWM||AR, Motifs|PWM||Ar, Motifs|PWM||GR
B83 B84	chr19	4421191	C	T	transition	CHAF1A	intronic	NA	NA	6	Motifs|PWM||LUN-1, Chromatin_Structure||FAIRE|Helas3
B113 B114	chr18	6348505	T	C	transition	L3MBTL4	intronic	NA	NA	6	Motifs|PWM||ESRRA, Motifs|PWM||Hbp1
B93 B94	chr18	10325164	C	T	transition	intergenic		NA	NA	6	Motifs|PWM||Oct-1
B103 B104	chr9	12145048	G	A	transition	intergenic		NA	NA	6	Motifs|PWM||Six6, Motifs|PWM||Sox7, Motifs|PWM||DMRT3, Motifs|PWM||Six3
B62 B63	chr20	13918102	A	G	transition	SEL1L2	intronic	NA	NA	6	Motifs|PWM||FOXG1
LJ LS	chr9	15398550	G	C	transversion	intergenic		NA	NA	6	Motifs|PWM||Pitx2, Motifs|Footprinting|Cll|Pitx2
B83 B84	chr21	16181595	A	G	transition	intergenic		NA	NA	6	Motifs|PWM||Gfi1b
B83	chr8	16483051	T	C	transition	intergenic		NA	NA	6	Motifs|PWM||NR2E1
B93 B94	chr2	16519005	G	C	transversion	intergenic		NA	NA	6	Motifs|PWM||Cart-1
B62 B63	chr3	16887859	C	T	transition	intergenic		NA	NA	6	Motifs|PWM||Hand1:E47, Motifs|PWM||SMAD4
B93 B94	chr7	19081637	C	A	transversion	intergenic		NA	NA	6	Motifs|PWM||HeliosA
B62 B63	chr19	20630678	T	C	transition	intergenic		NA	NA	6	Motifs|PWM||CIZ
B62 B63	chr21	21885587	G	A	transition	intergenic		NA	NA	6	Motifs|PWM||ATF4
B62 B63	chr10	24095479	T	C	transition	KIAA1217	intronic	NA	NA	6	Motifs|PWM||POU4F3, Motifs|PWM||POU4F2, Motifs|PWM||POU4F1
B53 B54	chr11	30052740	G	C	transversion	intergenic		NA	NA	6	Motifs|PWM||Meis1, Motifs|PWM||MAFB, Motifs|PWM||TGIF
B53 B54	chr11	31215844	C	G	transversion	intergenic		NA	NA	6	Motifs|PWM||PTF1-beta, Motifs|PWM||RARA, Motifs|PWM||RARB
B123 B124	chr19	31507732	T	G	transversion	intergenic		NA	NA	6	Motifs|PWM||HoxB5, Motifs|PWM||Hoxc8, Motifs|PWM||Pax-4, Motifs|PWM||HOXC-8, Motifs|PWM||Dlx5, Motifs|PWM||Dlx3, Motifs|PWM||Hoxa6, Motifs|PWM||Dlx2, Motifs|PWM||Dlx4, Motifs|PWM||Hoxb5, Motifs|PWM||Hlxb9
B113 B114	chr5	31616208	A	G	transition	intergenic		NA	NA	6	Motifs|PWM||Tcf3
B62 B63	chr10	31767758	G	A	transition	ZEB1	intronic	NA	NA	6	Motifs|PWM||TBX15, Motifs|PWM||FOXB1
B93 B94	chr2	32706159	A	G	transition	BIRC6	intronic	NA	NA	6	Motifs|PWM||POU3F3, Motifs|PWM||Mafb, Motifs|PWM||POU3F2, Motifs|PWM||POU3F1
B123 B124	chr5	34827544	A	C	transversion	RAI14	intronic	NA	NA	6	Motifs|PWM||HIC1, Motifs|PWM||RFX4
B62 B63	chr11	34949538	A	G	transition	PDHX	intronic	NA	NA	6	Motifs|PWM||POU3F3, Motifs|PWM||Oct-1, Motifs|PWM||POU3F1, Motifs|PWM||POU2F1, Motifs|PWM||POU2F2
B62	chr11	35864072	A	T	transversion	intergenic		NA	NA	6	Motifs|PWM||SPIB, Motifs|PWM||Srf, Motifs|PWM||SPIC, Motifs|PWM||SPI1, Motifs|PWM||SPI-B
B53 B54	chr14	37701455	T	C	transition	MIPOL1	intronic	NA	NA	6	Motifs|PWM||HNF1A
LJ LS	chr14	39073589	G	C	transversion	intergenic		NA	NA	6	Motifs|PWM||Mtf1, Motifs|PWM||Oct-4(POU5F1), Motifs|PWM||DMRT5
B53 B54	chr1	40982533	A	G	transition	EXO5	3downstream	NA	NA	6	Motifs|PWM||AIRE
B123 B124	chr2	41332857	C	T	transition	intergenic		NA	NA	6	Motifs|PWM||COMP1
B62 B63	chr12	41970530	A	G	transition	intergenic		NA	NA	6	Motifs|PWM||IRX5, Motifs|PWM||IRX2, Motifs|PWM||IRX3
B103 B104	chr12	42352274	A	C	transversion	intergenic		NA	NA	6	Motifs|PWM||Gfi-1
B123	chr14	43121265	G	A	transition	intergenic		CpG_site	NA	6	Motifs|PWM||FOXC1, Motifs|PWM||Freac-3, Motifs|PWM||Sox7
B53 B54	chr1	43588564	C	T	transition	intergenic		NA	NA	6	Motifs|PWM||FOXP1, Motifs|PWM||Six6
B84	chr13	45830708	C	T	transition	GTF2F2	intronic	NA	NA	6	Motifs|PWM||Oct-1
B93 B94	chr3	46151596	A	G	transition	intergenic		NA	NA	6	Motifs|PWM||Pax-4, Chromatin_Structure||FAIRE|Gm19239
B83	chr8	50040053	G	A	transition	intergenic		NA	NA	6	Motifs|PWM||HeliosA
