Harvard Medical School Curriculum Vitae

Date Prepar	ed:	January 14, 2013	Curriculu	ini vitae	
Name:		Robert M. Plenge			
Office Addr	ess:	New Research Buil 77 Avenue Louis P. Suite 168, Boston, I	asteur	Brigham and Womer 75 Francis Street, AS Boston, MA 02115	n's Hospital Arthritis Center SBII – 1
Home Addro	ess:	23 Boulder Brook H Wellesley, MA 024			
Work Phone	e:	617-525-4451			
Work Email	l:	rplenge@partners	.org		
Work FAX:		617-252-1902			
Place of Birt	th:	Phoenix, AZ			
Education 1992 1998 2000	B.S Ph. M.I		Biology Genetics Medicine		University of California- San Diego, San Diego, CA Case Western Reserve University, Cleveland, OH Case Western Reserve University
Postdoctora	l Trai	ining			
7/2000- 6/2001 7/2001-	Inte Res	ern	Medicine Medicine		University of California, San Francisco, CA University of California
6/2002 7/2002- 1/2006	Clin Fell	nical/Research low	Medicine/Div Rheumatolog and Allergy	vision of y, Immunology	Brigham and Women's Hospital (BWH), Boston, MA
7/2002- 11/2005 7/2003- 6/2008		earch Fellow	Medicine Genetics		Harvard Medical School (HMS), Boston, MA Broad Institute of MIT and Harvard, Cambridge, MA
Faculty Aca	demi	c Appointments			
7/2003- 6/2008		earch Scientist	Genetics		Broad Institute of MIT and Harvard
12/2005-	Inst	tructor	Medicine		HMS

6/2008 7/2008- Assistant Professor Medicine current

HMS

Appointments at Hospitals/Affiliated Institutions

2/2006- current	Associate Physician	Medicine, Division of Rheumatology, Immunology Allergy	BWH and
11/2011- current	Associate Member	Broad Institute, Inc.	MIT and Harvard
Other Profe	ssional Positions		
2005- current	Member	North American Rheumatoid Feinstein Institute, Manhasset	Arthritis Consortium (NARAC), t, NY
<u>Major Adm</u>	inistrative Leadership P	ositions	
Local 7/2008- current	Director, Genetics and Rheumatology, Immuno		BWH
2010-2011	Co-director, Immunolog		HMS
Committee S	<u>Service</u>		
Local 2005-2006 2008- current	Medical and Population Genetics planning committee Scientific Advisory Board		Broad Institute, Member Brigham Rheumatoid Arthritis Sequential Study (BRASS), BWH
2012	Lawrence Summers Fellowship: Application Review Panel		Broad Institute, Member
National 2012- current	Multidisiplinary Clinical Research Center (MCRC) Advisory Committee		University of California, San Francisco, Member
Professional	Societies		
2003- current	American College of RI 2003-2006 2006- 2010-	heumatology	Fellow Member Member, Committee on Research
2008-	American Society of Hu	uman Genetics	Member
current 2012-	Research and Education College of Rheumatolo 2012-	n Foundation of the American gy	Member, Scientific Advisory
	2012-		Council Member, Strategic Planning Committee
2012	American Society for C	linical Investigation	Member

Grant Review Activities

2009	Grant review committee	Psoriasis Foundation
	2009	Ad hoc member
2009-	Grant review committee	Alliance for Lupus Research
current	2009-current	Ad hoc member
2009-2011	Grant review committee	American College of Rheumatology,
		Education and Research Foundation
	2009-2011	Ad hoc member
2009-2011	Abstract review committee	European League Against Rheumatism, Annual Meeting
	2009-2011	Ad hoc member
2010	Grant review committee	Icelandic Research Foundation
	2010	Ad hoc member
2010-2011	Grant review committee	Wellcome Trust Foundation
	2010-2011	Ad hoc member
2011	Program review committee	Celiac Disease Consortium, Netherlands
	2011	Ad hoc member
2011	Grant review committee	David Phillips Fellowship
	2011	Ad hoc member
2012	NIH Study Section	Genetics of Human Disease
	2012	Ad hoc member
2012	NIH Study Section	IMM-M(08) Special Emphasis Panel
		National Institute of Allergy and Infectious
		Diseases (NIAID)
	2012	Ad hoc member
2012	NIH Study Section	ZDK1 GRB-N National Institute of
		Diabetes and Digestive and Kidney
		Diseases (NIDDK), Type 1 Diabetes Impact
		Award (DP3)
	2012	Ad hoc member
2012	Abstract review committee	American Society of Human Genetics,
		Annual Meeting
	2012	Ad hoc member

