SUPPLEMENTARY DATA

Supplementary Table 1. Primer Sequences used for the expression quantitative trait loci (eQTL) analysis of the genes and isoforms in LD block 16p13.13

NCBI Ref Seq	**Gene**	**Forward Primer**	**Reverse Primer**	**Size (bp)**
BQ221200	*CIITA III**	ggggaagctgagggcacg	gaagctccaggtagccaccttcta	182
BM147267	*CIITA IV **	gcggccccagagctgg	gaagctccaggtagccaccttcta	125
NM_015226.2	*CLEC16A Iso I*	cgcatgaagatgcagagaat	ggtggcgactgtgaggac	257
BC112897.1	*CLEC16A Iso II*	ctctgtggagcctgtgtgag	tgtttaatcttgcgctgtgc	102
AK126771.1	*CLEC16A Iso III*	acagcccagagtccagca	actcatgggggcatctgaat	157
NM_014015.3	*DEXI* †	agtagggtctcgactgc**t**gttc	acctctctgttggcacgaag	192
NM_003745.1	*SOCS*	gccccttctgtaggatggta	ggaggaagaggaggaaggtt	104
NM_002046.3	*GAPDH*	cagccgagccacatcgc	catgggtggaatcatattggaaca	186
Novel Transcript	*CIITA V*	acctgcaacaacaggattcac	caccaacacctcagacttcatc	105
Novel Transcript	*CIITA VI*	agctcgctgccagccttc	ggattctcgcttgtccctgt	159

**CIITA III* and *CIITA IV* primers were originally designed by Hornell 2003 (50)

†There are two intron-less *DEXI* pseudogenes (retro-DEXI) located on chromosome 15 at coordinates (Hg19) (29,033,933-29,035,470) and (23,157,046-23,158,548) whose genomic DNA matches almost exactly the cDNA for *DEXI*. The forward primer has one nucleotide difference at the 3' end (**bold**) that differ from the pseudogenes.

Supplementary Table 2. SNPs in *CLEC16A* most strongly associated with type 1 diabetes (total 48) from TDT analysis sorted in order of base position.