B113	chr20	52177664	A	G	transition	RP4_724E16.2	non-coding intronic	NA	NA	6	Motifs|Footprinting|A549|EWSR1-FLI1, Motifs|PWM||EWSR1-FLI1
B83 B84	chr3	53325297	C	T	transition	DCP1A	intronic	NA	NA	6	Motifs|PWM||POU3F3
B83 B84	chr5	53365411	A	C	transversion	ARL15	intronic	NA	NA	6	Motifs|PWM||Srf, Motifs|PWM||FOXP1, Motifs|PWM||Zfp105, Motifs|PWM||Mtf1, Motifs|PWM||AIRE
B113 B114	chr19	53997821	C	T	transition	ZNF813	3downstream	CpG_site	NA	6	Motifs|PWM||ZNF524
LJ LS	chr7	54008241	C	T	transition	intergenic		CpG_site	NA	6	Motifs|PWM||Obox1
B62 B63	chr14	54397605	T	C	transition	intergenic		NA	NA	6	Motifs|PWM||Dbx1, Motifs|PWM||C/EBPgamma, Motifs|PWM||SOX5
B93 B94	chr17	54433363	A	G	transition	ANKFN1	intronic	NA	NA	6	Motifs|PWM||Oct-1
B93 B94	chr17	54433372	T	C	transition	ANKFN1	intronic	NA	NA	6	Motifs|PWM||Oct-1

B123 B124	chr4	55065295	T	A	transversion	intergenic		NA	NA	6	Motifs|PWM||Barhl2, Motifs|PWM||Nkx5-2
B93 B94	chrX	55365659	C	A	transversion	intergenic		NA	NA	6	Motifs|PWM||HNF3alpha, Chromatin_Structure||FAIRE|Astrocy
B93 B94	chrX	55595693	C	A	transversion	intergenic		NA	NA	6	Motifs|PWM||MEF-2, Chromatin_Structure||FAIRE|A549
B103 B104	chr15	56398367	T	C	transition	RFX7	intronic	NA	NA	6	Motifs|PWM||Bcl6b, Chromatin_Structure||FAIRE|A549
B123 B124	chr6	56978734	T	G	transversion	ZNF451	intronic	NA	NA	6	Motifs|PWM||Tcf7, Motifs|PWM||Tbp
B53 B54	chr3	57476826	A	C	transversion	DNAH12	intronic	NA	NA	6	Motifs|PWM||FAC1, Motifs|PWM||Foxk1, Motifs|PWM||FOXC1, Motifs|PWM||Mtf1, Motifs|PWM||FOXO3, Motifs|PWM||Srf, Motifs|PWM||BCL6, Motifs|PWM||FOXP1, Motifs|PWM||Zfp105, Motifs|PWM||FOXO1
B53 B54	chr8	57645596	T	A	transversion	intergenic		NA	NA	6	Motifs|PWM||MEF-2
B93 B94	chr17	58787185	G	A	transition	BCAS3	intronic	NA	NA	6	Motifs|Footprinting|Mcf7|SRF, Motifs|Footprinting|Mcf7Hypoxlac|SRF, Motifs|PWM||SRF
B123 B124	chr17	60527119	A	G	transition	METTL2A	3utr	NA	NA	6	Motifs|Footprinting|GM12892|, Motifs|Footprinting|HelaS3|
B103 B104	chr16	63843879	C	T	transition	intergenic		NA	NA	6	Motifs|PWM||NR3C1
B93 B94	chr14	64047343	A	G	transition	intergenic		NA	NA	6	Motifs|PWM||Dbx2, Motifs|PWM||Vsx1, Motifs|PWM||Rhox6, Motifs|PWM||Pou4f3, Motifs|PWM||Hoxd8, Motifs|PWM||Sox1, Motifs|PWM||Isl2, Motifs|PWM||Pou3f1, Chromatin_Structure|FAIRE|Htr8
B113 B114	chr13	65852160	C	A	transversion	intergenic		NA	NA	6	Motifs|PWM||HOXA10, Motifs|PWM||HOXC11, Motifs|PWM||Hoxa9, Motifs|PWM||Cdx2, Motifs|PWM||Hoxd10, Motifs|PWM||Cdx1, Motifs|PWM||Cdx-2, Motifs|PWM||hoxa9, Motifs|PWM||HOXA11, Motifs|PWM||Hoxb9, Motifs|PWM||Sox7, Motifs|PWM||HOXD11
B103 B104	chr8	66834673	A	G	transition	intergenic		NA	NA	6	Motifs|PWM||AREB6, Motifs|PWM||Pitx2, Motifs|PWM||TCF4, Motifs|PWM||MEIS2
B114	chr9	66861389	T	C	transition	intergenic		NA	NA	6	Motifs|Footprinting|Helas3Ifna4h|HSF2, Motifs|Footprinting|Gm12878|HSF2, Motifs|Footprinting|Hpde6e6e7|HSF2, Motifs|PWM||HSF1, Motifs|PWM||HSF2
B114	chr12	67837996	G	A	transition	intergenic		CpG_site	NA	6	Motifs|PWM||RARA
B53 B54	chr2	68543768	A	T	transversion	CNRIP1	intronic	NA	NA	6	Motifs|PWM||Six6, Motifs|PWM||Tbp, Motifs|PWM||Dobox4
B93 B94	chr3	70268930	A	C	transversion	intergenic		NA	NA	6	Motifs|PWM||NR1H2::RXRA
B93 B94	chr15	70747621	C	G	transversion	intergenic		NA	NA	6	Motifs|PWM||Pitx2, Motifs|Footprinting|Mcf7|Pitx2
B93 B94	chr18	73917867	G	A	transition	intergenic		NA	NA	6	Motifs|PWM||CREB3L1, Motifs|PWM||CREB3L2, Chromatin_Structure|FAIRE|Medullo
