

## ORIGINAL ARTICLE

# Overview of the Rapid Response data

WM Brown<sup>1</sup>, JJ Pierce<sup>1</sup>, JE Hilner<sup>2</sup>, LH Perdue<sup>1</sup>, K Lohman<sup>1</sup>, L Lu<sup>1</sup>, PIW de Bakker<sup>3</sup>, K Irenze<sup>3</sup>, L Ziaugra<sup>3</sup>, DB Mirel<sup>3</sup> and the Type I Diabetes Genetics Consortium

<sup>1</sup>Division of Public Health Sciences, Department of Biostatistics, Wake Forest University Health Sciences, Winston-Salem, NC, USA;

<sup>2</sup>Department of Biostatistics, School of Public Health, University of Alabama at Birmingham, Birmingham, AL, USA and <sup>3</sup>Genetic Analysis Platform, The Broad Institute Center for Genotyping and Analysis, Broad Institute of MIT and Harvard, Cambridge, MA, USA

The Type I Diabetes Genetics Consortium (T1DGC) Rapid Response Workshop was established to evaluate published candidate gene associations in a large collection of affected sib-pair (ASP) families. We report on our quality control (QC) and preliminary family-based association analyses. A random sample of blind duplicates was analyzed for QC. Quality checks, including examination of plate-panel yield, marker yield, Hardy–Weinberg equilibrium, mismatch error rate, Mendelian error rate, and allele distribution across plates, were performed. Genotypes from 2324 families within nine cohorts were obtained from a panel of 21 candidate genes, including 384 single-nucleotide polymorphisms on two genotyping platforms performed at the Broad Institute Center for Genotyping and Analysis (Cambridge, MA, USA). The T1DGC Rapid Response project, following rigorous QC procedures, resulted in a 2297 family, 9688 genotyped individual database on a single-candidate gene panel. The available data include 9005 individuals with genotype data from both platforms and 683 individuals genotyped (276 in Illumina; 407 in Sequenom) on only one platform.

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## Introduction

The Type I Diabetes Genetics Consortium (T1DGC; <http://www.t1dgc.org>) is an international effort to identify genes that determine an individual's risk of type I diabetes (T1D). The creation of a resource base of well-characterized affected sib-pair (ASP) families and other collections has been implemented that will facilitate the localization and characterization of T1D genes that determine disease risk. The aim of the T1DGC Rapid Response project was to explore candidate genes previously reported to be associated with T1D (for example *INS*,<sup>1</sup> *CTLA4*,<sup>2</sup> *PTPN22*,<sup>3</sup> *SUMO4*<sup>4</sup>). Confirmation of candidate genes for T1D has been difficult for some but not for others. Although many of the initial studies were underpowered, some genes appeared to exhibit their association only in certain populations or ethnic groups.

In the T1DGC Rapid Response experiment, a series of candidate genes were chosen for evaluation in a large collection of Caucasian ASP families. The candidate genes had been published earlier with identified polymorphisms that suggested association and may (or may not) have been replicated in other studies. In a separate component of the project, candidate genes that emerged from recent genome-wide association scans for T1D, type II diabetes, and other autoimmune diseases were

included for follow-up. This report provides the background of the quality control (QC) procedures in the first Rapid Response experiment, whose objectives were to (1) establish evidence of replication of published candidate genes and (2) test hypotheses of population heterogeneity, the impact of stratification, potential genetic interaction on risk, and other modifying factors on T1D risk.

## Results

The T1DGC Rapid Response project had three data-set releases. The second and third data-set releases resulted in modified, updated, and more complete versions of the earlier Rapid Response data. The main modifications for each release were incorporation of re-called markers and resolution of problematic families. As major histocompatibility complex (MHC) fine mapping and genome-wide linkage scans were being performed on the same families, these additional data provided more specific relationship information to help resolve pedigree structure problems. All datasets were securely maintained in the T1DGC web site (<http://www.t1dgc.org>) with password-protected access. These and other T1DGC datasets are now available ([https://www.t1dgc.org/views/vw\\_databases.cfm](https://www.t1dgc.org/views/vw_databases.cfm)).

The initial data release (2007.03.RR) consisted of 2317 families from 9 cohorts with genotype data on 21 candidate genes as well as a set of DNA fingerprinting markers. For this release, seven families were removed because of high levels of Mendelian inconsistencies. In

Correspondence: Dr WM Brown, Division of Public Health Sciences, Department of Biostatistics, Wake Forest University Health Sciences, Winston-Salem, NC 27157, USA.  
E-mail: wmbrown@wfbmc.edu

**Table 1** Description of T1DGC Rapid Response data following QC procedures

T1DGC 2008.07.RR Data characteristics Family summary: post-modification											
Contributor	Families in dataset	Families with genotype data	Families (n (%)) with								
			Parents			Affected full siblings					
			0	1	2	0	1	2	3	4+	Half sib
Asia-Pacific	191	191	27	53	111	1	11	168	8	1	2
BDA	417	417	6	71	340	6	63	347	1	0	0
Danish	146	146	17	41	88	1	18	115	12	0	0
Europe	475	475	51	114	310	0	30	420	22	1	2
HBDI	431	431	1	40	390	0	24	365	35	6	1
North America	334	334	38	103	193	0	28	293	10	0	3
United Kingdom	114	114	5	24	85	0	6	104	3	1	0
Joslin	112	111	38	24	49	1	12	90	5	0	3
Sardinian	78	78	4	22	52	0	8	70	0	0	0
All	2298	2297	187	492	1618	9	200	1972	96	9	11

Abbreviations: BDA, British Dietetic Association; HBDI, Human Biological Data Interchange; QC, quality control; T1DGC, Type I Diabetes Genetics Consortium.

addition, 135 individuals had either a pedigree change (that is classified new parent, sample switch, or gender reclassification) or were considered unrelated to other family members. The number of single-nucleotide polymorphisms (SNPs) genotyped per candidate gene ranged from 1 to 66. There were 357 SNPs released that were genotyped using the Illumina GoldenGate assay and 375 SNPs that were genotyped using the Sequenom platform; 334 SNPs were common to the two platforms. The Sequenom genotyping resulted in a slightly higher number of inconsistencies within families. Two SNPs were eliminated in the Illumina panel, as they resulted in problems in >20% of the families. Four SNPs were set to missing in the Sequenom panel that resulted in problems in >10% of the families. In addition, eight SNPs genotyped with the Illumina assay and two SNPs genotyped with the Sequenom assay were missing because of <80% call rate.

The second data release (2007.12.RR) were generated on 2297 families from 9 cohorts on 21 candidate genes and included DNA fingerprinting markers and re-genotyped data for seven SNPs on the Sequenom platform. These seven SNPs (*INS* rs1003483; *PTPN22* rs1746860; *IL13* rs1881457; *IL12B* rs2569253; *IL4R* rs3024613; *IL2RA* rs4147359; *VDR* rs7975232) had their data re-scored based on significant deviation from Hardy–Weinberg equilibrium assumptions. It was also determined that these seven SNPs had a low genotyping concordance compared with that observed on the Illumina panel. Thus, these SNPs were determined to have erroneously scored genotypes. The repeated scoring resulted in releasing 377 markers on the Sequenom platform and increasing the number to 336 SNPs that were scored on both platforms.

Earlier, the datasets that were released included all SNPs (regardless of genotyping status), three families

whose Mendelian inconsistencies were resolved, elimination of one member from a pair of monozygotic twins, and removal of 23 duplicate families. This dataset incorporated 137 individuals who had either a pedigree change (that is classified new parent, sample switch, or gender reclassification), were members of a family who were determined to be unrelated, or were a duplicate sample and, therefore, had their genotype dataset to 'missing.'