Editorial Activities

Ad hoc Reviewer

American Journal of Human Genetics Annals of the Rheumatic Diseases Arthritis & Rheumatism Arthritis Care and Research Arthritis Research and Therapy Clinical Immunology Clinical Rheumatology Diabetes European Journal of Human Genetics Genes & Immunity

Genetic Epidemiology Human Molecular Genetics Journal of American Medical Association (JAMA) Journal of Experimental Medicine Journal of Rheumatology Molecular Medicine Nature Communications Nature Genetics Nature Medicine Nature Reviews Genetic Nature Reviews Immunology Nature Reviews Rheumatology New England Journal of Medicine (NEJM) **PLos Genetics PLoS Medicine Tissue Antigens**

Other Editorial Roles

current		
2012-	Associate Editor	Arthritis and Rheumatism
2011-2012	Advisory Editor	Arthritis and Rheumatism
2007-2012	Editorial Board	Arthritis Research and Therapy

1992	<i>Phi Beta Delta</i> International Honor Society	University of California-San Diego	Educational achievement
1995	<i>Alpha Omega Alpha</i> Research Award	Case Western Reserve University, Lepow Research Day	Research
1995	The 1995 American Society of Human Genetics Predoctoral Clinical Award	American Society of Human Genetics Annual Meeting	Research
2007, 2008	Excellence in Tutoring Award	Harvard Medical School	Human Genetics
2008	Young Investigator Award	Department of Medicine, BWH	Research

Report of Funded and Unfunded Projects

Past	
2004-2009	Genetic basis of the immune response
	NIH, K08 AI55314
	PI
	Major goals: Project to learn population genetics, genomic variation, and disease associations relevant to immune-related diseases
2005-2006	Subsidized genotyping of a genome-wide association study

	NIH, U54 RR020278
	PI
	Major goals: Provide additional support for a genome-wide association study of rheumatoid arthritis to search for new risk loci
2007-2008	Genome-wide association study to search for genetic predictors of HIV viral load
	Gate's Foundation
	PI, Subcontract
	Major goals: Conduct a a genome-wide association study of HIV viral load to search for
	new loci
2007-2010	Driving Biology Project: genetic basis of rheumatoid arthritis
	NIH, U54 LM 008748
	PI, Subcontract
	Major Goals: Develop an infrastructure to use electronic medical records to study clinical
	and genetic aspects of rheumatoid arthritis
2008-2009	Young Investigator Award
	William Randolph Hearst Foundation
	PI
	Major Goals: Start a laboratory to study the genetics of rheumatoid arthritis
	Major Goals: Conduct a candidate gene and genome-wide association study to find alleles
2009-2011	that predict response to treatment with anti-TNF therapy
2009-2011	<i>Meta-analysis of GWAS data to identify novel rheumatoid arthritis risk loci</i> NIH, R01 AR057108
	PI (BWH)
	Major Goals: Conduct a meta-analysis of 6 genome-wide association studies to identify
	novel RA risk loci
2007-2012	Gene-environment interactions in rheumatoid arthritis
2007 2012	NIH, P60 AR47782
	Co-investigator
	Multidisciplinary Clinical Research Center Major Goals: Understanding the relationship
	between genes and the environment for rheumatoid arthritis
2010-2012	Deep sequencing of rheumatoid arthritis risk loci
	NIH, R01 AR059648
	PI (Broad Institute)
	Major Goals: next-generation sequencing of 25 RA risk genes identified by GWAS
Current	
2008-2013	Genome-wide association study of gout
2000 2012	NIH, R01 AR056291
	Co-investigator
	Major Goals: Conduct a genome-wide association study of uric acid levels and gout to
	search for new loci
2008-2013	Genotype-phenotype correlations of rheumatoid arthritis susceptibility genes
	Burroughs Wellcome Fund, Career Award for Medical Scientists
	PI (BWH)(\$700,000)
	Major Goals: Study genotype-phenotype relationships for rheumatoid arthritis risk loci
2009-2014	Dissection of two rheumatoid arthritis risk loci from genome-wide studies
	NIH, R01 AR056768
	PI (BWH)(\$1,558,082)
	Major Goals: Sequence, genotype and functional studies of two RA risk loci