B83 B84	chr15	75719027	T	C	transition	SIN3A	intronic	NA	NA	6	Motifs|Footprinting|AosmcSerumfree|Ikaros, Motifs|Footprinting|Cll|Lyf-1, Motifs|PWM||Ikaros, Motifs|Footprinting|Helas3|Ikaros, Motifs|Footprinting|A549|Ikaros, Motifs|Footprinting|Cll|Ikaros, Motifs|Footprinting|A549|Lyf-1, Motifs|PWM||Lyf-1, Motifs|Footprinting|Gm12878|Lyf-1, Motifs|Footprinting|Gm12878|Ikaros, Motifs|Footprinting|Panisd|Ikaros, Motifs|Footprinting|Huh7|Ikaros, Motifs|Footprinting|Mcf7Hypoxlac|Lyf-1, Motifs|Footprinting|Helas3Ifna4h|Ikaros,
B62 B63	chr3	76252246	C	T	transition	intergenic		CpG_site	NA	6	Motifs|PWM||TCFAP2A, Motifs|PWM||TFAP2B, Motifs|PWM||Tcfap2e, Motifs|PWM||TFAP2C, Motifs|PWM||TFAP2A, Motifs|PWM||Tcfap2b
LJ LS	chr16	76707882	C	T	transition	intergenic		NA	NA	6	Motifs|PWM||Lmx1a, Motifs|PWM||Lhx1, Motifs|PWM||HMGIY, Motifs|PWM||Arid3a, Motifs|PWM||Lmx1b, Motifs|PWM||Lhx5, Motifs|PWM||Hoxd13
B53 B54	chr3	76877788	A	G	transition	intergenic		NA	NA	6	Motifs|PWM||Tlx2
B103 B104	chr15	78321335	T	G	transversion	TBC1D2B	intronic	NA	NA	6	Motifs|PWM||Sox18, Motifs|PWM||Sry, Motifs|PWM||EMX2, Motifs|PWM||Cdx, Motifs|PWM||Nkx2-5, Motifs|PWM||Hmx2, Motifs|PWM||Sox5, Motifs|PWM||EN1, Motifs|PWM||Sox21, Motifs|PWM||Hmx1, Motifs|PWM||Nkx5-2, Motifs|PWM||Sox15, Motifs|PWM||HMX1, Motifs|PWM||MSX1, Motifs|PWM||Hmx3, Motifs|PWM||Sox12, Motifs|PWM||EMX1
B103 B104	chr12	79083015	G	A	transition	intergenic		NA	NA	6	Motifs|PWM||AP-2alphaA

B93 B94	chr2	79194279	T	A	transversion	intergenic		NA	NA	6	Motifs|PWM||NF-AT, Chromatin_Structure||FAIRE|Gm12891
LJ LS	chr1	79265388	C	T	transition	intergenic		NA	NA	6	Motifs|PWM||FOXJ3, Motifs|PWM||Zfp187, Motifs|PWM||FOXJ2
B93 B94	chr13	79350867	A	G	transition	intergenic		NA	NA	6	Motifs|PWM||VDR
B113 B114	chr12	80731159	G	A	transition	OTOGL	intronic	CpG_site	NA	6	Motifs|PWM||MECP2
B62 B63	chr1	81565925	A	G	transition	intergenic		NA	NA	6	Motifs|PWM||BCL6
B53 B54	chr4	81742124	C	T	transition	C4orf22	intronic	NA	NA	6	Motifs|PWM||SPDEF
B93 B94	chr7	85163682	G	A	transition	intergenic		NA	NA	6	Motifs|PWM||HNF3, Motifs|PWM||MEF2A, Motifs|PWM||MEF-2, Motifs|PWM||FOXD3, Motifs|PWM||RSRFC4, Motifs|PWM||Freac-4, Motifs|PWM||Foxj3, Motifs|PWM||Hoxd8, Motifs|PWM||Foxa2
B62 B63	chr13	85569294	C	T	transition	intergenic		NA	NA	6	Motifs|PWM||AIRE, Chromatin_Structure||FAIRE|Panislets, Chromatin Structure||FAIRE|Gliobla
B62	chr13	87882896	A	C	transversion	intergenic		NA	NA	6	Motifs|PWM||Dbx1, Motifs|PWM||FOXJ3, Motifs|PWM||FOXP1, Motifs|PWM||MEF-2, Motifs|PWM||Tlx2, Motifs|PWM||FOXJ2, Motifs|PWM||Sox8
B103 B104	chr3	89209126	T	G	transversion	EPHA3	intronic	NA	NA	6	Motifs|PWM||Srf, Motifs|PWM||FOXP1
B113 B114	chr4	89304876	G	A	transition	HERC6	intronic	NA	NA	6	Motifs|PWM||Zbtb3
B83	chr8	94260080	G	T	transversion	intergenic		NA	NA	6	Motifs|PWM||FOXJ3, Motifs|PWM||FOXJ2
B84	chr9	94497890	T	C	transition	ROR2	intronic	NA	NA	6	Motifs|PWM||Srf, Motifs|PWM||FOXP1, Motifs|PWM||Zfp105, Motifs|PWM||Mtf1
B62 B63	chr15	95452112	T	A	transversion	intergenic		NA	NA	6	Motifs|PWM||Tcf3, Motifs|PWM||Srf, Motifs|PWM||Tcfap2e, Motifs|PWM||Tcf7, Motifs|PWM||Zfp105
B103 B104	chr3	95967254	T	G	transversion	intergenic		NA	NA	6	Motifs|PWM||AR, Motifs|PWM||Gfi1b
B113	chr15	96155017	C	A	transversion	intergenic		NA	NA	6	Motifs|PWM||Barhl-1, Motifs|PWM||Nkx5-2
B62 B63	chrX	96306108	C	T	transition	DIAPH2	intronic	NA	NA	6	Motifs|PWM||ZFP652, Motifs|PWM||Gfi-1
B123 B124	chr5	97928714	T	G	transversion	intergenic		NA	NA	6	Motifs|PWM||Oct-1, Motifs|PWM||HMGIY, Motifs|PWM||HSF1, Motifs|PWM||Sox7, Motifs|PWM||NFAT5, Motifs|PWM||Nanog, Motifs|PWM||HSF2
B113 B114	chr1	101012039	G	T	transversion	intergenic		CpG_site	NA	6	Motifs|PWM||RBP-Jkappa
B53 B54	chr11	101016266	T	C	transition	LOC10105452[illegible]	non-coding intronic	NA	NA	6	Motifs|PWM||Hoxa5, Motifs|PWM||Lhx4, Motifs|PWM||Evx1, Motifs|PWM||Meox1, Motifs|PWM||Hoxa6, Motifs|PWM||Hoxa2, Motifs|PWM||Hoxd3, Motifs|PWM||Dlx2, Motifs|PWM||Hoxb3, Motifs|PWM||Csh2, Motifs|PWM||Inf1, Motifs|PWM||HOXD3