SNP Name	Position (Hg19)	MAF*	Minor Allele	Major Allele	Transmitted Minor Allele Count	Untransmitted Minor Allele Count	TDT *P*-value†	Odds Ratio†	Location‡
rs12708713	11072459	0.167	T	C	1045	1239	4.92E-05	0.843 (0.777-0.916)	Intron 8
rs12708714	11072525	0.168	C	T	1048	1244	4.24E-05	0.842 (0.776-0.915)	Intron 8
rs7403919	11085968	0.341	C	T	1691	1938	4.13E-05	0.873 (0.818-0.931)	Intron 10
rs8062322	11092319	0.289	A	C	1529	1768	3.15E-05	0.865 (0.808-0.926)	Intron 10
rs9926367	11093179	0.288	C	T	1520	1759	3.00E-05	0.864 (0.807-0.926)	Intron 10
rs7200940	11164567	0.307	G	C	1556	1804	1.88E-05	0.863 (0.806-0.923)	Intron 19
rs11860603	11165017	0.305	C	T	1546	1796	1.53E-05	0.861 (0.804-0.921)	Intron 19
rs9934969	11166002	0.303	A	G	1540	1795	1.01E-05	0.858 (0.802-0.918)	Intron 19
rs11865121§	11166688	0.304	A	C	1544	1798	1.11E-05	0.859 (0.802-0.919)	Intron 19
rs7198621	11167458	0.304	G	C	1538	1779	2.86E-05	0.865 (0.808-0.926)	Intron 19
rs725613	11169683	0.336	C	A	1625	1874	2.56E-05	0.867 (0.811-0.927)	Intron 19
rs12925642	11171602	0.304	G	A	1548	1797	1.67E-05	0.861 (0.805-0.922)	Intron 19
rs2058531	11172109	0.305	G	A	1554	1807	1.28E-05	0.860 (0.804-0.920)	Intron 19
rs9652601	11174365	0.306	A	G	1557	1792	4.89E-05	0.869 (0.812-0.93)	Intron 19
rs9652582	11174564	0.300	A	G	1536	1783	1.81E-05	0.862 (0.805-0.922)	Intron 19
rs2041670	11174652	0.300	T	C	1537	1785	1.69E-05	0.861 (0.804-0.922)	Intron 19
rs11648679	11175984	0.335	G	A	1623	1877	1.76E-05	0.865 (0.809-0.924)	Intron 19
rs12708715	11177824	0.301	T	C	1542	1795	1.19E-05	0.859 (0.803-0.92)	Intron 19
rs9929994	11178245	0.336	G	A	1617	1873	1.47E-05	0.863 (0.808-0.923)	Intron 19
rs12708716‖	11179873	0.333	G	A	1615	1866	2.10E-05	0.866 (0.810-0.925)	Intron 19
rs9888908	11181244	0.300	A	C	1539	1790	1.36E-05	0.860 (0.803-0.92)	Intron 19
rs7203793	11182134	0.335	G	C	1626	1876	2.39E-05	0.867 (0.811-0.926)	Intron 19
rs12924729¶	11187783	0.305	A	G	1565	1806	3.31E-05	0.867 (0.810-0.927)	Intron 19
rs12928537	11191400	0.303	A	G	1550	1798	1.82E-05	0.862 (0.806-0.923)	Intron 19
rs12927355	11194771	0.302	T	C	1556	1802	2.18E-05	0.864 (0.807-0.924)	Intron 19
rs741172	11200798	0.300	T	C	1551	1803	1.35E-05	0.860 (0.804-0.921)	Intron 19
rs9746695	11207894	0.302	C	T	1550	1799	1.69E-05	0.862 (0.805-0.922)	Intron 19
rs12935413	11210447	0.333	A	G	1617	1865	2.64E-05	0.867 (0.811-0.927)	Intron 19
rs34306440#	11215035	0.209	G	A	1203	1436	5.74E-06	0.838 (0.776-0.904)	Intron 20
rs35032408	11215424	0.208	G	T	1204	1429	1.16E-05	0.843 (0.780-0.910)	Intron 20
rs8064154	11219419	0.338	G	A	1610	1870	1.05E-05	0.861 (0.806-0.920)	Intron 21
rs12924112	11219720	0.336	G	T	1589	1849	9.24E-06	0.859 (0.804-0.919)	Intron 21
rs34540843	11221287	0.242	G	A	1332	1555	3.32E-05	0.857 (0.796-0.922)	Intron 22
rs36045143	11224966	0.242	G	A	1332	1564	1.62E-05	0.852 (0.792-0.916)	Intron 22
rs2241099	11225064	0.241	G	C	1325	1555	1.82E-05	0.852 (0.792-0.917)	Intron 22
rs12919828	11226779	0.243	G	A	1335	1567	1.66E-05	0.852 (0.792-0.917)	Intron 22
rs35732840	11228355	0.336	G	T	1606	1860	1.60E-05	0.863 (0.808-0.923)	Intron 22
rs62026376	11228712	0.243	T	C	1331	1569	9.89E-06	0.848 (0.789-0.913)	Intron 22

rs62026377	11229128	0.243	T	G	1327	1564	1.04E-05	0.849 (0.789-0.913)	Intron 22
rs7203459	11230703	0.242	C	T	1324	1558	1.31E-05	0.850 (0.790-0.914)	Intron 22
rs2867880	11231857	0.334	A	G	1604	1856	1.84E-05	0.864 (0.808-0.924)	Intron 22
rs12919732	11235123	0.244	G	A	1321	1558	1.00E-05	0.848 (0.788-0.912)	Intron 22
rs2903692‖	11238783	0.330	A	G	1581	1844	6.99E-06	0.857 (0.802-0.917)	Intron 22
rs4322688	11238991	0.332	G	C	1590	1856	5.86E-06	0.857 (0.801-0.916)	Intron 22
rs12930373	11239599	0.331	A	G	1590	1847	1.17E-05	0.861 (0.805-0.921)	Intron 22
rs12917893	11239978	0.331	T	A	1584	1842	1.04E-05	0.860 (0.804-0.92)	Intron 22
rs62026379	11241806	0.241	A	G	1314	1556	6.27E-06	0.845 (0.785-0.909)	Intron 22
rs17673553	11241906	0.242	G	A	1326	1558	1.56E-05	0.851 (0.791-0.916)	Intron 22

*MAF was calculated from the founders (4,655 individuals). †TDT *P*-values and Odds Ratio were calculated from the 3,070 multiplex families (12,485 individuals). ‡ Intronic regions were based on Ensembl (CLEC16A - 001 ENST00000409790) (51). §GWAS reported SNP in association with multiple sclerosis (8). ‖ GWAS reported SNPs in association with type 1 diabetes (3,4). ¶GWAS reported SNP previously reported in association with primary biliary cirrhosis (11).GWAS reported SNPs were limited to the dbGAP NHGRI GWAS catalog. #Most significant SNP associated with type 1 diabetes. MAF, minor allele frequency.