The third and final data release (2008.07.RR) consisted of 2297 families from 9 cohorts with genotype data on 21 candidate genes and a set of DNA fingerprinting markers (Table 1). For this release, the raw genotype data generated from the Sequenom platform were scored using the new Typer 4.0 software. A comparison of scoring 10 SNPs that were considered 'poor performing' markers and a second comparison of 20 SNPs using Typer 4.0 software suggested that well-called SNPs remained so, whereas poorly called SNPs improved when scored with the Typer 4.0 software. The Sequenom assay for *TCF7* rs5742913 was originally designed to detect C and T alleles in dbSNP using ss65832708 ([http://www.ncbi.nlm.nih.gov/SNP/snp\\_ref.cgi?rs=5742913](http://www.ncbi.nlm.nih.gov/SNP/snp_ref.cgi?rs=5742913)). This assay resulted in the SNP appearing monomorphic for the C allele; however, earlier data<sup>5,6</sup> reported that the correct alleles are C and A (not T) as defined by ss93257904. By virtue of the Sequenom iPLEX chemistry use of four mass-distinct chain-terminating nucleotides, a re-definition by the Typer 4.0 software to identify and measure the mass peak corresponding to the A-extended primer (earlier attempting to detect the absent T-extended primer) permitted the accurate re-call of genotypes. To provide consistent data, all SNPs in the Sequenom dataset were re-called using Typer 4.0 software. These data were then subjected to standard QC procedures. The Illumina data were unchanged from the earlier release (2007.12.RR).

## Discussion

The T1DGC Rapid Response project resulted in a dataset containing 9688 individuals from 2297 ASP families with family members genotyped for 21 T1D candidate genes on two genotype platforms. Five of the candidate genes were ‘fully investigated,’ in that the structure of the gene was evaluated using tagging SNPs. Sixteen of the genes were subjected to genotyping using only the reported SNPs that exhibited the strongest association with T1D (for more detail, see Julier *et al.*,<sup>7</sup> this volume). In this dataset, there are 9005 individuals with genotype data derived from both platforms. There are 683 individuals genotyped only on one platform (276 in Illumina; 407 in Sequenom).

There are two important aspects of the Rapid Response project. The first was that a majority of the families were included in a genome-wide linkage scan panel (6000 SNPs).<sup>8,9</sup> Second, all families were included in the MHC Fine Mapping project.<sup>10</sup> As a result of the additional genetic information, the T1DGC Coordinating Center was able to better resolve QC issues (such as questionable family structure) within the Rapid Response dataset. The T1DGC Rapid Response project permitted the creation of a valuable resource of well-characterized families from multiple geographic sites that showed the localization and characterization of T1D genes.

## Materials and methods

### Genotyping

The T1DGC Molecular Technology Subcommittee established a list of genes for investigation at the Rapid Response Laboratory (Broad Institute of Harvard/Massachusetts Institute of Technology). An initial set of published candidate genes and associated SNPs was selected, based on a literature search. Two lists of candidate genes were constructed for consideration in the experiment. List 1 included those genes (*INS*, *PTPN22*, *CTLA4*, *IL2R/CD25*, and *SUMO4*) that had been confirmed or replicated, excluding those in the MHC. List 2 included those genes (*IL12B*, *IL4R*, *IL4*, *IL13*, *OAS1*, *VDR*, *SDF1*, *PAX4*, *FOXP3*, *IRS1*, *TCF7*, *IFIH1*, *EFHB*, *CAPSL*, *Q7Z4C4(5Q)*, and *CEACAM21*) that had not been replicated earlier.

From the initial set of candidate genes, a panel of 384 SNPs was identified using the ‘Tagger’ algorithm<sup>11</sup> such that all observed SNPs in HapMap-CEU with minor allele frequency >3% were captured with a perfect proxy ( $r^2 = 1$ ) for genes on List 1, or with a tagging SNP at  $r^2 \geq 0.8$  for genes on List 2. SNPs were preferentially chosen with high-design scores for the Illumina GoldenGate assay.

The panel of 384 SNPs was genotyped on two platforms, using the Illumina GoldenGate assay and the Sequenom MALDI-TOF assay. A preliminary round of design and assay testing using the HapMap CEU sample panel was performed on both platforms. For genotyping using the Sequenom platform, 19 SNPs were not included because of an inability to design robust assays. The 19 excluded SNPs were replaced with 16 SNPs potentially useful for sample tracking for Affymetrix whole-genome genotyping. The 381 SNPs genotyped using the Sequenom iPLEX technology were distributed among 20 iPLEX pools (13–21 SNPs/pool; median pool

size 19). The SNPs were part of 21 candidate genes: 5 fully investigated (158 SNPs) and 16 replicated (226 SNPs). The number of SNPs genotyped for each gene ranged from 1 to 69 SNPs. Four genes contained a single SNP; 3 genes contained 5–6 SNPs; 7 genes contained 10–16 SNPs; 4 genes contained 21–28 SNPs; 3 genes contained 37–42 SNPs; and one gene contained 69 SNPs.

In the Illumina assay, 367 SNPs were adequately genotyped, whereas the remaining 17 SNPs could not be accurately called and were considered ‘missing.’ For the Sequenom assay, all 381 SNPs were genotyped. For the genotyped SNPs, 8 in the Illumina panel and 2 in the Sequenom panel had <80% call rate and were set to ‘missing.’ Using strand information, genotypes were examined and calls were adjusted on the Sequenom panel to reflect the strand order of the Illumina panel. This procedure required ‘flipping’ genotype calls.

For the original release of the Rapid Response data, the Sequenom platform used Typer 3.0 software (<http://www.sequenom.com> (software no longer available)) to call genotypes. The Sequenom genotype data were later re-called using Typer 4.0 software (<http://sequenom.com/Genetic-Analysis/Applications/iPLEX-Genotyping/iPLEX-Literature>). This version of the Sequenom genotype calling software allows the simultaneous viewing and consistent clustering of multiple production plates of data, ensuring that genotype class definitions are stable across plates. (For a complete list of SNPs, see Appendix A)

### Samples

DNA samples from members of 2324 ASP families obtained from nine cohorts were used for genotyping. The families selected consisted primarily of nuclear families with an ASP with T1D. A total of 9982 DNA samples were shipped. A total of 9985 samples were used in the genotyping on both platforms: 9982 production samples (including 339 QC duplicate samples) and 3 CEPH control samples. For the Illumina platform, 9479 of the production samples and 322 QC samples genotyped with >90% call rate on 367 SNPs. For the Sequenom platform, 9581 production samples and 315 QC samples genotyped at >90% call rate on 380 SNPs.

The initial QC procedure of the Coordinating Center consisted of reviewing the failed status of the SNPs and samples, based on reports from the genotyping facility. Using the production and duplicate QC samples, concordance rates were generated between the pairs. This rate was based on both samples having a called genotype for a given SNP. The total number of concordant SNPs was divided by the total number of SNPs where both samples had a called genotype. ‘Missingness’ was also examined between the two samples. Samples that were discordant (that is concordance rate <96%) were reviewed within families to identify the sample with Mendelian consistency. For concordant samples, the sample that had the greatest number of called genotypes was preserved for analysis. If a production sample or QC sample failed genotyping, the sample that passed evaluation was preserved for analysis.