B62 B63	chr4	102822941	A	T	transversion	BANK1	intronic	NA	NA	6	Motifs|PWM||DMRT5
B113 B114	chr7	108858722	C	A	transversion	intergenic		NA	NA	6	Motifs|PWM||FOXC1, Motifs|PWM||SPIC
B53 B54	chr7	113264399	A	G	transition	intergenic		NA	NA	6	Motifs|PWM||Srf, Motifs|PWM||FOXP1, Motifs|PWM||Zfp105, Motifs|PWM||Mtf1
LJ LS	chr4	115909880	T	A	transversion	NDST4	intronic	NA	NA	6	Motifs|PWM||NF-E2, Motifs|PWM||AP-1
LJ LS	chr4	115909881	C	A	transversion	NDST4	intronic	NA	NA	6	Motifs|PWM||NF-E2, Motifs|PWM||AP-1
B103 B104	chr8	118537055	G	A	transition	MED30	intronic	CpG_site	NA	6	Motifs|PWM||Tcf3, Motifs|PWM||IRF8, Motifs|PWM||IRF2, Motifs|PWM||HMGA2, Motifs|PWM||IRF3, Motifs|PWM||ISGF-3, Motifs|PWM||IRF7, Motifs|PWM||DMRT7, Motifs|PWM||STAT1
B62 B63	chr2	122634222	A	G	transition	intergenic		NA	NA	6	Motifs|PWM||Brachyury
LS	chr12	123034737	A	T	transversion	KNTC1	intronic	NA	NA	6	Motifs|PWM||Pou2f3, Motifs|PWM||POU5F1P1, Motifs|PWM||Pou2f2, Motifs|PWM||POU3F3, Motifs|PWM||Oct-1, Motifs|PWM||Octamer, Motifs|PWM||POU2F3, Motifs|PWM||POU3F1, Motifs|PWM||OCT-x, Motifs|PWM||POU3F4, Motifs|PWM||POU2F1, Motifs|PWM||POU1F1, Motifs|PWM||POU2F2, Motifs|PWM||POU3F2
B62 B63	chrX	123121415	C	T	transition	STAG2	intronic	NA	NA	6	Motifs|PWM||Elf3, Motifs|PWM||Gata6
B62 B63	chr10	127420577	G	A	transition	EDRF1	intronic	CpG_site	NA	6	Motifs|PWM||Mybl1, Motifs|PWM||Myb
B113	chr9	127918364	G	A	transition	PPP6C	intronic	NA	NA	6	Motifs|PWM||Oct-1, Motifs|PWM||C/EBP, Motifs|PWM||Mafb, Motifs|PWM||Oct-4(POU5F1)
LJ LS	chr4	129175380	T	G	transversion	intergenic		NA	NA	6	Motifs|PWM||STAT1:STAT1
B93 B94	chr11	131293441	T	C	transition	NTM	intronic	NA	NA	6	Motifs|PWM||FOXP3, Chromatin_Structure||FAIRE|Nhbe

B123 B124	chr3	139887457	G	T	transversion	CLSTN2	intronic	NA	NA	6	Motifs|PWM||BARX1
B113 B114	chr6	145886359	G	C	transversion	intergenic		NA	NA	6	Motifs|PWM||FXR/RXR-alpha
LJ LS	chr2	146609689	G	A	transition	intergenic		NA	NA	6	Motifs|PWM||HNF4A
B93 B94	chr4	154787026	A	G	transition	intergenic		NA	NA	6	Motifs|PWM||HIC2
LJ LS	chr3	155766601	G	A	transition	intergenic		NA	NA	6	Motifs|PWM||MTF1, Motifs|PWM||C/EBP
B93 B94	chr6	163212623	C	A	transversion	PACRG	intronic	CpG_site	NA	6	Motifs|PWM||AR
B83 B84	chr3	164228118	A	C	transversion	intergenic		NA	NA	6	Motifs|PWM||RUSH-1alpha, Motifs|PWM||Hltf
B93 B94	chr6	165600454	T	C	transition	intergenic		NA	NA	6	Motifs|PWM||TP53, Motifs|PWM||p53
B62 B63	chr5	166938981	A	G	transition	TENM2	intronic	NA	NA	6	Motifs|PWM||GR
B123 B124	chr3	168345596	T	G	transversion	EGFEM1P	non-coding intronic	NA	NA	6	Motifs|PWM||E2F2, Motifs|PWM||Foxa2
LS	chr2	174321128	C	G	transversion	intergenic		NA	NA	6	Motifs|PWM||EWSR1-FLI1
B113 B114	chr3	174392522	T	C	transition	intergenic		NA	NA	6	Motifs|PWM||PU.1
B53 B54	chr1	182887203	C	A	transversion	SHCBP1L	intronic	NA	NA	6	Motifs|PWM||Tbp
B93 B94	chr2	182975989	A	T	transversion	PPP1R1C	intronic	NA	NA	6	Motifs|PWM||HFH8(FOXF1A), Motifs|PWM||Freac-7, Motifs|PWM||FOXO6, Motifs|PWM||FOXO3, Motifs|PWM||Zscan4, Motifs|PWM||FOXO4, Motifs|PWM||FOXO1
B103 B104	chr2	185753314	C	A	transversion	ZNF804A	intronic	NA	NA	6	Motifs|PWM||Zbtb3
LJ LS	chr2	186167711	T	G	transversion	intergenic		NA	NA	6	Motifs|PWM||HNF3alpha, Motifs|PWM||FOXJ2, Motifs|PWM||HFH3(FOXI1)
LJ LS	chr2	186167712	A	T	transversion	intergenic		NA	NA	6	Motifs|PWM||HNF3alpha, Motifs|PWM||FOXJ2, Motifs|PWM||HFH3(FOXI1)