2010-2014	Informatics for integrating biology and the bedside (i2b2), Driving Biology Project NIH, U54 LM008748
	PI, Subcontract (BWH)(Core 2: \$412,720)
	Major Goals: Develop an infrastructure to use electronic medical records to study clinical and genetic aspects of autoimmune diseases
2010-2015	Genetic predictors of response to anti-TNF therapy in rheumatoid arthritis
	NIH, U01 GM092691
	PI (BWH)(\$4,369,033)
	Major Goals: conduct genetic studies to understand treatment response in RA
2012-2013	Novel autoantigen discovery with a synthetic human peptidome in juvenile idiopathic
2012 2010	arthritis
	Harvard Catalyst
	PI (BWH)(\$50,000)
	Major Goals: pilot study to identify novel autoantigens that confer risk of JIA
2012-2013	Genomic analysis of the arthritis internet registry (AIR) cohort
	Broad SPARC and Arthritis Foundation
	PI (Broad Institute)(\$200,000)
	Major Goals: pilot study of GWAS and expression analysis in 300 AIR patients
2012-2014	The personalization of therapeutic efficacy and risk
	NIH, R24/U54 HL117798
	PI, subcontract (BWH)(\$142,777)
	Major Goals: identify biomarkers of efficacy and toxicity to NSAIDs
Current Un	funded Projects
2010-	
	VDJ sequencing in immune-mediated diseases
current	PI (BWH) Major Cooley rilet accurating of the T cell beta sheir and the Is becaus sheir in different
	Major Goals: pilot sequencing of the T-cell beta chain and the Ig heavy chain in different
2011	immune-mediated diseases
2011-	GWAS to drugs: small molecule screen of CD40 signaling pathway
current	PI (BWH) Maior Coolar diagonar pour lamellando inhibitary of the CD40 signaling notherway in P
	Major Goals: discover novel small molecule inhibitors of the CD40 signaling pathway in B
2011	lymphocyte cells
2011-	Systematic perturbation of autoimmune risk genes in primary CD4+ T cells
current	PI (BWH) Major Cools: understand the contribution of autoimmune risk games to the biology of
	Major Goals: understand the contribution of autoimmune risk genes to the biology of

Report of Local Teaching and Training

CD4+ T cells in humans

Teaching of S	<u>Students in Courses</u>	
UNIV. OF	CALIFORNIA – SAN DIEGO	
1991	Molecular Endocrinology	
	Class of 200 undergraduate students	10 hrs per week for 12 weeks
CASE		
WESTERN		
RESERVE		
1992-1993	Human Anatomy	
	15 doctoral nursing students	7 hrs per week for 10 weeks

HMS

2002-2003	Musculoskeletal Section	
	7 2 nd yr medical students	5 hrs per week for 1 week per year
2006	Molecular Biology and Genetics in	
	Moderns Medicine (HST160)	
	50 1 st yr graduate students	1 hr per week
2006-2012	Human Genetics (IN755.0)	
	7 1 st yr medical students	1.5 hrs per week for 8 weeks per year
2009	Genetics (202)	
	8 students	2 hrs per year as lecturer
2010-2011	Immunology 300	
	10-15 1 st -3 rd yr graduate students	2 hrs per year as invited lecturer
2012	Immunology 302qc	
	7 1 st -3 rd yr graduate students	2 hrs per year as invited lecturer

Formal Teaching of Residents, Clinical Fellows and Research Fellows (post-docs)

2005-2006	Genetic basis of common disease	Genetic Resident Workshop Series, BWH
	3-4 medical residents in combined	2 hrs per year
	Residency in Internal Medicine and Medical	1
	Genetics	
2007-2010	Genetic basis of rheumatic disease	Division of Rheumatology, Immunology and Allergy, BWH
	25 fellows in both Allergy and	1 hr lecture per year
	Rheumatology clinical training programs	
	Introductory Course	
<u>Clinical Sup</u>	ervisory and Training Responsibilities	
2006-current	Ambulatory Rheumatology Clinic	One half session/week for 8 weeks
	Facilitator for Residents at BWH	throughout the year
Laboratory a	and Other Research Supervisory and Training	ng Responsibilities
2008-current	Supervision of a research scientists in	Daily ongoing mentorship

2008-current	Supervision of a research scientists in	Daily ongoing mentorship
	computational genetics at BWH	
2008-current	Supervision of a research scientists in	Daily ongoing mentorship
	molecular immunology at BWH	

Formally Supervised Trainees and Faculty

2007-2010	Soumya Raychaudhuri, M.DPh.D., Assistant Professor of Medicine, HMS and BWH
	Provided co-mentorship with Dr. Mark Daly (MGH).
	Published 10 manuscripts, including 2 first-author manuscripts in Nature Genetics.
2008-2012	Eli Stahl, Ph.D., Assistant Professor of Medicine, Mt. Sinai School of Medicine
	Provided primary mentorship.
	Published 10 manuscripts, including 2 first-author manuscripts in Nature Genetics.
2008-	Jing Cui, Ph.D., Assistant Professor of Medicine, BWH and HMS
current	Provide primary mentorship.
	Dr. Cui is studying genetic predictors of response to anti-TNF therapy.
2009-2012	Bibi (Fina) Kurreeman, Ph.D., Assistant Professor of Medicine, Leiden University Medical