Genotypes from each of the two marker panels (Illumina and Sequenom) were reviewed initially as separate datasets. The results of each QC procedure were then compared across both analyses to detect similar (or different) problems. In each genotyping platform, there

were 339 QC samples. In both panels, two (different) samples failed for both the production and QC genotyping. For the Illumina panel, nine samples failed the production sample but passed for the QC sample. For the Sequenom panel, 16 samples failed QC genotyping but passed for the production sample. Four failed samples were common to the two platforms. For the Illumina panel, 15 samples failed QC genotyping but passed the production sample. For the Sequenom panel, 21 samples failed QC genotyping but passed for the production sample.

The overall concordance rate in the Illumina platform between the production sample and QC sample was 98.6%. There were 10 samples with concordance <96%. Only one of these 10 samples had >80% of total SNPs genotyped. In the Sequenom platform, there were seven samples with concordance rate <96%. Three of these samples had >80% of total SNPs genotyped. Once the production and QC sample concordance and comparison estimates were made, family structures and genotypic data were used to check for Mendelian inconsistency, relationship misclassification, and existence of duplicate samples.

Families were examined for Mendelian inconsistencies to detect relationship misclassification using the PedCheck<sup>12</sup> software. After summarizing PedCheck results, the Coordinating Center enumerated 'Mendelian Inconsistency Errors' (MIE) within each family. If the total number of inconsistencies was >2% of the total number of SNPs, the family was considered to be problematic and individually reviewed. From these in-depth reviews, pedigrees were re-arranged, restructured, or individuals coded as missing all SNP genotypes. Results from both genotyping datasets were reviewed. As part of this QC procedure, individual SNPs were independently checked for excess MIE. If an SNP had MIE in >20% of families, the SNP was considered to be problematic and the entire set of SNP genotypes was coded as missing. SNPs with MIE counts of 10–20% were reviewed on an individual basis. To aid in detection of family structure problems for the MHC data, we used the PREST<sup>13</sup> results from the T1DGC genome-wide linkage scans.<sup>6,8</sup>

The Coordinating Center reviewed pairwise comparisons within families and between families to detect potential duplicate samples. The identical by state (IBS) statistics for pairwise individuals were obtained using Graphical Relationship Representation software.<sup>14</sup> For pairs of relatives that had IBS>1.98, the data were reviewed to determine whether they were within families (that is twins or duplicate samples) or between families (that is same person belonging to two distinct families, multiple individuals common between two distinct families, or duplicate sample between families). In combination with the IBS information, MIE results were used to determine whether a sample switch or a duplicate sample had occurred. The Coordinating Center examined individuals and families across all genotyping datasets released earlier. Using these data, the Coordinating Center decided whether there were sample switches, relatedness issues, gender discrepancies, or duplicate samples (that is within families, across families, or twins). After all issues were resolved, datasets were assembled for final MIE checks and families were re-examined. Families that continued to exhibit high-MIE rates were removed from the dataset.

**Table 2** Description of the T1DGC Rapid Response genetic data

T1DGC 2008.07.RR Pedigree summary Data characteristics and preparation	DNA		Filtered LMN		Filtered SQNM		Pedigree changes				Modifications				Genotypes dropped	
	Source	DNA	Filtered LMN	Filtered SQNM	New relationship	Added members	Samples switched	Sex difference	Ungenotyped	Problematic families removed	Duplicate families removed	Within family duplicates removed	Valid marker count LMN	Valid marker count SQNM		
Asia-Pacific	782	763 (97.6)	756 (96.7)		2	3	1	0	3	0	0	0	357	376		
BDA	1808	1668 (92.3)	1646 (91.0)		5	2	2	3	50	3	5	1	357	377		
Danish	686	678 (98.8)	636 (92.7)		4	1	1	0	6	1	0	0	357	377		
Europe	1953	1941 (99.4)	1898 (97.2)		2	2	3	0	11	0	0	4	357	377		
HBDI	2143	1959 (91.4)	2042 (95.3)		1	1	3	0	7	0	7	2	357	377		
North America	1376	1323 (96.1)	1301 (94.5)		1	1	4	1	8	0	4	4	357	377		
United Kingdom	484	442 (91.3)	459 (94.8)		0	0	0	2	8	0	2	6	357	377		
Joslin	386	368 (95.3)	335 (86.8)		2	2	1	0	4	1	5	0	357	377		
Sardinian	364	337 (92.6)	339 (93.1)		0	0	0	0	8	0	0	1	357	377		
Total	9982	9479 (95.0)	9412 (94.3)		17	12	15	6	105	5	23	18				

Abbreviations: BDA, British Diabetic Association; HBDI, Human Biological Data Interchange; T1DGC, Type 1 Diabetes Genetics Consortium.



The remaining families were deemed to have random MIE. A family that was deemed problematic and a reasonable solution was not available was completely removed from the analysis dataset. Once families were considered 'clean,' the family was included the final analysis data file (Table 2).

#### Summary

The Coordinating Center for the T1DGC performed QC and initial family-based association analyses on data from two genotyping platforms (Illumina Golden Gate and Sequenom iPLEX) for the T1DGC Rapid Response project. A random sample of blind duplicates was evaluated for QC. DNA samples collected from participants were shipped to the genotyping laboratory from several T1DGC DNA Repository sites. Quality checks, including examination of plate-panel yield, marker yield, Hardy-Weinberg equilibrium, mismatch error rate, Mendelian error rate, and allele distribution across plates, were performed. Genotypes from 2324 families within nine cohorts were genotyped for 21 candidate genes (384 SNPs). The final data consisted of genotypes from 2297 families (9688 individuals) that enabled robust estimation of candidate gene effects on T1D risk.

#### Conflict of interest

The authors declare no conflict of interest.