B93 B94	chr1	186835728	A	G	transition	PLA2G4A	intronic	NA	NA	6	Motifs|PWM||FOXD3, Motifs|PWM||Irx2, Motifs|PWM||Irx6, Motifs|PWM||Irx-3, Motifs|PWM||BARHL2, Motifs|PWM||Tlx2, Motifs|PWM||Irx5, Motifs|PWM||Irx3, Motifs|PWM||RSRFC4, Motifs|PWM||Irx4, Motifs|PWM||AFP1
B83 B84	chr3	189327482	C	T	transition	intergenic		NA	NA	6	Motifs|PWM||Oct-1, Chromatin_Structure||FAIRE||Gliobla
B83 B84	chr1	218352394	A	G	transition	intergenic		NA	NA	6	Motifs|PWM||ISGF-3
B83 B84	chr2	230145171	G	A	transition	intergenic		NA	NA	6	Motifs|PWM||TFE, Motifs|PWM||Sox17, Motifs|PWM||Sox8
B53 B54	chr2	230274367	C	T	transition	DNER	intronic	CpG_site	NA	6	Motifs|PWM||SP1, Motifs|PWM||Sp1, Motifs|PWM||Ascl2
LJ LS	chr2	238513142	C	T	transition	intergenic		NA	NA	6	Motifs|Footprinting|Fibrop|E2A, Motifs|Footprinting|Hpde6e6e7|E2A, Motifs|Footprinting|Cll|E2A, Motifs|Footprinting|Medullo|E2A, Motifs|Footprinting|Hepg2|E2A, Motifs|Footprinting|Myometr|E2A, Motifs|Footprinting|Gliobla|E2A, Motifs|Footprinting|Gm12891|E2A, Motifs|Footprinting|Huh75|E2A, Motifs|Footprinting|LncapAndro|E2A, Motifs|Footprinting|Gm12878|E2A, Motifs|Footprinting|Gm12892|E2A, Motifs|Footprinting|Gm19238|E2A, Motifs|PWM||E2A, Motifs|Footprinting|Htr8|E2A, Motifs|Footprinting|8988t|E2A,
B62 B63	chr2	242283282	A	G	transition	SEPT2	nonsyn p.N271S	NA	NA	6	Motifs|PWM||ESR2
B103 B104	chr17	2421858	C	T	transition	intergenic		CpG_site	NA	7	No data
LJ	chr19	3354318	T	C	transition	intergenic		NA	NA	7	No data
B123 B124	chr3	3361010	G	C	transversion	intergenic		NA	NA	7	No data
LJ LS	chr8	3381974	C	A	transversion	CSMD1	intronic	NA	NA	7	No data
B103 B104	chr8	5214640	G	A	transition	intergenic		NA	NA	7	No data
B93 B94	chr4	5962283	A	G	transition	intergenic		NA	NA	7	No data
B123 B124	chr3	7391648	G	A	transition	GRM7	intronic	CpG_site	NA	7	No data
B84	chr7	8681356	T	C	transition	NXPH1	intronic	NA	NA	7	No data
B123 B124	chr9	11027822	C	T	transition	intergenic		NA	NA	7	No data
B83 B84	chr5	11250985	G	A	transition	CTNND2	intronic	CpG_site	NA	7	No data
B103 B104	chr9	11417777	A	T	transversion	intergenic		NA	NA	7	No data
B103 B104	chr12	11507171	G	A	transition	PRB1	intronic	NA	NA	7	No data
B62 B63	chr17	12179178	A	G	transition	intergenic		NA	NA	7	No data
B53 B54	chr4	12439530	C	A	transversion	intergenic		NA	NA	7	No data

LJ LS	chr17	13733612	C	T	transition	intergenic		NA	NA	7	No data
B83 B84	chr17	13734863	G	A	transition	intergenic		NA	NA	7	No data
B53 B54	chr1	14607191	C	T	transition	intergenic		NA	NA	7	No data
B123 B124	chr9	15437524	C	T	transition	SNAPC3	intronic	NA	NA	7	No data
B123 B124	chr10	15589618	C	T	transition	ITGA8	intronic	NA	NA	7	No data
B123 B124	chr4	15798530	G	T	transversion	CD38	intronic	NA	NA	7	No data
B83 B84	chr7	16224899	A	G	transition	ISPD	intronic	NA	NA	7	No data
B62 B63	chr2	17073960	C	T	transition	intergenic		NA	NA	7	No data
B62 B63	chr12	17246962	A	G	transition	intergenic		NA	NA	7	No data
B53 B54	chr6	17265790	C	T	transition	intergenic		NA	NA	7	No data
LJ LS	chrY	17459241	A	G	transition	intergenic		NA	NA	7	No data
B83 B84	chr21	17868492	A	G	transition	LINC00478	non-coding intronic	NA	NA	7	No data
B93 B94	chr7	17914832	C	A	transversion	SNX13	intronic	NA	NA	7	No data
B123 B124	chr12	19654480	A	T	transversion	AEBP2	intronic	NA	NA	7	No data
B123 B124	chr10	20845041	C	T	transition	intergenic		CpG_site	NA	7	No data
B62 B63	chr13	20923983	G	A	transition	intergenic		CpG_site	NA	7	No data
B93 B94	chr19	21062165	C	T	transition	intergenic		NA	NA	7	No data
B123 B124	chr3	21088283	C	T	transition	intergenic		NA	NA	7	No data
B83 B84	chr10	21149815	G	C	transversion	NEBL	intronic	NA	NA	7	No data
B93 B94	chr10	21979949	C	G	transversion	MLLT10	intronic	NA	NA	7	No data
B93 B94	chr8	22898794	T	C	transition	TNFRSF10B	intronic	NA	NA	7	No data