	Center		
	Provided primary mentorship.		
	Published 9 manuscripts, including 3 first-author manuscripts in American Journal of		
	Human Genetics.		
2011-2012	John Hutchinson, Ph.D., Research Associate Harvard School of Public Health		
	Provided primary mentorship.		
	Dr. Hutchinson studied epigenetics; has a first-author manuscript that is in preparation.		
2011-2012	Paul Klarenbeek, Ph.D., Medical Student, Amsterdam Medical College		
	Provided primary mentorship.		
	Dr. Klarenbeek studied T cell receptor repertoire sequencing; has a first-author manuscript		
	in preparation.		
2011-	Dorothee Diogo, Ph.D., Research Fellow in Medicine, BWH and HMS		
current	Provide primary mentorship.		
	Dr. Diogo is studying sequence variants in the human genome.		
2011-	Di Wu, Ph.D., Research Fellow in Medicine, BWH and HMS		
current	Provide primary mentorship.		
	Dr. Wu is studying the CD40 signaling pathway, genetics and drug discovery.		
2011-	Rumey Ishizawar, M.D., Ph.D., Instructor in Medicine, BWH and HMS		
current	Provide primary mentorship.		
	Dr. Ishizawar is studying autoimmune risk genes in CD4+ T cells.		
2012-	Yukinori Okada, MD, Ph.D., Research Fellow in Medicine, BWH and HMS		
current	Provide primary mentorship.		
	Dr. Okada is studying sequence variants in the human genome for risk of RA.		
2012-	Malgorzata (Gosia) Trynka, Ph.D., Research Fellow in Medicine, BWH and HMS		
current	Provide co-mentorship with Dr. Soumya Raychaudhuri (BWH).		
	Dr. Trynka is studying genomic pathways/networks in autoimmunity.		

Formal Teaching of Peers

No presentations below were sponsored by outside entities.

Genetic basis of rheumatoid arthritis	lecture
CME, HMS	Boston, MA
Genetic basis of rheumatoid arthritis	lecture
CME, New York University School of Medicine	New York, NY
Genetic basis of rheumatoid arthritis	lecture
CME, Johns Hopkins School of Medicine	Baltimore, MD
	CME, HMS Genetic basis of rheumatoid arthritis CME, New York University School of Medicine Genetic basis of rheumatoid arthritis

Local Invited Presentations

No presentations below were sponsored by outside entities.

2008	Genetic basis of rheumatoid arthritis/Grand Rounds
	Pediatric Rheumatology, Children's Hospital, Boston, MA
2008-2012	Genetic basis of rheumatoid arthritis/ i2b2 Summer Student Course Lecture
	BWH
2008	Genetic basis of rheumatoid arthritis/Rheumatology Review Lecture
	HMS
2008	Genetic basis of rheumatoid arthritis/Medical and Population Genetics Meeting Lecture
	Broad Institute, Cambridge, MA
2010	Genetic basis of rheumatoid arthritis/Program in Molecular and Genetic Epidemiology

	Harvard School of Public Health, Boston, MA
2011	Genetic basis of rheumatoid arthritis/Rheumatology Grand Rounds, BWH
2013	Genomes before clinical trials/Rheumatology Grand Rounds, BWH

<u>Report of Regional, National and International Invited Teaching and</u> <u>Presentations</u>