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## Appendix

## Appendix A: Rapid Response reference SNP list

<i>Study</i>	<i>Chromosome</i>	<i>Gene</i>	<i>Marker</i>	<i>Physical position</i>	<i>Illumina</i>	<i>Sequenom</i>
Full	1	<i>PTPN22</i>	rs3827733	114050631	Genotyped	Genotyped
Full	1	<i>PTPN22</i>	rs3789602	114051793	Genotyped	Genotyped
Full	1	<i>PTPN22</i>	rs1217379	114056125	Genotyped	Genotyped
Full	1	<i>PTPN22</i>	rs6537798	114063748	Genotyped	Genotyped
Full	1	<i>PTPN22</i>	rs3789607	114078476	Genotyped	Genotyped
Full	1	<i>PTPN22</i>	rs2476600	114081776	Genotyped	Genotyped
Full	1	<i>PTPN22</i>	rs1217395	114086477	Genotyped	Ungenotyped
Full	1	<i>PTPN22</i>	rs1970559	114089190	Genotyped	Ungenotyped
Full	1	<i>PTPN22</i>	rs2476601	114089610	Genotyped	Genotyped
Full	1	<i>PTPN22</i>	rs1775759	114100846	Genotyped	Genotyped
Full	1	<i>PTPN22</i>	rs3765598	114106505	Genotyped	Genotyped
Full	1	<i>PTPN22</i>	rs11582409	114111475	Ungenotyped	Genotyped
Full	1	<i>PTPN22</i>	rs1217418	114113273	Genotyped	Genotyped
Full	1	<i>PTPN22</i>	rs1217414	114124709	Genotyped	Genotyped
Full	1	<i>PTPN22</i>	rs17510162	114125773	Genotyped	Genotyped
Full	1	<i>PTPN22</i>	rs2488457	114127410	Genotyped	Genotyped
Full	1	<i>PTPN22</i>	rs1235005	114129479	Genotyped	Genotyped
Full	1	<i>PTPN22</i>	rs6665194	114129885	Genotyped	Genotyped
Full	1	<i>PTPN22</i>	rs1217384	114131802	Genotyped	Genotyped
Full	1	<i>PTPN22</i>	rs12566340	114132370	Genotyped	Genotyped
Full	1	<i>PTPN22</i>	rs7529353	114132504	Genotyped	Genotyped
Full	1	<i>PTPN22</i>	rs7524200	114138866	Genotyped	Genotyped
Full	1	<i>PTPN22</i>	rs1217423	114139335	Genotyped	Genotyped
Full	1	<i>PTPN22</i>	rs1746860	114141382	Ungenotyped	Genotyped
Full	1	<i>PTPN22</i>	rs2358994	114141503	Genotyped	Genotyped
Full	1	<i>PTPN22</i>	rs1539438	114142398	Genotyped	Genotyped
Full	1	<i>PTPN22</i>	rs1217394	114145701	Genotyped	Genotyped
Full	1	<i>PTPN22</i>	rs1217393	114145988	Genotyped	Ungenotyped
Full	2	<i>CTLA4</i>	rs231811	204539397	Genotyped	Genotyped
Full	2	<i>CTLA4</i>	rs6741283	204540316	Genotyped	Genotyped
Full	2	<i>CTLA4</i>	rs11571293	204543219	Genotyped	Genotyped
Full	2	<i>CTLA4</i>	rs2162610	204544235	Genotyped	Genotyped
Full	2	<i>CTLA4</i>	rs926169	204548258	Genotyped	Genotyped
Full	2	<i>CTLA4</i>	rs11571290	204548647	Genotyped	Genotyped
Full	2	<i>CTLA4</i>	rs231770	204554659	Genotyped	Genotyped
Full	2	<i>CTLA4</i>	rs733618	204556450	Genotyped	Genotyped
Full	2	<i>CTLA4</i>	rs11571316	204556595	Genotyped	Genotyped
Full	2	<i>CTLA4</i>	rs16840252	204557025	Genotyped	Genotyped
Full	2	<i>CTLA4</i>	rs11571317	204557514	Genotyped	Genotyped
Full	2	<i>CTLA4</i>	rs5742909	204557853	Genotyped	Genotyped
Full	2	<i>CTLA4</i>	rs231777	204559094	Genotyped	Genotyped
Full	2	<i>CTLA4</i>	rs231779	204559993	Genotyped	Genotyped
Full	2	<i>CTLA4</i>	rs3087243	204564425	Genotyped	Ungenotyped
Full	2	<i>CTLA4</i>	rs1427676	204566672	Genotyped	Genotyped
Full	2	<i>CTLA4</i>	rs231727	204567056	Genotyped	Genotyped
Full	2	<i>CTLA4</i>	rs231731	204570036	Genotyped	Genotyped
Full	2	<i>CTLA4</i>	rs11571300	204572273	Genotyped	Genotyped
Full	2	<i>CTLA4</i>	rs960792	204574756	Ungenotyped	Ungenotyped
Full	2	<i>CTLA4</i>	rs1365965	204577376	Genotyped	Genotyped
Full	2	<i>CTLA4</i>	rs231757	204578993	Genotyped	Genotyped
Full	2	<i>CTLA4</i>	rs231755	204579075	Genotyped	Genotyped
Full	2	<i>CTLA4</i>	rs7600322	204579859	Genotyped	Ungenotyped
Full	2	<i>CTLA4</i>	rs6748358	204582411	Genotyped	Genotyped
Full	6	<i>SUMO4</i>	rs12204461	149743228	Genotyped	Genotyped
Full	6	<i>SUMO4</i>	rs7742990	149746219	Genotyped	Genotyped
Full	6	<i>SUMO4</i>	rs9373589	149748981	Genotyped	Genotyped
Full	6	<i>SUMO4</i>	rs9404034	149753676	Genotyped	Genotyped
Full	6	<i>SUMO4</i>	rs2789490	149756012	Genotyped	Genotyped
Full	6	<i>SUMO4</i>	rs237032	149757865	Genotyped	Genotyped
Full	6	<i>SUMO4</i>	rs237025	149763383	Genotyped	Genotyped
Full	6	<i>SUMO4</i>	rs2789488	149766983	Genotyped	Genotyped
Full	6	<i>SUMO4</i>	rs2789489	149769038	Genotyped	Genotyped
Full	6	<i>SUMO4</i>	rs652921	149772539	Genotyped	Genotyped
Full	6	<i>SUMO4</i>	rs366905	149776790	Genotyped	Genotyped
Full	6	<i>SUMO4</i>	rs480034	149777123	Genotyped	Genotyped
Full	6	<i>SUMO4</i>	rs236999	149781068	Genotyped	Genotyped
Full	6	<i>SUMO4</i>	rs513923	149782364	Genotyped	Genotyped