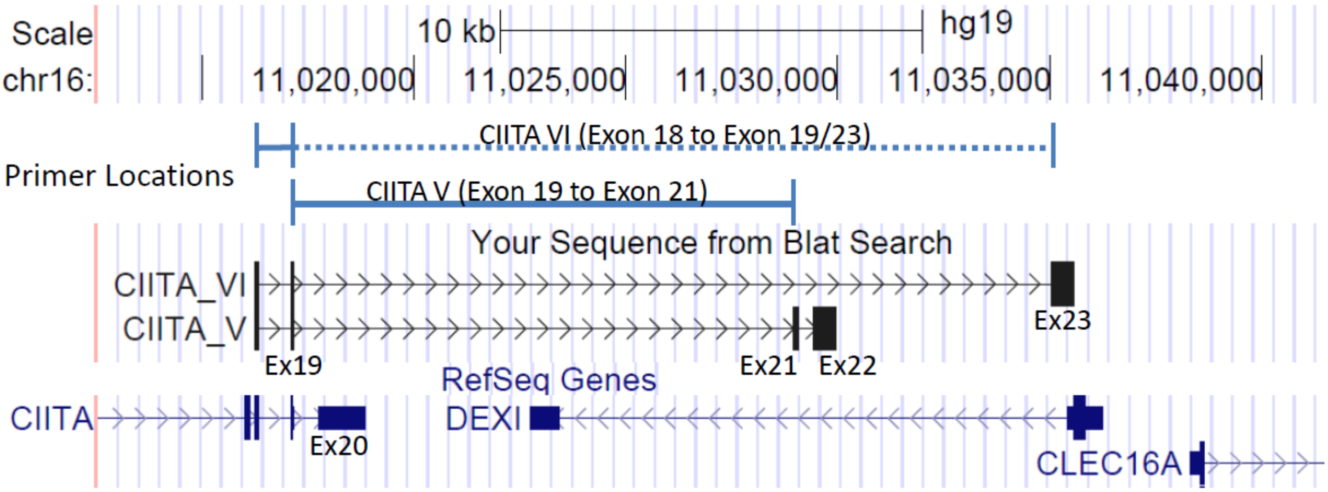
Supplementary Table 3. SNPs Significantly Associated with Type 1 Diabetes with their Corresponding eQTL Results for *DEXI*