B113 B114	chr16	24377252	A	G	transition	intergenic		NA	NA	7	No data
B53 B54	chr7	24594488	T	C	transition	intergenic		NA	NA	7	No data
B123 B124	chr9	24903909	G	A	transition	intergenic		NA	NA	7	No data
B62 B63	chr3	24933640	C	T	transition	intergenic		NA	NA	7	No data
B93 B94	chr9	25091094	G	C	transversion	intergenic		CpG_site	NA	7	No data
B83 B84	chr21	25258273	G	C	transversion	intergenic		CpG_site	NA	7	No data
B62 B63	chr9	25628943	T	C	transition	intergenic		NA	NA	7	No data
B113	chr12	26742696	A	G	transition	ITPR2	intronic	NA	NA	7	No data
B62 B63	chr12	28411429	G	T	transversion	CCDC91	intronic	NA	NA	7	No data
B83 B84	chr14	30014118	A	G	transition	MIR548AI	non-coding intronic	NA	NA	7	No data
B83 B84	chr3	31020364	A	G	transition	intergenic		NA	NA	7	No data
B83 B84	chr17	31223286	C	T	transition	intergenic		NA	NA	7	No data
B113 B114	chr18	32683856	G	C	transversion	MAPRE2	intronic	NA	NA	7	No data
B123 B124	chr4	33398722	C	T	transition	intergenic		CpG_site	NA	7	No data
B113 B114	chr6	34401949	G	A	transition	intergenic		CpG_site	NA	7	No data
B53 B54	chr9	34617204	C	T	transition	DCTN3	intronic	NA	NA	7	No data
B103 B104	chr20	34752676	C	T	transition	EPB41L1	intronic	CpG_site	NA	7	No data
B123 B124	chr16	35013471	T	A	transversion	intergenic		NA	NA	7	No data
B53 B54	chr13	36215362	A	C	transversion	NBEA	intronic	NA	NA	7	No data
B84	chr4	36920104	A	C	transversion	intergenic		NA	NA	7	No data
B103 B104	chr20	37200837	G	T	transversion	RALGAPB	intronic	NA	NA	7	No data
LJ LS	chr18	38732423	A	G	transition	intergenic		NA	NA	7	No data
B93 B94	chr15	39986654	C	G	transversion	FSIP1	intronic	NA	NA	7	No data
B123 B124	chr21	42082429	G	A	transition	DSCAM	intronic	CpG_site	NA	7	No data
B103 B104	chrX	42471130	T	G	transversion	intergenic		NA	NA	7	No data
B103 B104	chr13	43170170	G	A	transition	TNFSF11	intronic	CpG_site	NA	7	No data
B53 B54	chr8	43297461	T	G	transversion	intergenic		NA	NA	7	No data
B62 B63	chr2	43609005	G	C	transversion	THADA	intronic	NA	NA	7	No data
B62 B63	chr4	46434769	G	A	transition	intergenic		NA	NA	7	No data

B123 B124	chr14	46455725	T	G	transversion	intergenic		NA	NA	7	No data
B103 B104	chr8	47649158	A	G	transition	intergenic		NA	NA	7	No data
LJ LS	chr16	47797029	T	C	transition	intergenic		NA	NA	7	No data
B83 B84	chr15	48506290	T	C	transition	SLC12A1	intronic	NA	NA	7	No data
LJ LS	chr10	53069364	T	C	transition	PRKG1	intronic	NA	NA	7	No data
B62 B63	chr18	53310663	A	G	transition	intergenic		NA	NA	7	No data
B123 B124	chr20	55421177	T	G	transversion	intergenic		NA	NA	7	No data
B53 B54	chr8	56129326	G	A	transition	XKR4	intronic	NA	NA	7	No data
B123 B124	chr6	56419279	C	T	transition	DST	intronic	NA	NA	7	No data
B53 B54	chr11	57175782	C	T	transition	SLC43A3	intronic	NA	NA	7	No data
B62 B63	chr2	57552402	T	A	transversion	intergenic		NA	NA	7	No data
B62 B63	chrX	58380768	A	C	transversion	intergenic		NA	NA	7	No data
B103 B104	chr12	58842837	C	G	transversion	intergenic		NA	NA	7	No data
B123 B124	chr3	60760326	C	G	transversion	FHIT	intronic	NA	NA	7	No data
B62 B63	chr8	62392971	G	A	transition	CLVS1	intronic	CpG_site	NA	7	No data
B103 B104	chr5	63649074	T	A	transversion	RNF180	intronic	NA	NA	7	No data
B123	chr1	63702337	A	C	transversion	LINC00466	non-coding intronic	NA	NA	7	No data
B93 B94	chr1	64191122	G	A	transition	intergenic		NA	NA	7	No data
B103 B104	chr18	64205104	G	A	transition	CDH19	intronic	NA	NA	7	No data
B62 B63	chr7	64318208	C	T	transition	intergenic		NA	NA	7	No data
B62 B63	chr14	64473123	C	T	transition	SYNE2	intronic	CpG_site	NA	7	No data
B83 B84	chr13	64665172	G	T	transversion	intergenic		NA	NA	7	No data