Appendix A Continued

<i>Study</i>	<i>Chromosome</i>	<i>Gene</i>	<i>Marker</i>	<i>Physical position</i>	<i>Illumina</i>	<i>Sequenom</i>
Full	6	SUMO4	rs9485389	149782907	Genotyped	Genotyped
Full	10	IL2R/CD25	rs6602363	6076150	Genotyped	Genotyped
Full	10	IL2R/CD25	rs7898880	6077559	Genotyped	Genotyped
Full	10	IL2R/CD25	rs7911500	6077732	Genotyped	Genotyped
Full	10	IL2R/CD25	rs17147986	6078484	Ungenotyped	Genotyped
Full	10	IL2R/CD25	rs6602364	6078859	Genotyped	Genotyped
Full	10	IL2R/CD25	rs1323653	6079064	Genotyped	Genotyped
Full	10	IL2R/CD25	rs7477011	6080600	Genotyped	Genotyped
Full	10	IL2R/CD25	rs10795731	6082040	Genotyped	Genotyped
Full	10	IL2R/CD25	rs17322780	6082478	Genotyped	Genotyped
Full	10	IL2R/CD25	rs10795733	6083484	Genotyped	Genotyped
Full	10	IL2R/CD25	rs764851	6087950	Genotyped	Genotyped
Full	10	IL2R/CD25	rs10795737	6089350	Genotyped	Genotyped
Full	10	IL2R/CD25	rs12359875	6091113	Genotyped	Genotyped
Full	10	IL2R/CD25	rs12722608	6092847	Genotyped	Genotyped
Full	10	IL2R/CD25	rs12722605	6093169	Genotyped	Genotyped
Full	10	IL2R/CD25	rs12244380	6093380	Genotyped	Genotyped
Full	10	IL2R/CD25	rs12722598	6095156	Genotyped	Genotyped
Full	10	IL2R/CD25	rs9663421	6095610	Genotyped	Genotyped
Full	10	IL2R/CD25	rs12722596	6096300	Genotyped	Genotyped
Full	10	IL2R/CD25	rs2386841	6097738	Genotyped	Genotyped
Full	10	IL2R/CD25	rs7899538	6099904	Genotyped	Genotyped
Full	10	IL2R/CD25	rs12722588	6100439	Genotyped	Genotyped
Full	10	IL2R/CD25	rs2274037	6102114	Genotyped	Genotyped
Full	10	IL2R/CD25	rs2076846	6103259	Genotyped	Genotyped
Full	10	IL2R/CD25	rs7093069	6103325	Genotyped	Genotyped
Full	10	IL2R/CD25	rs11596355	6104187	Genotyped	Genotyped
Full	10	IL2R/CD25	rs12722574	6106468	Genotyped	Genotyped
Full	10	IL2R/CD25	rs2031229	6106574	Genotyped	Ungenotyped
Full	10	IL2R/CD25	rs2025345	6107694	Genotyped	Genotyped
Full	10	IL2R/CD25	rs12722561	6109899	Genotyped	Genotyped
Full	10	IL2R/CD25	rs7910961	6117802	Genotyped	Genotyped
Full	10	IL2R/CD25	rs6602391	6118038	Genotyped	Genotyped
Full	10	IL2R/CD25	rs12722521	6118802	Genotyped	Ungenotyped
Full	10	IL2R/CD25	rs11256448	6119485	Ungenotyped	Genotyped
Full	10	IL2R/CD25	rs7072398	6119852	Genotyped	Genotyped
Full	10	IL2R/CD25	rs12722518	6120643	Genotyped	Genotyped
Full	10	IL2R/CD25	rs11256456	6120718	Genotyped	Genotyped
Full	10	IL2R/CD25	rs12722516	6121223	Genotyped	Genotyped
Full	10	IL2R/CD25	rs4749924	6122402	Genotyped	Genotyped
Full	10	IL2R/CD25	rs6602398	6122959	Genotyped	Genotyped
Full	10	IL2R/CD25	rs4749926	6125318	Genotyped	Genotyped
Full	10	IL2R/CD25	rs10905656	6126099	Genotyped	Genotyped
Full	10	IL2R/CD25	rs942201	6126298	Genotyped	Genotyped
Full	10	IL2R/CD25	rs706780	6127032	Genotyped	Genotyped
Full	10	IL2R/CD25	rs1107345	6127301	Genotyped	Genotyped
Full	10	IL2R/CD25	rs11256497	6127800	Genotyped	Genotyped
Full	10	IL2R/CD25	rs791587	6128705	Genotyped	Genotyped
Full	10	IL2R/CD25	rs791589	6129577	Genotyped	Ungenotyped
Full	10	IL2R/CD25	rs791590	6130328	Genotyped	Genotyped
Full	10	IL2R/CD25	rs10905669	6132099	Genotyped	Genotyped
Full	10	IL2R/CD25	rs1323658	6134360	Genotyped	Genotyped
Full	10	IL2R/CD25	rs2476491	6135416	Genotyped	Genotyped
Full	10	IL2R/CD25	rs2256774	6137171	Genotyped	Genotyped
Full	10	IL2R/CD25	rs706779	6138830	Genotyped	Genotyped
Full	10	IL2R/CD25	rs706778	6138955	Genotyped	Genotyped
Full	10	IL2R/CD25	rs2104286	6139051	Genotyped	Ungenotyped
Full	10	IL2R/CD25	rs3134883	6140731	Genotyped	Genotyped
Full	10	IL2R/CD25	rs3118470	6141719	Genotyped	Genotyped
Full	10	IL2R/CD25	rs12722486	6143768	Genotyped	Genotyped
Full	10	IL2R/CD25	rs7072793	6146272	Genotyped	Genotyped
Full	10	IL2R/CD25	rs7073236	6146558	Genotyped	Genotyped
Full	10	IL2R/CD25	rs4147359	6148445	Ungenotyped	Genotyped
Full	10	IL2R/CD25	rs7089861	6150332	Genotyped	Genotyped
Full	10	IL2R/CD25	rs7090512	6150835	Genotyped	Genotyped
Full	10	IL2R/CD25	rs1887027	6153788	Genotyped	Genotyped
Full	10	IL2R/CD25	rs4749955	6158972	Genotyped	Genotyped
Full	10	IL2R/CD25	rs11594656	6162015	Genotyped	Genotyped
Full	10	IL2R/CD25	rs12251307	6163501	Genotyped	Genotyped
Full	10	IL2R/CD25	rs7100400	6164086	Genotyped	Genotyped
Full	11	INS	rs4244808	2119686	Genotyped	Ungenotyped

## Appendix A Continued

<i>Study</i>	<i>Chromosome</i>	<i>Gene</i>	<i>Marker</i>	<i>Physical position</i>	<i>Illumina</i>	<i>Sequenom</i>
Full	11	<i>INS</i>	rs1003483	2124119	Genotyped	Genotyped
Full	11	<i>INS</i>	rs3741208	2126350	Ungenotyped	Genotyped
Full	11	<i>INS</i>	rs1004446	2126719	Genotyped	Genotyped
Full	11	<i>INS</i>	rs4320932	2128177	Genotyped	Genotyped
Full	11	<i>INS</i>	rs7924316	2130023	Genotyped	Genotyped
Full	11	<i>INS</i>	rs3842753	2137636	Ungenotyped	Genotyped
Full	11	<i>INS</i>	rs3842748	2137971	Genotyped	Genotyped
Full	11	<i>INS</i>	rs2070762	2142911	Genotyped	Genotyped
Full	11	<i>INS</i>	rs6356	2147527	Genotyped	Genotyped
Full	11	<i>INS</i>	rs10840490	2150393	Ungenotyped	Genotyped
Full	11	<i>INS</i>	rs10743149	2150751	Genotyped	Genotyped
Full	11	<i>INS</i>	rs10840491	2150966	Genotyped	Ungenotyped
Full	11	<i>INS</i>	rs7119275	2151386	Genotyped	Genotyped
Full	11	<i>INS</i>	rs10840495	2152413	Genotyped	Ungenotyped
Full	11	<i>INS</i>	rs4930046	2153724	Genotyped	Ungenotyped
Full	11	<i>INS</i>	rs4929966	2154012	Genotyped	Genotyped
Full	11	<i>INS</i>	rs11042978	2154994	Genotyped	Genotyped
Full	11	<i>INS</i>	rs11564710	2156905	Genotyped	Genotyped
Full	11	<i>INS</i>	rs6578993	2157739	Genotyped	Genotyped
Full	11	<i>INS</i>	rs11564709	2157914	Genotyped	Genotyped
Replication	2	<i>IFIH1</i>	rs2111485	162936043	Genotyped	Genotyped
Replication	2	<i>IFIH1</i>	rs1990760	162949558	Genotyped	Genotyped
Replication	2	<i>IFIH1</i>	rs3747517	162954331	Genotyped	Genotyped
Replication	2	<i>IFIH1</i>	rs3788964	163032987	Genotyped	Genotyped
Replication	2	<i>IFIH1</i>	rs984971	163050028	Genotyped	Genotyped
Replication	2	<i>IFIH1</i>	rs2068330	163062897	Genotyped	Genotyped
Replication	2	<i>IRS1</i>	rs16822551	227407085	Genotyped	Genotyped
Replication	2	<i>IRS1</i>	rs11683087	227412111	Genotyped	Genotyped
Replication	2	<i>IRS1</i>	rs2251692	227415285	Genotyped	Ungenotyped
Replication	2	<i>IRS1</i>	rs17208239	227423202	Genotyped	Genotyped
Replication	2	<i>IRS1</i>	rs17208470	227429410	Genotyped	Genotyped
Replication	2	<i>IRS1</i>	rs2435185	227461928	Genotyped	Genotyped
Replication	2	<i>IRS1</i>	rs4675095	227477472	Genotyped	Genotyped
Replication	2	<i>IRS1</i>	rs1801278	227486049	Genotyped	Genotyped
Replication	2	<i>IRS1</i>	rs1801123	227486548	Genotyped	Genotyped
Replication	2	<i>IRS1</i>	rs6725330	227492362	Genotyped	Genotyped
Replication	2	<i>IRS1</i>	rs6725556	227492497	Genotyped	Genotyped
Replication	2	<i>IRS1</i>	rs13018009	227493906	Genotyped	Genotyped
Replication	2	<i>IRS1</i>	rs4675096	227494446	Genotyped	Genotyped
Replication	2	<i>IRS1</i>	rs956115	227500069	Genotyped	Ungenotyped
Replication	2	<i>IRS1</i>	rs957797	227502248	Genotyped	Genotyped
Replication	2	<i>IRS1</i>	rs13417106	227509333	Genotyped	Genotyped
Replication	3	<i>EFHB</i>	rs2929366	19934760	Genotyped	Genotyped
Replication	5	<i>CAPSL</i>	rs1445898	35946286	Genotyped	Genotyped
Replication	5	<i>IL12B</i>	rs7730126	158662525	Genotyped	Genotyped
Replication	5	<i>IL12B</i>	rs1549922	158664126	Genotyped	Genotyped
Replication	5	<i>IL12B</i>	rs4921466	158665350	Genotyped	Genotyped
Replication	5	<i>IL12B</i>	rs6859018	158669570	Genotyped	Genotyped
Replication	5	<i>IL12B</i>	rs17875325	158673108	Genotyped	Genotyped
Replication	5	<i>IL12B</i>	rs3181225	158673201	Genotyped	Genotyped
Replication	5	<i>IL12B</i>	rs17875324	158673428	Ungenotyped	Genotyped
Replication	5	<i>IL12B</i>	rs3212227	158675528	Genotyped	Genotyped
Replication	5	<i>IL12B</i>	rs2853696	158677238	Genotyped	Genotyped
Replication	5	<i>IL12B</i>	rs919766	158680142	Genotyped	Genotyped
Replication	5	<i>IL12B</i>	rs2853694	158681666	Genotyped	Genotyped
Replication	5	<i>IL12B</i>	rs2569253	158683571	Genotyped	Ungenotyped
Replication	5	<i>IL12B</i>	rs3181219	158684717	Genotyped	Genotyped
Replication	5	<i>IL12B</i>	rs17875303	158685556	Genotyped	Genotyped
Replication	5	<i>IL12B</i>	rs1433048	158688423	Genotyped	Genotyped
Replication	5	<i>IL12B</i>	rs2546893	158688538	Genotyped	Genotyped
Replication	5	<i>IL12B</i>	rs2546890	158692478	Genotyped	Genotyped
Replication	5	<i>IL12B</i>	rs10052709	158693055	Genotyped	Genotyped
Replication	5	<i>IL12B</i>	rs7709212	158696755	Genotyped	Genotyped
Replication	5	<i>IL12B</i>	rs6868898	158696998	Genotyped	Genotyped
Replication	5	<i>IL12B</i>	rs17056704	158700244	Genotyped	Genotyped
Replication	5	<i>IL12B</i>	rs13188370	158701707	Genotyped	Genotyped
Replication	5	<i>IL12B</i>	rs17056705	158701831	Genotyped	Genotyped
Replication	5	<i>IL12B</i>	rs17056706	158703333	Genotyped	Genotyped
Replication	5	<i>IL12B</i>	rs953861	158705160	Genotyped	Genotyped
Replication	5	<i>IL12B</i>	rs4244437	158705695	Genotyped	Genotyped
Replication	5	<i>IL12B</i>	rs11747112	158707187	Genotyped	Genotyped