SNP	Position (Hg19)	Minor Allele	β*	STAT*	*P*-value*	r^2 rs7403919†	r^2 rs34306440†	Location‡
rs7403919	11085968	C	0.9029	4.404	1.25E-04	1.000	0.533	Intron 10
rs725613	11169683	C	0.8884	4.289	1.72E-04	0.940	0.501	Intron 19
rs11648679	11175984	G	0.8884	4.289	1.72E-04	0.940	0.501	Intron 19
rs9929994	11178245	G	0.8884	4.289	1.72E-04	0.940	0.501	Intron 19
rs12708716	11179873	G	0.8884	4.289	1.72E-04	0.940	0.501	Intron 19
rs7203793	11182134	G	0.8884	4.289	1.72E-04	0.940	0.501	Intron 19
rs12935413	11210447	A	0.8884	4.289	1.72E-04	0.940	0.501	Intron 19
rs8064154	11219419	G	0.8884	4.289	1.72E-04	0.940	0.501	Intron 21
rs12924112	11219720	G	0.8884	4.289	1.72E-04	0.940	0.501	Intron 21
rs4322688	11238991	G	0.8884	4.289	1.72E-04	0.940	0.501	Intron 22
rs12930373	11239599	A	0.8884	4.289	1.72E-04	0.940	0.501	Intron 22
rs2903692	11238783	A	0.8884	4.289	1.72E-04	0.940	0.529	Intron 22
rs35732840	11228355	G	0.8934	4.306	1.64E-04	0.875	0.467	Intron 22
rs2867880	11231857	A	0.8934	4.306	1.64E-04	0.875	0.467	Intron 22
rs12917893	11239978	T	0.8518	3.994	3.88E-04	0.875	0.467	Intron 22
rs7200940	11164567	G	0.7586	3.799	6.61E-04	0.844	0.645	Intron 19
rs8062322	11092319	A	0.7974	3.502	1.47E-03	0.834	0.578	Intron 10
rs9926367	11093179	C	0.7974	3.502	1.47E-03	0.834	0.578	Intron 10
rs11860603	11165017	C	0.7596	3.586	1.17E-03	0.790	0.622	Intron 19
rs9934969	11166002	A	0.7596	3.586	1.17E-03	0.790	0.622	Intron 19
rs11865121	11166688	A	0.7596	3.586	1.17E-03	0.790	0.622	Intron 19
rs7198621	11167458	G	0.7596	3.586	1.17E-03	0.790	0.622	Intron 19
rs12925642	11171602	G	0.7596	3.586	1.17E-03	0.790	0.622	Intron 19
rs2058531	11172109	G	0.7596	3.586	1.17E-03	0.790	0.622	Intron 19
rs9652601	11174365	A	0.7596	3.586	1.17E-03	0.790	0.622	Intron 19
rs9652582	11174564	A	0.7596	3.586	1.17E-03	0.790	0.622	Intron 19
rs2041670	11174652	T	0.7596	3.586	1.17E-03	0.790	0.622	Intron 19
rs12708715	11177824	T	0.7596	3.586	1.17E-03	0.790	0.622	Intron 19
rs9888908	11181244	A	0.7596	3.586	1.17E-03	0.790	0.622	Intron 19
rs12928537	11191400	A	0.7596	3.586	1.17E-03	0.790	0.622	Intron 19
rs12927355	11194771	T	0.7596	3.586	1.17E-03	0.790	0.622	Intron 19
rs741172	11200798	T	0.7596	3.586	1.17E-03	0.790	0.622	Intron 19
rs9746695	11207894	C	0.7596	3.586	1.17E-03	0.790	0.622	Intron 19
rs12924729	11187783	A	0.6429	3.1	4.19E-03	0.737	0.511	Intron 19
rs34540843	11221287	G	0.9382	2.98	5.67E-03	0.600	0.680	Intron 22
rs62026379	11241806	A	0.9382	2.98	5.67E-03	0.600	0.680	Intron 22
rs17673553	11241906	G	0.9382	2.98	5.67E-03	0.600	0.680	Intron 22
rs34306440§	11215035	G	0.8521	2.561	1.57E-02	0.533	1.000	Intron 20