Invited Presentations and Courses

Regional, National, and International Invited Presentations and Courses

No presentations below were sponsored by outside entities. Regional 2009 Genetics of rheumatoid arthritis Innovative Therapies Conference, American College of Rheumatology, Philadelphia, PA 2009 Genetic basis of rheumatoid arthritis Advances in Rheumatology, New York University School of Medicine and Hospital of Joint Disease 2010 Genetic basis of rheumatoid arthritis/Rheumatology Grand Rounds UMass Medical Center, Worcester, MA 2012 Genetic basis of rheumatoid arthritis/Rheumatology Grand Rounds Rhode Island Hospital, Providence, RI 2012 Genetics and Drug Discovery/Lecture Series Novartis Research Foundation, Cambridge, MA 2012 Future of Genomics in Rheumatology/Invited Lecture Challenge the expert in Rheumatology II, Boston, MA National 1995 X-chromosome inactivation/Annual Meeting of Medical Student HHMI Fellows Howard Hughes Medical Institute, Chevy Chase, MD X-chromosome inactivation/45th Annual Meeting (abstract) 1995 American Society of Human Genetics 45th Annual Meeting, Minneapolis, MN 1997 X-chromosome inactivation/Human Molecular Genetics Conference (abstract) Gordon Research Conference, Newport, RI X-chromosome inactivation/47th Annual Meeting (abstract) 1997 American Society of Human Genetics 47th Annual Meeting, Baltimore, MD 1998 X-chromosome inactivation/Invited lecturer Genome Mapping, Sequencing and Biology, Cold Spring Harbor, NY X-chromosome inactivation/48th Annual Meeting (abstract) 1998 American Society of Human Genetics 48th Annual Meeting, Denver, CO 2005-Bi-weekly phone calls Member, North American Rheumatoid Arthritis Consortium (NARAC). I participate in ongoing research activities that pertain to the genetic basis of rheumatoid arthritis. We have bi-weekly phone calls to discuss our research projects with graduate students, fellows and senior investigators. 2007 Genetic basis of rheumatoid arthritis/ Idec B-cell Summit/Invited lecturer and consultant Genentech-Biogen, Los Angeles, CA Genetic basis of rheumatoid arthritis/Invited lecturer 2007

	American College of Rheumatology Innovative Therapies in Autoimmune Diseases, Washington, DC
2007	Genetic basis of common diseases/Invited lecturer
2007	Feinstein Institute, 1 st Year Human Genetic Graduate Student Series,
2007	Genetic basis of rheumatoid arthritis/Invited lecturer
2007	Federation of Clinical Immunological Societies Annual Meeting, San Diego, CA
2007	Genetic basis of rheumatoid arthritis/Grand Rounds
2007	Rheumatology, Oklahoma Medical Research Foundation
2007	Genetic basis of rheumatoid arthritis/Grand Rounds
2007	Rheumatology, University of Southern California Medical School
2008	Genetic basis of rheumatoid arthritis/Grand Rounds
	Rheumatology, Duke University, Durham, NC
2008	Genetic basis of rheumatoid arthritis/Invited lecturer
	Rheumatology Review, Johns Hopkins School of Medicine, Baltimore, MD
2008	Genetic basis of rheumatoid arthritis/Invited lecturer
	American College of Rheumatology "Within Our Reach" Meeting, Chicago, IL
2008	Genetic basis of rheumatoid arthritis/Invited lecturer
	Lupus Genetics Meeting, Oklahoma Medical Research Foundation, Oklahoma City
2008	CD40 is a genetic risk factor for rheumatoid arthritis/ Invited lecturer
	2008 Annual Meeting of American Society of Human Genetics, Philadelphia, PA
2009	Genetic basis of rheumatoid arthritis/Rheumatology Review Invited lecturer
	New York University School of Medicine, New York, NY
2009	Genetic basis of rheumatoid arthritis/Seminar in Advanced Rheumatology/Invited lecturer
	New York University School of Medicine and Hospital of Joint Diseases, New York, NY
2009	Genetic basis of rheumatoid arthritis/Invited lecturer
	American College of Rheumatology Innovative Therapies Conference, Washington, DC
2009	Genetic basis of rheumatoid arthritis/Invited lecturer
	American College of Rheumatology "Within Our Reach" Meeting, San Diego, CA
2009	Genetic basis of rheumatoid arthritis/Invited lecturer
• • • • •	Lowe's Annual Lecture, University of Alabama, Birmingham, AL
2009	Genetic basis of treatment response in rheumatoid arthritis/Invited lecturer
2000	American College of Rheumatology 2009 Annual Meeting, Philadelphia, PA
2009	Functional studies of rheumatoid arthritis risk genes/Invited lecturer
2000	American College of Rheumatology 2009 Annual Meeting, Philadelphia, PA
2009	Genetics of RA: from genotype to patient care/Invited lecturer
2000	Third Annual Symposium on Systemic Lupus Erythematosus, Boston, MA
2009	Square wheels: electronic medical records in discovery research/Invited lecturer
	Workshop on Opportunities to Maximize the Utility of Genomics Research Data through
2000	Electronic Health Information, Washington, DC
2009	Genetic Architecture of rheumatoid arthritis/Invited lecturer
2000	Department of Genetics, University of Alabama, Birmingham, AL Square wheels: electronic medical records in discovery research/Invited lecturer
2009	Using EHR Data for Discovery Research, Washington, DC
2009	The genetic architecture of rheumatoid arthritis/Invited lecturer
2009	Multidisciplinary Clinical Research Center (MCRC), Boston University, Boston, MA
2010	The genetic architecture of rheumatoid arthritis/Invited lecturer
2010	Rheumatology Grand Rounds, Feinberg School of Medicine, Northwestern University
2010	The genetic architecture of rheumatoid arthritis/Invited lecturer
2010	