Appendix A Continued

<i>Study</i>	<i>Chromosome</i>	<i>Gene</i>	<i>Marker</i>	<i>Physical position</i>	<i>Illumina</i>	<i>Sequenom</i>
Replication	5	<i>IL13</i>	rs2240032	132005026	Ungenotyped	Genotyped
Replication	5	<i>IL13</i>	rs1881457	132020308	Ungenotyped	Genotyped
Replication	5	<i>IL13</i>	rs1800925	132020708	Genotyped	Genotyped
Replication	5	<i>IL13</i>	rs1295686	132023742	Genotyped	Genotyped
Replication	5	<i>IL13</i>	rs20541	132023863	Genotyped	Genotyped
Replication	5	<i>IL4</i>	rs848	132024399	Genotyped	Genotyped
Replication	5	<i>IL4</i>	rs1295683	132026775	Genotyped	Genotyped
Replication	5	<i>IL4</i>	rs2243210	132029285	Genotyped	Genotyped
Replication	5	<i>IL4</i>	rs2243248	132036543	Genotyped	Genotyped
Replication	5	<i>IL4</i>	rs2243250	132037053	Genotyped	Genotyped
Replication	5	<i>IL4</i>	rs2227284	132040624	Genotyped	Genotyped
Replication	5	<i>IL4</i>	rs2243263	132041198	Genotyped	Genotyped
Replication	5	<i>IL4</i>	rs2243274	132042731	Genotyped	Genotyped
Replication	5	<i>IL4</i>	rs11242122	132052607	Genotyped	Genotyped
Replication	5	<i>IL4</i>	rs1468216	132064151	Genotyped	Genotyped
Replication	5	<i>Q7Z4C4(5Q)</i>	rs9127	96397921	Genotyped	Genotyped
Replication	5	<i>TCF7</i>	rs30503	133465245	Genotyped	Genotyped
Replication	5	<i>TCF7</i>	rs30500	133467775	Genotyped	Genotyped
Replication	5	<i>TCF7</i>	rs30499	133469625	Genotyped	Genotyped
Replication	5	<i>TCF7</i>	rs244948	133472960	Genotyped	Genotyped
Replication	5	<i>TCF7</i>	rs5742913	133479582	Ungenotyped	Genotyped
Replication	5	<i>TCF7</i>	rs244692	133480434	Genotyped	Genotyped
Replication	5	<i>TCF7</i>	rs151822	133483860	Genotyped	Genotyped
Replication	5	<i>TCF7</i>	rs152404	133486613	Genotyped	Genotyped
Replication	5	<i>TCF7</i>	rs152406	133489485	Genotyped	Genotyped
Replication	5	<i>TCF7</i>	rs249611	133492556	Ungenotyped	Genotyped
Replication	5	<i>TCF7</i>	rs17653687	133495899	Genotyped	Genotyped
Replication	7	<i>PAX4</i>	rs3779536	126827928	Genotyped	Genotyped
Replication	7	<i>PAX4</i>	rs806213	126828941	Genotyped	Genotyped
Replication	7	<i>PAX4</i>	rs3735640	126830289	Genotyped	Genotyped
Replication	7	<i>PAX4</i>	rs806216	126831813	Genotyped	Genotyped
Replication	7	<i>PAX4</i>	rs3824006	126835931	Genotyped	Genotyped
Replication	7	<i>PAX4</i>	rs10229583	126840854	Genotyped	Genotyped
Replication	7	<i>PAX4</i>	rs712701	126845139	Genotyped	Genotyped
Replication	7	<i>PAX4</i>	rs327518	126845743	Genotyped	Genotyped
Replication	7	<i>PAX4</i>	rs7801118	126857837	Genotyped	Genotyped
Replication	7	<i>PAX4</i>	rs806187	126863590	Genotyped	Genotyped
Replication	10	<i>CXCL12</i>	rs731336	44159260	Genotyped	Genotyped
Replication	10	<i>CXCL12</i>	rs1147882	44166437	Genotyped	Genotyped
Replication	10	<i>CXCL12</i>	rs266109	44170430	Genotyped	Genotyped
Replication	10	<i>CXCL12</i>	rs1147879	44170656	Ungenotyped	Genotyped
Replication	10	<i>CXCL12</i>	rs266108	44172126	Genotyped	Genotyped
Replication	10	<i>CXCL12</i>	rs266105	44175669	Genotyped	Genotyped
Replication	10	<i>CXCL12</i>	rs11595588	44175746	Genotyped	Genotyped
Replication	10	<i>CXCL12</i>	rs17391002	44175933	Ungenotyped	Genotyped
Replication	10	<i>CXCL12</i>	rs4948876	44176752	Genotyped	Genotyped
Replication	10	<i>CXCL12</i>	rs11239011	44176969	Genotyped	Genotyped
Replication	10	<i>CXCL12</i>	rs185545	44178846	Genotyped	Genotyped
Replication	10	<i>CXCL12</i>	rs17156191	44179353	Genotyped	Genotyped
Replication	10	<i>CXCL12</i>	rs266098	44181674	Genotyped	Ungenotyped
Replication	10	<i>CXCL12</i>	rs10900029	44182226	Genotyped	Genotyped
Replication	10	<i>CXCL12</i>	rs266093	44186214	Genotyped	Genotyped
Replication	10	<i>CXCL12</i>	rs266092	44186281	Genotyped	Genotyped
Replication	10	<i>CXCL12</i>	rs1801157	44188263	Genotyped	Genotyped
Replication	10	<i>CXCL12</i>	rs197452	44190246	Ungenotyped	Genotyped
Replication	10	<i>CXCL12</i>	rs266087	44191068	Ungenotyped	Genotyped
Replication	10	<i>CXCL12</i>	rs2297630	44191554	Genotyped	Genotyped
Replication	10	<i>CXCL12</i>	rs2839693	44194573	Genotyped	Genotyped
Replication	10	<i>CXCL12</i>	rs4948878	44194827	Genotyped	Genotyped
Replication	10	<i>CXCL12</i>	rs2839689	44195465	Genotyped	Genotyped
Replication	10	<i>CXCL12</i>	rs3780891	44198719	Genotyped	Genotyped
Replication	10	<i>CXCL12</i>	rs2839685	44201644	Ungenotyped	Genotyped
Replication	10	<i>CXCL12</i>	rs1413519	44202567	Genotyped	Genotyped
Replication	10	<i>CXCL12</i>	rs2861442	44204965	Genotyped	Genotyped
Replication	10	<i>CXCL12</i>	rs754617	44206348	Genotyped	Genotyped
Replication	10	<i>CXCL12</i>	rs7088285	44207935	Genotyped	Genotyped
Replication	10	<i>CXCL12</i>	rs6593412	44208136	Genotyped	Genotyped
Replication	10	<i>CXCL12</i>	rs1855531	44209658	Genotyped	Genotyped
Replication	10	<i>CXCL12</i>	rs11595460	44211083	Genotyped	Genotyped
Replication	10	<i>CXCL12</i>	rs1023264	44213762	Genotyped	Genotyped
Replication	10	<i>CXCL12</i>	rs1023262	44213870	Genotyped	Genotyped