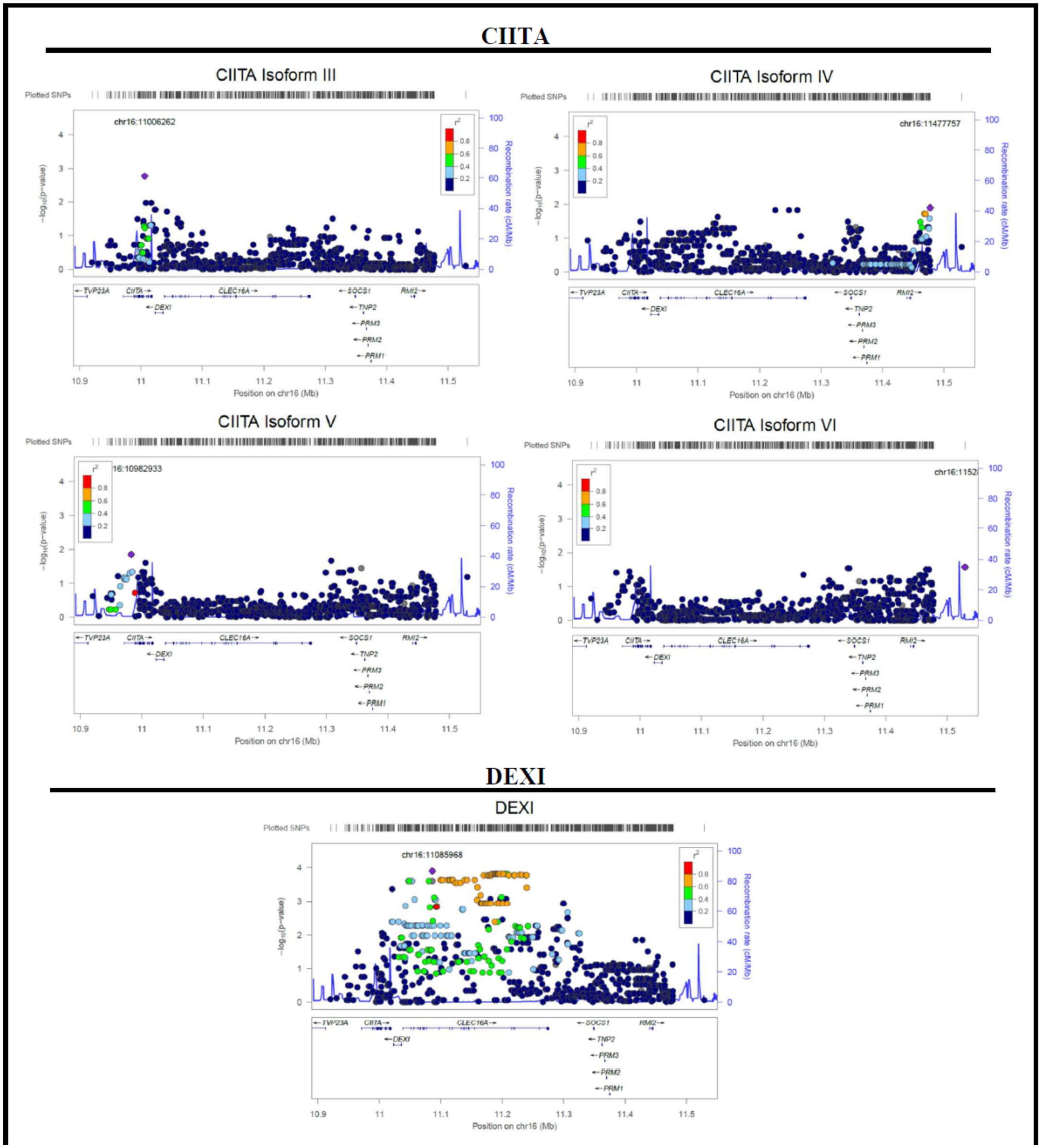
rs35032408	11215424	G	0.8521	2.561	1.57E-02	0.533	1.000	Intron 20
rs36045143	11224966	G	0.9253	2.95	6.10E-03	0.500	0.600	Intron 22
rs2241099	11225064	G	0.9253	2.95	6.10E-03	0.500	0.600	Intron 22
rs62026376	11228712	T	0.9253	2.95	6.10E-03	0.500	0.600	Intron 22
rs62026377	11229128	T	0.9253	2.95	6.10E-03	0.500	0.600	Intron 22
rs12919732	11235123	G	0.8494	2.649	1.28E-02	0.500	0.600	Intron 22
rs12919828	11226779	G	0.9253	2.95	6.10E-03	0.485	0.592	Intron 22
rs7203459	11230703	C	0.8583	2.663	1.23E-02	0.412	0.529	Intron 22
rs12708713	11072459	T	0.5901	1.6	1.20E-01	0.394	0.638	Intron 8
rs12708714	11072525	C	0.5901	1.6	1.20E-01	0.394	0.638	Intron 8

Samples are sorted in order of r^2 relative to rs7403919 (most significant SNP that is associated with *DEXI* expression levels). *Statistical analysis performed using expression and genotyping data. †r^2 values were calculated using genotyping data for 33 HapMap samples and the designated SNP as reference. ‡Intronic regions were based on Ensembl (CLEC16A - 001 ENST00000409790) (51). §Most Significant SNP associated with type 1 diabetes. β, linear regression coefficient; STAT, coefficient t-statistic; r^2, correlation.