	Rheumatology Grand Rounds, Cedars Sinai Medical Center, Los Angeles, CA
2011	The genetic architecture of rheumatoid arthritis/Invited lecturer
	Rheumatology Grand Rounds, UT Houston, Houston, Texas
2011	Electronic medical records for discovery research/Invited lecturer
	Internal Medicine Grand Rounds, UT Houston, Houston, Texas
2011	Shared data in genome-wide association studies/Invited lecturer
	Advancing Scientific Progress through Genomic Data Sharing and Access, National
	Cancer Institute Washington, DC
2011	The genetic architecture of rheumatoid arthritis/Invited lecturer
	Rheumatology Grand Rounds, Hospital for Special Surgery, New York, NY
2011	The genetic architecture of rheumatoid arthritis/Invited lecturer
	Internal Medicine Grand Rounds, Vanderbilt University, Nashville, TN
2011	Rheumatology Research Workshop/Organizer
	American College of Rheumatology, Miami, FL
2011	The genetic architecture of rheumatoid arthritis/Invited lecturer
	Epigenomics, Sequencing and SNiPs-2011, Boston, MA
2012	The cycle of discovery research in rheumatoid arthritis/Invited lecturer
	Keystone Symposium, Rheumatoid Arthritis, Sante Fe, NM
2012	The Future of Pharmacogenomics/Invited lecturer
	Rheumatology Research Workshop, Denver, CO
2012	Identifying TV-Relevant Variants to Phenotype /Invited speaker
	Joint NIH-Industry Target Validation Workstreams Workshop, Cambridge, MA
2012	Genetics and Drug Discovery/Invited speaker
	Autoimmunity Summit, Eli Lilly, San Diego, CA
2012	Polygenic architecture of pharmacological traits/Invited speaker
	NIDDK Workshop on the Pharmacogenomics of Metformin Responses, Washington, DC
2012	Embedded genomics/Invited speaker
	Rheumatology Grand Rounds, UCSD School of Medicine, San Diego, CA
2012	GWAS to high-throughput screen at the CD40 locus/Plenary presentation
	American College of Rheumatology 2012 Annual Meeting, Washington, DC

International

2007	Genetic basis of rheumatoid arthritis/Invited lecturer		
	Advances in Targeted Therapies Workshop, Athens, Greece		
2007, 2008	Genetic basis of rheumatoid arthritis/Invited lecturer		
	AutoCure Workshop, Stockholm, Sweden		
2009	Genetic basis of rheumatoid arthritis/Invited lecturer		
	European Leagues Against Rheumatism 2009 Annual Meeting, Copenhagen, Denmark		
2009	Genetic basis of rheumatoid arthritis/Invited lecturer		
	German Rheumatology Society 2009 Annual Meeting, Cologne, Germany		
2009	Genetic architecture of rheumatoid arthritis/Invited lecturer		
	Canadian Arthritis Network's Annual Scientific Conference, Vancouver BC, Canada		
2010	Genetic architecture of autoimmune diseases/Fundamental Lecture		
	European Workshop of Immune-Mediated Inflammatory Diseases, Barcelona, Spain		
2011	Genetic architecture of rheumatoid arthritis/Invited Lecture		
	Rheumatology Grand Rounds, Amsterdam Medical Center, Amsterdam, Netherlands		
2011	Genetic architecture of autoimmune diseases/Invited Lecture		
	European League Against Rheumatism, Annual Meeting, London, England		

2011	The cycle of discovery research in rheumatoid arthritis/Visiting Professor	
	Centre of Molecular Medicine, Karolinska Institute, Stockholm, Sweden	
2011	Genetic basis of complex traits/Invited lecturer	
	Post-graduate course on myositis, AutoCure Network, Sigtuna, Sweden	
2012	Genomic aspects of rheumatologic disorders/Invited lecturer, Annual Meeting	
	Asia Pacific League of Associations for Rheumatology, Dead Sea, Jordan	
2013	Human genetics to drug screens/Invited lecturer	
	Functional Genomics Workshop, London, England	

Report of Clinical Activities and Innovations

Current Licensure and Certification

2002-2012 Massachusetts Medical License2005 Board Certification in Rheumatology

Practice Activities

2006-2012	Ambulatory care	Rheumatology, BWH	One Session per week
2006-2012	Inpatient consult care	Rheumatology, BWH	Three weeks per year

