

To my parents, Vicki and John Zuvich,
my biggest fans, for their infinite support.

In loving memory of my grandmother, Vera Laudermilch,
who was a woman ahead of her time,
for her great example and encouragements.

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LIST OF ABBREVIATIONS

3UTR	3' untranslated region
5UTR	5' untranslated region
ACP	Accelerated Cure Project
AITD	autoimmune thyroid disease
AS	ankylosing spondylitis
BWH	Brigham and Women's Hospital
CD	Crohn's disease
CD4+	"helper" T cells
CD8+	cytotoxic T cells
CDCV	common disease/common variant hypothesis
CEU	US residents with northern and western European ancestry collected by the Centre d'Etude du Polymorphisme Humain (CEPH)
CHB	Chinese people from Beijing
CIS	clinically isolated syndrome
CMS	University of Cambridge, England
CNS	central nervous system
CSF	cerebrospinal fluid
dbSNP	database of single nucleotide polymorphisms
DNA	deoxyribonucleic acid
EDSS	Extended Disability Status Score
FS	functional systems
GD	Graves' disease
GRRs	genotype relative risks
GWAS	genome-wide association studies

HLA	human leukocyte antigen
HWE	Hardy-Weinberg equilibrium
IgG	immunoglobulin G
IMSGC	International Multiple Sclerosis Genetics Consortium
JPT	Japanese people from Tokyo area
kb	kilobases
LD	linkage disequilibrium
MALDI TOF MS	Matrix-assisted laser desorption/ionization time-of-flight mass spectrometry
MDR	Multi-factor dimensionality reduction
MHC	Major Histocompatibility Complex
MRI	magnetic resonance imaging
MRV	multiple rare variants hypothesis
MS	multiple sclerosis
MSSS	Multiple Sclerosis Severity Score
nsSNP	nonsynonymous single nucleotide polymorphism
OR	odds ratio
PCR	polymerase chain reaction
PPMS	primary progressive multiple sclerosis
PRMS	progressive relapsing multiple sclerosis
PSA	psoriatic arthritis
PSO	psoriasis
QC	quality control
RA	rheumatoid arthritis
RRMS	relapsing-remitting multiple sclerosis
rs	reference sequence (NCBI)
RUSH	Rush University of Chicago

SLE	systemic lupus erythematosus
SNP	single nucleotide polymorphism
SPMS	secondary progressive multiple sclerosis
T1D	type 1 diabetes
Th1	T helper cells type 1
Th2	T helper cells type 2
Th17	T helper cells type 17
UCSF	University of California San Francisco
UK	United Kingdom
US	United States
WashU	Washington University
WASP	Whole-genome Association Study Pipeline
YRI	Yoruba people of Ibadan, Nigeria

Nucleotide Abbreviations:

A	adenine
C	cytosine
G	guanine
T	thymine

Amino Acid Abbreviations:

A	alanine
G	glycine
I	isoleucine
P	proline
Q	glutamine

R	arginine
S	serine
T	threonine
Y	tyrosine

Gene Abbreviations:

<i>AKT</i>	v-akt murine thymoma viral oncogene homolog 1
<i>AP-1</i>	jun oncogene (JUN)
<i>Bad</i>	Bcl-2-associated agonist of cell death
<i>Bax</i>	Bcl-2-associated X protein
<i>Bcl-2</i>	B-cell CLL/lymphoma 2
<i>Bcl-2L1</i>	Bcl-2-like 1
<i>Bcl-3</i>	B-cell CLL/lymphoma 3
<i>BID</i>	BH3 interacting domain death agonist
<i>Bim</i>	Bcl-2-like 11 (apoptosis facilitator) (Bcl2l11)
<i>c-myc</i>	myelocytomatosis oncogene homolog
<i>CIITA</i>	major histocompatibility complex class II transactivator
<i>C16orf75</i>	chromosome 16 open reading frame 75
<i>CD6</i>	Cluster of Differentiation 6
<i>CD58</i>	Cluster of Differentiation 58
<i>CD226</i>	Cluster of Differentiation 226
<i>Cdc25a</i>	cell division cycle 25 homolog A
<i>CLEC16A</i>	C-type lectin domain family 16 member A
<i>CPLA2</i>	cytosolic phospholipase a2
<i>CRLF2</i>	cytokine receptor-like factor 2 (thymic stromal lymphopoietin receptor)
<i>CSK</i>	c-src tyrosine kinase