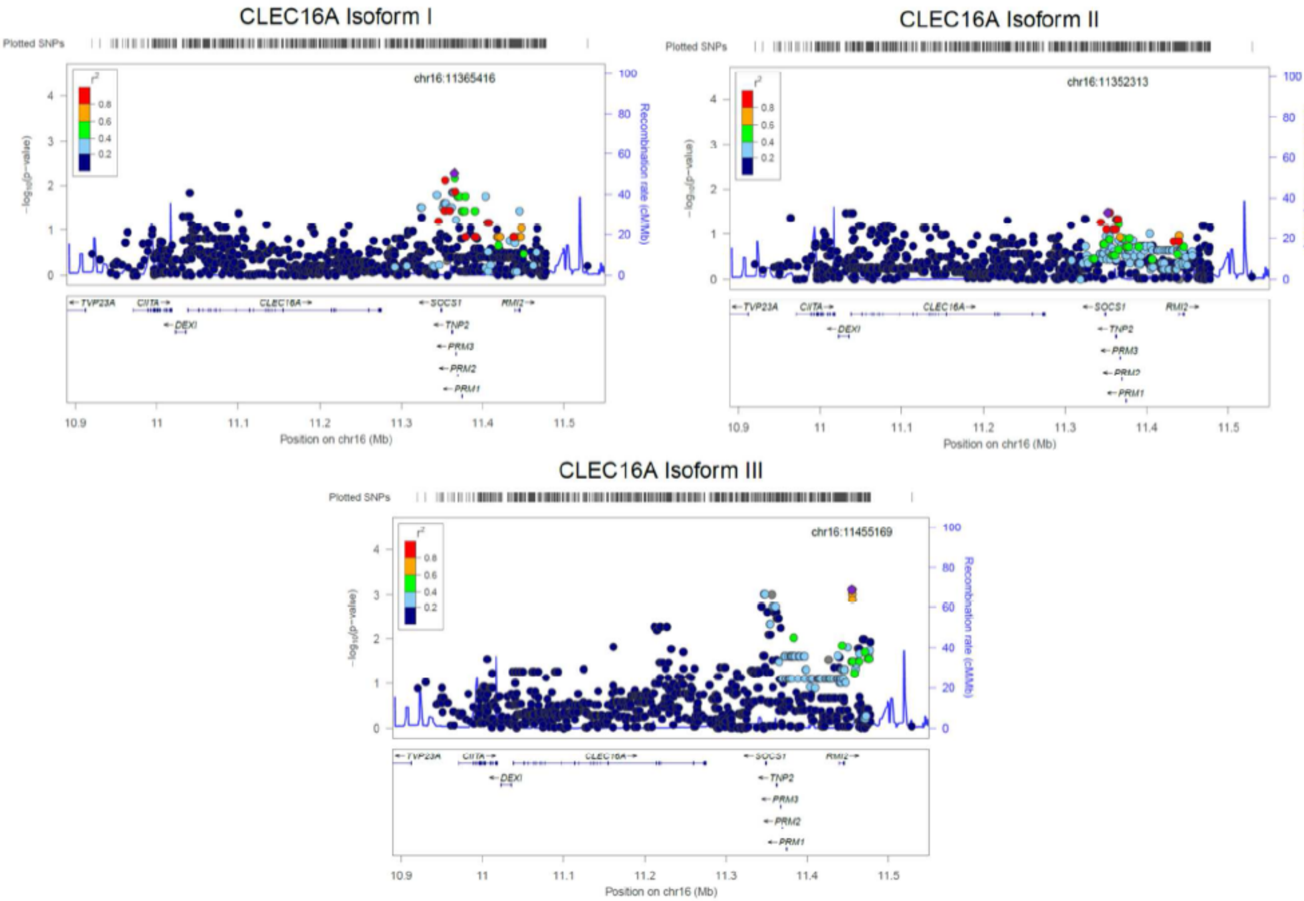
Supplementary Figure 1. New Isoforms of *CIITA* were identified that have exons that splice either into the middle intronic region of *DEXI* or splice into exon 1 of *DEXI*. These two isoforms have been named *CIITA V*, and *CIITA VI*. Start sites of the new isoforms are not known. Primers were designed that amplified these two new isoforms. The exact locations of primers are noted in the above figure. The reverse primer for *CIITA VI* straddled two exons (designated by the dotted line).



Supplementary Figure 2. Association between SNP alleles and transcript levels for genes in chromosomal region 16p13.13. The –log (*P*-value) for the association is plotted relative to physical position on the chromosome for each SNP. The r^2 is plotted relative to the most significant SNP.



CLEC16A



SOCS1