Report of Scholarship

Peer reviewed publications in print or other media

Research Investigations

- 1. Naumova AK, **Plenge RM**, Bird LM, Leppert M, Morgan K, Willard HF, Sapienza C. *Heritability* of X chromosome inactivation in a large family. Am J Hum Genet. 1996;58(6):1111-19.
- 2. Hendrich BH, **Plenge RM**, Willard HF. *Identification and characterization of the human XIST gene promoter: implications for models of X chromosome inactivation*. Nuc Acids Res. 1997;25(13):2661-71.
- 3. **Plenge RM**, Hendrich BD, Schwartz C, Arena JF, Naumova A, Sapienza C, Winter RM, Willard HF. *A promoter mutation in the XIST gene in two unrelated families with skewed X chromosome inactivation*. Nat Genet. 1997;17(3):353-6.
- 4. **Plenge RM**, Tranebjaerg L, Schwartz C, Willard HF. *Evidence that mutations in the X-linked DDP gene cause incompletely penetrant and variable skewed X inactivation*. Am J Hum Genet. 1999;64(3):759-67.
- 5 **Plenge RM**, Percec I, Nadeau J, Willard HF. *Expression-based assay of an X-linked gene to examine effects of the X-controlling element (Xce) locus*. Mamm Genome 2000;11(5):405-8.
- 6. **Plenge RM**, Stevenson RA, Lubs HA, Schwartz C, Willard HF. *Skewed X chromosome inactivation is a common feature of X-linked mental retardation disorders*. Am J Hum Genet. 2002;71(1):168-73.

- 7. Percec I, **Plenge RM**, Nadeau JH, Bartolomei MS, Willard HF. *Autosomal dominant mutations affecting X inactivation choice in the mouse*. Science 2002;296(5570):1136-39.
- 8. Percec I, Thorvaldsen JL, **Plenge RM**, Krapp CJ, Nadeau JH, Willard HF, Bartolomei MS. *An N-ethyl-N-nitrosourea mutagenesis screen for epigenetic mutations in the mouse*. Genetics 2003;164(4):1481-94.
- 9. **Plenge RM,** Padyukov L, Remmers EF, Purcell S, Lee AT, Karlson EW, Wolfe F, Kastner DL, Alfredsson L, Altshuler D, Gregersen PK, Klareskog L, Rioux JD. *Replication of putative candidate gene associations with rheumatoid arthritis in over 4,000 samples from North America and Sweden: association of susceptibility with PTPN22, CTLA4 and PADI4.* Am J Hum Genet. 2005;77(6):1044-60.
- 10. Bonnen PE, Pe'er I, **Plenge RM**, Salit J, Lowe JK, Shapero MH, Lifton RP, Breslow JL, Daly MJ, Reich DE, Jones KW, Stoffel M, Altshuler D, Friedman JM. *Evaluating potential for whole-genome studies in Kosrae, an isolated population in Micronesia.* Nat Genet. 2006;38(2):214-17.
- 11. Graham RR, Kozyrev SV, Baechler EC, Reddy MVPL, **Plenge RM**, Bauer JW, Ortmann WA, Koeuth T, Escribano MFG, The Argentine and Spanish Collaborative Groups, Pons-Estel B, Petri M, Daly M, Gregersen PK, Martín J, Altshuler D, Behrens TW, & Alarcón-Riquelme ME. *A common haplotype of interferon regulatory factor 5 (IRF5) regulates splicing and expression and is associated with increased risk of systemic lupus erythematosus*. Nat Genet. 2006;38(5):550-5.
- 12. Price AL, Patterson NJ, **Plenge RM**, Weinblatt ME, Shadick NA, Reich D. *Principal components analysis corrects for stratification in genome-wide association studies*. Nat Genet. 2006;38(8):904-9.
- 13. Amos-Landgraf JM, Cottle A, **Plenge RM**, Friez M, Schwartz CE, Longshore J, Willard HF. *X chromosome-inactivation patterns of 1005 phenotypically unaffected females*. Am J Hum Genet. 2006;79(3):493-9.
- 14. Graham RR, Kyogoku C, Sigurdsson S, Vlasova IA, Davies LRL, Baechler EC, Plenge RM, Koeuth T, Ortmann WA, Hom G, Bauer JW, Gillett C, Burtt N, Cunninghame G, Onofrio R, Petri M, Gunnarsson I, Svenungsson E, Ronnblom L, Nordmark G, Gregersen PK, Moser K, Gaffney PM, Criswell LA, Vyse TJ, Syvanen A, Bohjanen PR, Daly MJ, Behrens TW, Altshuler D. *Three functional variants of IFN regulatory factor 5 (IRF5) define risk and protective haplotypes for human lupus*. Proc Natl Acad Sci USA 2007; 104:6758-63.
- 15. Kallberg H, Padyukov L, **Plenge RM**, Ronnelid J, Gregersen PK, van der Helm-van Mil AH, Toes RE, Huizinga TW, Klareskog L, Alfredsson L *Gene-Gene and Gene-Environment Interactions Involving HLA-DRB1, PTPN22, and Smoking in Two Subsets of Rheumatoid Arthritis.* Am J Hum Genet. 2007;80:867-75.
- 16. Remmers EF*, Plenge RM*, Lee AT, Graham RR, Hom G, Behrens TW, de Bakker PIW, Le JM, Lee H, Batliwalla F, Li W, Masters SL, Booty MG, Carulli JP, Padyukov L, Alfredsson L, Klareskog L, Chen WV, Amos CI, Criswell LA, Seldin MF, Kastner DL, and Gregersen PK STAT4 and the Risk of Rheumatoid Arthritis and Systemic Lupus Erythematosus. New Eng J Med. 2007;357