<i>GCET2</i>	germinal center expressed transcript 2
<i>GSK-3</i>	frequently rearranged in advanced T-cell lymphomas (FRAT1)
<i>HLA</i>	human leukocyte antigen
<i>IL1R2</i>	interleukin-1-receptor type II
<i>IL2</i>	interleukin-2
<i>IL2RA</i>	interleukin-2 receptor subunit alpha
<i>IL2RG</i>	interleukin-2 receptor subunit gamma (gamma c)
<i>IL4</i>	interleukin-4
<i>IL4R</i>	interleukin-4 receptor
<i>IL5</i>	interleukin-5
<i>IL7</i>	interleukin-7
<i>IL7RA</i>	interleukin-7 receptor subunit alpha
<i>IL9</i>	interleukin-9
<i>IL12A</i>	interleukin-12 subunit alpha
<i>IL12B</i>	interleukin-12 subunit beta
<i>IL12RB1</i>	interleukin-12 receptor, beta 1
<i>IL12RB2</i>	interleukin-12 receptor, beta 2
<i>IL13</i>	interleukin-13
<i>IL13RA1</i>	interleukin-13-receptor alpha 1
<i>IL15</i>	interleukin-15
<i>IL17</i>	interleukin-17
<i>IL17A</i>	interleukin-17 subunit alpha
<i>IL17RA</i>	interleukin-17 receptor subunit alpha
<i>IL17RD</i>	interleukin-17 receptor subunit delta
<i>IL18</i>	interleukin-18
<i>IL21</i>	interleukin-21
<i>IL23</i>	interleukin-23

<i>IL27RA</i>	interleukin-27-receptor subunit alpha
<i>INFG</i>	interferon gamma
<i>IRF1</i>	interferon regulatoyr factor 1
<i>IRF8</i>	interferon regulatory factor 8
<i>IRS1</i>	insulin receptor substrate 1
<i>IRS2</i>	insulin receptor substrate 2
<i>Jak1</i>	Janus kinase 1
<i>Jak2</i>	Janus kinase 2
<i>Jak3</i>	Janus kinase 3
<i>LAG3</i>	lymphocyte-activation gene 3
<i>LOC400499</i>	hypothetical protein 400499
<i>LOC729954</i>	hypothetical protein 729954
<i>MCL-1</i>	myeloid cell leukemia sequence 1
<i>NFATC1</i>	nuclear factor of activated T-cells, cytoplasmic, calcineurin-dependent 1
<i>NFATC2</i>	nuclear factor of activated T-cells, cytoplasmic, calcineurin-dependent 2
<i>p27</i>	Cyclin-dependent kinase inhibitor member 1B (<i>CDKN1B</i>)
<i>p38</i>	mitogen-activated protein kinase 14 (<i>MAPK14</i>)
<i>PI3K</i>	phosphoinositide-3-kinase (alpha, beta, gamma, delta subunits)
<i>PLA2G4A</i>	phospholipase A2, group IVA
<i>PLA2G4B</i>	phospholipase A2, group IVB
<i>PLA2G4C</i>	phospholipase A2, group IVc
<i>PRKCA</i>	protein kinase C subunit alpha
<i>PRKCE</i>	protein kinase C subunit epsilon
<i>PRM1/2/3</i>	protamine 1, 2, and 3
<i>PTEN</i>	phosphatase and tensin homolog

<i>PTK2B</i>	protein tyrosine kinase 2 beta
<i>SHC1</i>	SHC (Src homolog 2 domain containing) transforming protein 1
<i>SHIP</i>	inositol polyphosphate-5-phosphatase (<i>INPP5D</i>)
<i>SKAP2</i>	src kinase associated phosphoprotein 2
<i>SOCS</i>	suppressors of cytokine signaling (<i>SOCS1-7</i>)
<i>Stat1</i>	signal transducer and activator of transcription 1
<i>Stat3</i>	signal transducer and activator of transcription 3
<i>Stat5a</i>	signal transducer and activator of transcription 5A
<i>Stat5b</i>	signal transducer and activator of transcription 5B
<i>Stat6</i>	signal transducer and activator of transcription 6
<i>TBET</i>	Th-1-specific T box factor
<i>TNFRSF1A</i>	tumor necrosis factor receptor superfamily member 1A
<i>TNP2</i>	transition protein 2
<i>TSLP</i>	thymic stromal lymphopoietin
<i>TYK2</i>	tyrosine kinase 2

Statistic Abbreviations/Symbols:

$\lambda_{\square\square}$ recurrence risk ratio for first degree relatives

LIST OF WEBSITES

<http://genome.ucsc.edu>

<http://www.ensembl.org/index.html>

<http://www.r-project.org/>

www.genome.gov/19518660#3

www.mult-sclerosis.org

www.ncbi.nlm.nih.gov