## Appendix A Continued

<i>Study</i>	<i>Chromosome</i>	<i>Gene</i>	<i>Marker</i>	<i>Physical position</i>	<i>Illumina</i>	<i>Sequenom</i>
Replication	10	<i>CXCL12</i>	rs1779384	44214449	Genotyped	Genotyped
Replication	10	<i>CXCL12</i>	rs4948881	44216041	Genotyped	Genotyped
Replication	10	<i>CXCL12</i>	rs1761325	44216995	Genotyped	Genotyped
Replication	12	<i>OAS1</i>	rs3741982	111791701	Genotyped	Genotyped
Replication	12	<i>OAS1</i>	rs12177	111798145	Genotyped	Genotyped
Replication	12	<i>OAS1</i>	rs2240193	111798381	Genotyped	Genotyped
Replication	12	<i>OAS1</i>	rs2240191	111798451	Genotyped	Genotyped
Replication	12	<i>OAS1</i>	rs12309946	111798975	Genotyped	Genotyped
Replication	12	<i>OAS1</i>	rs4766662	111808419	Genotyped	Genotyped
Replication	12	<i>OAS1</i>	rs3741981	111811590	Genotyped	Genotyped
Replication	12	<i>OAS1</i>	rs10774671	111819913	Genotyped	Genotyped
Replication	12	<i>OAS1</i>	rs7135579	111829602	Genotyped	Genotyped
Replication	12	<i>OAS1</i>	rs3803057	111831454	Genotyped	Genotyped
Replication	12	<i>OAS1</i>	rs7967461	111832479	Genotyped	Genotyped
Replication	12	<i>VDR</i>	rs1859281	46502016	Genotyped	Genotyped
Replication	12	<i>VDR</i>	rs2544028	46502697	Genotyped	Genotyped
Replication	12	<i>VDR</i>	rs10747524	46509741	Genotyped	Genotyped
Replication	12	<i>VDR</i>	rs11608702	46515035	Genotyped	Genotyped
Replication	12	<i>VDR</i>	rs12721364	46517697	Genotyped	Genotyped
Replication	12	<i>VDR</i>	rs7968585	46518360	Genotyped	Genotyped
Replication	12	<i>VDR</i>	rs6580639	46518439	Genotyped	Genotyped
Replication	12	<i>VDR</i>	rs731236	46525024	Ungenotyped	Genotyped
Replication	12	<i>VDR</i>	rs7975232	46525104	Genotyped	Genotyped
Replication	12	<i>VDR</i>	rs11574113	46525167	Genotyped	Genotyped
Replication	12	<i>VDR</i>	rs1544410	46526102	Genotyped	Genotyped
Replication	12	<i>VDR</i>	rs11574085	46537061	Genotyped	Genotyped
Replication	12	<i>VDR</i>	rs11168267	46537809	Genotyped	Genotyped
Replication	12	<i>VDR</i>	rs11574077	46539194	Genotyped	Genotyped
Replication	12	<i>VDR</i>	rs2239182	46541678	Genotyped	Genotyped
Replication	12	<i>VDR</i>	rs2107301	46541837	Genotyped	Genotyped
Replication	12	<i>VDR</i>	rs2239180	46542313	Genotyped	Genotyped
Replication	12	<i>VDR</i>	rs2239179	46544033	Genotyped	Genotyped
Replication	12	<i>VDR</i>	rs11574066	46544902	Genotyped	Genotyped
Replication	12	<i>VDR</i>	rs12717991	46545393	Genotyped	Genotyped
Replication	12	<i>VDR</i>	rs886441	46549231	Genotyped	Genotyped
Replication	12	<i>VDR</i>	rs2189480	46550095	Genotyped	Genotyped
Replication	12	<i>VDR</i>	rs3819545	46551273	Genotyped	Genotyped
Replication	12	<i>VDR</i>	rs3782905	46552434	Genotyped	Genotyped
Replication	12	<i>VDR</i>	rs2239186	46555677	Genotyped	Genotyped
Replication	12	<i>VDR</i>	rs10735810	46559162	Genotyped	Genotyped
Replication	12	<i>VDR</i>	rs2254210	46559981	Genotyped	Genotyped
Replication	12	<i>VDR</i>	rs2238136	46563980	Genotyped	Genotyped
Replication	12	<i>VDR</i>	rs2853564	46564754	Genotyped	Genotyped
Replication	12	<i>VDR</i>	rs4760648	46566932	Genotyped	Genotyped
Replication	12	<i>VDR</i>	rs11168287	46571681	Genotyped	Genotyped
Replication	12	<i>VDR</i>	rs4328262	46571915	Ungenotyped	Ungenotyped
Replication	12	<i>VDR</i>	rs4237855	46573470	Ungenotyped	Genotyped
Replication	12	<i>VDR</i>	rs11574027	46573640	Genotyped	Genotyped
Replication	12	<i>VDR</i>	rs11574026	46574513	Genotyped	Ungenotyped
Replication	12	<i>VDR</i>	rs10875695	46579304	Genotyped	Genotyped
Replication	12	<i>VDR</i>	rs11168292	46579872	Genotyped	Genotyped
Replication	12	<i>VDR</i>	rs12721377	46581618	Genotyped	Genotyped
Replication	12	<i>VDR</i>	rs10783219	46581755	Genotyped	Genotyped
Replication	12	<i>VDR</i>	rs7299460	46582535	Genotyped	Genotyped
Replication	12	<i>VDR</i>	rs4760658	46582753	Genotyped	Genotyped
Replication	16	<i>IL4R</i>	rs2057768	27229596	Genotyped	Genotyped
Replication	16	<i>IL4R</i>	rs2107356	27230905	Genotyped	Genotyped
Replication	16	<i>IL4R</i>	rs6498012	27239475	Genotyped	Genotyped
Replication	16	<i>IL4R</i>	rs1110470	27243928	Ungenotyped	Genotyped
Replication	16	<i>IL4R</i>	rs4787948	27248560	Genotyped	Genotyped
Replication	16	<i>IL4R</i>	rs2283563	27253855	Ungenotyped	Genotyped
Replication	16	<i>IL4R</i>	rs3024530	27258188	Genotyped	Ungenotyped
Replication	16	<i>IL4R</i>	rs3024537	27260320	Genotyped	Genotyped
Replication	16	<i>IL4R</i>	rs1805010	27263704	Genotyped	Genotyped
Replication	16	<i>IL4R</i>	rs3024560	27264168	Genotyped	Genotyped
Replication	16	<i>IL4R</i>	rs3024571	27265428	Genotyped	Genotyped
Replication	16	<i>IL4R</i>	rs2301807	27265599	Genotyped	Genotyped
Replication	16	<i>IL4R</i>	rs3024578	27265852	Genotyped	Genotyped
Replication	16	<i>IL4R</i>	rs2239347	27266522	Genotyped	Genotyped
Replication	16	<i>IL4R</i>	rs3116578	27267337	Ungenotyped	Genotyped
Replication	16	<i>IL4R</i>	rs3024613	27271754	Genotyped	Genotyped