B62 B63	chr6	64923693	C	T	transition	EYS	intronic	CpG_site	NA	7	No data
LJ LS	chr10	64995156	A	G	transition	JMJD1C	intronic	NA	NA	7	No data
B93 B94	chr4	66240782	A	G	transition	EPHA5	intronic	NA	NA	7	No data
B103 B104	chr17	66580685	C	T	transition	FAM20A	intronic	CpG_site	NA	7	No data
B114	chr9	66861395	T	C	transition	intergenic		NA	NA	7	No data
B62 B63	chr2	68293158	A	G	transition	intergenic		NA	NA	7	No data
LJ LS	chr4	70243771	G	C	transversion	intergenic		NA	NA	7	No data
B93 B94	chr18	72619462	C	T	transition	ZNF407	intronic	NA	NA	7	No data
B103 B104	chr13	72813354	G	T	transversion	intergenic		NA	NA	7	No data
B93 B94	chr10	72842217	A	G	transition	intergenic		NA	NA	7	No data
LJ LS	chr10	73143664	G	A	transition	intergenic		NA	NA	7	No data
LJ LS	chr13	73798380	G	A	transition	intergenic		CpG_site	NA	7	No data
B103 B104	chr13	74635919	C	T	transition	KLF12	intronic	CpG_site	NA	7	No data
B83 B84	chr10	76753997	G	A	transition	KAT6B	intronic	NA	NA	7	No data
B123 B124	chr13	76971899	G	C	transversion	intergenic		NA	NA	7	No data
B93 B94	chr9	77343121	T	A	transversion	TRPM6	intronic	NA	NA	7	No data
B103 B104	chr14	78571385	T	C	transition	intergenic		NA	NA	7	No data
B123 B124	chr5	78999514	T	C	transition	CMYA5	intronic	NA	NA	7	No data
B93 B94	chr16	81098864	G	A	transition	C16orf46	intronic	NA	NA	7	No data
B103 B104	chrX	81626563	A	G	transition	intergenic		NA	NA	7	No data
B103 B104	chr2	82569412	T	C	transition	intergenic		NA	NA	7	No data
B123 B124	chr9	82729515	A	G	transition	intergenic		NA	NA	7	No data
B83 B84	chr11	87164141	C	T	transition	intergenic		CpG_site	NA	7	No data
B123 B124	chr10	87386677	A	C	transversion	GRID1	intronic	NA	NA	7	No data
B93 B94	chr3	88598299	A	G	transition	intergenic		NA	NA	7	No data
B103 B104	chr2	88639215	C	T	transition	intergenic		CpG_site	NA	7	No data
B93 B94	chr9	89456759	C	T	transition	intergenic		CpG_site	NA	7	No data
B113 B114	chr16	89961140	C	A	transversion	TCF25	intronic	CpG_site	NA	7	No data

B93 B94	chr11	90849241	A	G	transition	intergenic		NA	NA	7	No data
B62 B63	chr5	92461001	A	C	transversion	intergenic		NA	NA	7	No data
B103 B104	chr11	94241750	C	A	transversion	intergenic		NA	NA	7	No data
B62 B63	chr10	95127010	C	T	transition	MYOF	intronic	CpG_site	NA	7	No data
LJ LS	chr11	97265884	G	C	transversion	intergenic		NA	NA	7	No data
B123 B124	chr9	99136093	G	C	transversion	SLC35D2	intronic	CpG_site	NA	7	No data
B62 B63	chr3	99546146	G	A	transition	CMSS1	intronic	CpG_site	NA	7	No data
B123 B124	chr8	100386318	A	G	transition	VPS13B	intronic	NA	NA	7	No data
B93 B94	chr4	103002276	C	T	transition	intergenic		CpG_site	NA	7	No data
B113 B114	chr12	103506236	C	T	transition	intergenic		NA	NA	7	No data
B93 B94	chr7	103763484	C	T	transition	intergenic		NA	NA	7	No data
B62 B63	chr11	104908704	C	T	transition	intergenic		NA	NA	7	No data
B83 B84	chr1	105309724	G	A	transition	intergenic		CpG_site	NA	7	No data
LJ LS	chr9	105381829	C	T	transition	LINC00587	non-coding intronic	CpG_site	NA	7	No data
B103 B104	chr8	106280832	T	G	transversion	intergenic		NA	NA	7	No data
B62 B63	chr4	106901329	G	A	transition	intergenic		CpG_site	NA	7	No data
B62 B63	chrX	108826763	C	T	transition	intergenic		CpG_site	NA	7	No data
B113 B114	chr7	108858732	G	T	transversion	intergenic		NA	NA	7	No data
B123 B124	chr3	111154558	G	T	transversion	intergenic		NA	NA	7	No data
B62 B63	chrX	111417043	G	A	transition	ZCCHC16	intronic	NA	NA	7	No data
B53 B54	chr8	111879636	T	A	transversion	intergenic		NA	NA	7	No data
B62 B63	chr5	113042944	C	T	transition	intergenic		CpG_site	NA	7	No data
B53 B54	chr4	114760417	T	A	transversion	intergenic		NA	NA	7	No data
B93 B94	chr11	116421503	A	T	transversion	intergenic		NA	NA	7	No data
LJ LS	chr10	118484331	C	T	transition	HSPA12A	intronic	NA	NA	7	No data
LJ LS	chr1	118539438	T	C	transition	SPAG17	intronic	NA	NA	7	No data
B113 B114	chr5	118782822	G	A	transition	intergenic		CpG_site	NA	7	No data
B83 B84	chr10	118876496	A	G	transition	KIAA1598	intronic	NA	NA	7	No data
B83	chr4	119231662	C	T	transition	PRSS12	intronic	CpG_site	NA	7	No data
B53 B54	chr12	120569035	C	G	transversion	GCN1L1	nonsyn p.S2506T	NA	NA	7	No data
B53 B54	chr6	123917045	A	G	transition	TRDN	intronic	NA	NA	7	No data
B53 B54	chr2	124712603	A	C	transversion	intergenic		NA	NA	7	No data
B123 B124	chr7	125495096	C	T	transition	intergenic		NA	NA	7	No data
B103 B104	chr10	128776253	G	T	transversion	DOCK1	intronic (splice_site)	NA	NA	7	No data
B83 B84	chr4	132378202	C	T	transition	intergenic		NA	NA	7	No data
B93 B94	chr7	132653469	G	A	transition	CHCHD3	intronic	CpG_site	NA	7	No data
B53 B54	chr6	133263812	G	C	transversion	intergenic		NA	NA	7	No data
B83 B84	chr2	137786478	C	T	transition	THSD7B	intronic	CpG_site	NA	7	No data
B93 B94	chr2	138486744	A	G	transition	intergenic		NA	NA	7	No data
B83 B84	chr8	138597674	G	C	transversion	intergenic		NA	NA	7	No data
B53 B54	chr6	140126330	A	G	transition	LOC10013273	non-coding intronic	NA	NA	7	No data
B103 B104	chr3	140390293	C	T	transition	intergenic		NA	NA	7	No data
B113 B114	chr6	142436831	C	G	transversion	intergenic		NA	NA	7	No data
B123 B124	chrX	144020346	G	A	transition	intergenic		NA	NA	7	No data
B93 B94	chr6	146456876	G	A	transition	GRM1	intronic	NA	NA	7	No data
B62 B63	chr2	149183277	G	A	transition	MBD5	intronic	CpG_site	NA	7	No data
B84	chr6	149959476	T	A	transversion	KATNA1	intronic	NA	NA	7	No data
B62 B63	chrX	150711814	G	T	transversion	intergenic		NA	NA	7	No data
B103 B104	chr7	152835331	A	G	transition	intergenic		NA	NA	7	No data
B104	chr2	153664053	G	A	transition	intergenic		CpG_site	NA	7	No data

B93 B94	chr2	156799239	A	G	transition	intergenic		NA	NA	7	No data
B103 B104	chr4	162128420	C	T	transition	intergenic		NA	NA	7	No data
B62 B63	chr6	163579077	G	A	transition	PACRG	intronic	NA	NA	7	No data
B93 B94	chr2	164338918	G	A	transition	intergenic		NA	NA	7	No data
B84	chr2	167878685	A	G	transition	XIRP2	intronic	NA	NA	7	No data
B83 B84	chr2	172987977	C	T	transition	intergenic		NA	NA	7	No data
B103 B104	chr3	178375930	C	T	transition	KCNMB2	intronic	NA	NA	7	No data
B53 B54	chr2	179436794	C	T	transition	TTN	nonsyn p.V23048I	NA	NA	7	No data
B53 B54	chr1	191364591	T	G	transversion	intergenic		NA	NA	7	No data
B113 B114	chr2	207501019	T	C	transition	intergenic		NA	NA	7	No data
B93 B94	chr2	212168526	A	G	transition	intergenic		NA	NA	7	No data
B113 B114	chr2	220870844	T	C	transition	intergenic		NA	NA	7	No data
B93 B94	chr1	224422130	T	C	transition	NVL	intronic	NA	NA	7	No data
B62 B63	chr1	231116417	A	G	transition	TTC13	5upstream	NA	NA	7	No data
B93 B94	chr2	241957475	G	A	transition	SNED1	intronic	CpG_site	NA	7	No data

* Notes:
1a -- eQTL + TF binding + matched TF motif + matched DNase Footprint + DNase peak
1b -- eQTL + TF binding + any motif + DNase Footprint + DNase peak
1c -- eQTL + TF binding + matched TF motif + DNase peak
1d -- eQTL + TF binding + any motif + DNase peak
1e -- eQTL + TF binding + matched TF motif
1f -- eQTL + TF binding / DNase peak
2a -- TF binding + matched TF motif + matched DNase Footprint + DNase peak
2b -- TF binding + any motif + DNase Footprint + DNase peak
2c -- TF binding + matched TF motif + DNase peak
3a -- TF binding + any motif + DNase peak
3b -- TF binding + matched TF motif
4 -- TF binding + DNase peak
5 -- TF binding or DNase peak
6 -- other