Appendix A Continued

<i>Study</i>	<i>Chromosome</i>	<i>Gene</i>	<i>Marker</i>	<i>Physical position</i>	<i>Illumina</i>	<i>Sequenom</i>
Replication	16	<i>IL4R</i>	rs3024614	27271846	Genotyped	Genotyped
Replication	16	<i>IL4R</i>	rs3024622	27272954	Genotyped	Genotyped
Replication	16	<i>IL4R</i>	rs4787423	27274835	Genotyped	Genotyped
Replication	16	<i>IL4R</i>	rs3024668	27279450	Genotyped	Genotyped
Replication	16	<i>IL4R</i>	rs2234897	27281113	Genotyped	Genotyped
Replication	16	<i>IL4R</i>	rs1805011	27281373	Genotyped	Genotyped
Replication	16	<i>IL4R</i>	rs1805012	27281465	Genotyped	Genotyped
Replication	16	<i>IL4R</i>	rs1805015	27281681	Genotyped	Ungenotyped
Replication	16	<i>IL4R</i>	rs1801275	27281901	Ungenotyped	Genotyped
Replication	16	<i>IL4R</i>	rs1805016	27282428	Genotyped	Genotyped
Replication	16	<i>IL4R</i>	rs1805014	27282530	Genotyped	Ungenotyped
Replication	16	<i>IL4R</i>	rs2074570	27282658	Ungenotyped	Genotyped
Replication	16	<i>IL4R</i>	rs8832	27283288	Genotyped	Genotyped
Replication	16	<i>IL4R</i>	rs3024685	27284411	Genotyped	Genotyped
Replication	16	<i>IL4R</i>	rs12102586	27285554	Genotyped	Genotyped
Replication	16	<i>IL4R</i>	rs4787956	27285750	Genotyped	Genotyped
Replication	16	<i>IL4R</i>	rs16976728	27289213	Genotyped	Genotyped
Replication	16	<i>IL4R</i>	rs4787426	27292232	Ungenotyped	Genotyped
Replication	16	<i>IL4R</i>	rs12445135	27293007	Genotyped	Genotyped
Replication	16	<i>IL4R</i>	rs4787427	27293895	Genotyped	Genotyped
Replication	16	<i>IL4R</i>	rs7191188	27296912	Genotyped	Genotyped
Replication	16	<i>IL4R</i>	rs6498015	27299125	Genotyped	Genotyped
Replication	16	<i>IL4R</i>	rs6498016	27299289	Genotyped	Genotyped
Replication	16	<i>IL4R</i>	rs2382722	27300127	Genotyped	Genotyped
Replication	16	<i>IL4R</i>	rs9944340	27301092	Genotyped	Genotyped
Replication	16	<i>IL4R</i>	rs6498017	27302359	Genotyped	Genotyped
Replication	19	<i>CEACAM21</i>	rs2302188	46777713	Genotyped	Genotyped
Replication	X	<i>FOXP3</i>	rs11796927	48844942	Genotyped	Genotyped
Replication	X	<i>FOXP3</i>	rs2294016	48849899	Genotyped	Genotyped
Replication	X	<i>FOXP3</i>	rs5952519	48851586	Genotyped	Genotyped
Replication	X	<i>FOXP3</i>	rs3761548	48874612	Genotyped	Genotyped
Replication	X	<i>FOXP3</i>	rs4824747	48885394	Genotyped	Genotyped
Replication	X	<i>FOXP3</i>	rs5906761	48887202	Genotyped	Genotyped
	3	<i>FINGERPRINT</i>	rs11130795	60873474	Ungenotyped	Genotyped
	3	<i>FINGERPRINT</i>	rs39639	124861224	Ungenotyped	Genotyped
	4	<i>FINGERPRINT</i>	rs6834736	57035453	Ungenotyped	Genotyped
	4	<i>FINGERPRINT</i>	rs6841061	186374692	Ungenotyped	Genotyped
	6	<i>FINGERPRINT</i>	rs4870405	156367804	Ungenotyped	Genotyped
	8	<i>FINGERPRINT</i>	rs2014286	16951496	Ungenotyped	Genotyped
	8	<i>FINGERPRINT</i>	rs1367972	62166007	Ungenotyped	Genotyped
	9	<i>FINGERPRINT</i>	rs12682834	87292377	Ungenotyped	Genotyped
	11	<i>FINGERPRINT</i>	rs1025412	14202872	Ungenotyped	Genotyped
	12	<i>FINGERPRINT</i>	rs10748087	66481362	Ungenotyped	Genotyped
	13	<i>FINGERPRINT</i>	rs1408229	36702353	Ungenotyped	Genotyped
	13	<i>FINGERPRINT</i>	rs2639486	82495935	Ungenotyped	Genotyped
	15	<i>FINGERPRINT</i>	rs12909691	91645766	Ungenotyped	Genotyped
	16	<i>FINGERPRINT</i>	rs8045964	80374234	Ungenotyped	Genotyped
	20	<i>FINGERPRINT</i>	rs6038115	5209132	Ungenotyped	Genotyped
	20	<i>FINGERPRINT</i>	rs6512586	47734553	Ungenotyped	Genotyped