(10): 977-86. *contributed equally

- 17. Plenge RM*, Seielstad M*, Padyukov L, Lee AT, Remmers EF, Ding B, Liew A, Khalili H, Chandrasekaran A, Davies LRL, Li W, Tan AKS, Bonnard C, Ong RTH, Thalamuthu A, Pettersson S, Liu C, Tian C, Chen WV, Carulli J, Beckman EM, Altshuler D, Alfredsson L, Criswell LA, Amos CI, Seldin MF, W, Kastner DA, Klareskog L, and Gregersen PK *TRAF1-C5 as a risk locus for Rheumatoid Arthritis a genomewide study*. New Eng J Med. 2007;357(12):1199-209. *contributed equally
- 18. Karlson EW, Chibnik LB, Cui J, **Plenge RM**, Glass RJ, Maher NE, Parker A, Roubenoff R, Izmailova E, Coblyn JS, Weinblatt ME, Shadick NA. *Associations between HLA, PTPN22, CTLA4 genotypes and RA phenotypes of autoantibody status, age at diagnosis, and erosions in a large cohort study.* Ann Rheum Dis. 2008 Mar;67(3):358-63.
- 19. Plenge RM, Cotsapas C, Davies L, Price AL, de Bakker PIW, Maller J, Pe'er I, Burtt NP, Blumenstiel B, DeFelice M, Parkin M, Barry R, Winslow W, Healy C, Graham RR, Izmailova E, Roubenoff R, Parker AN, Coblyn J, Weinblatt ME, Glass R, Karlson EW, Maher N, Hafler DA, Lee DM, Brenner MB, Seldin MF, Remmers EF, Lee AT, Padyukov L, Alfredsson L, Gabriel SB, Purcell S, Klareskog L, Gregersen PK, Shadick NA, Daly MJ, and Altshuler D. *Two independent alleles at 6q23 associated with risk of rheumatoid arthritis*. Nat Genet. 2007;39(12):1477-82.
- 20. Tian C, **Plenge RM**, Ransom M, Lee A, Villoslada P, Selmi C, Klareskog L, Pulver AE, Qi L, Gregersen PK, and Seldin MF. *Analysis and application of European genetic substructure using 300 K SNP information*. PLoS Genet. 2008 Apr;4(1):e4.
- 21. Fernando MM, Stevens CR, Walsh EC, De Jager PL, Goyette P, **Plenge RM**, Vyse TJ, Rioux JD. *Defining the role of the MHC in autoimmunity: a review and pooled analysis.* PLoS Genet. 2008 Apr;4(4):e1000024.
- 22. Lee HS, Lee AT, Criswell LA, Seldin MF, Amos CI, Carulli JP, Navarrete C, Remmers EF, Kastner DL, **Plenge RM**, Li W, Gregersen PK. *Several regions in the major histocompatibility complex confer risk for anti-CCP- antibody positive rheumatoid arthritis, independent of the DRB1 locus.* Mol Med. 2008;14(5-6): 293-300.
- 23. Johnsen AK, **Plenge RM**, Butty V, Campbell C, Dieguez-Gonzalez R, Gomez-Reino JJ, Shadick N, Weinblatt M, Gonzalez A, Gregersen PK, Benoist C, and Mathis D. *A Broad Analysis of IL1 Polymorphism and Rheumatoid Arthritis.* Arthritis Rheum. 2008;58(7):1947-57.
- 24. Costenbader KH, Chang S, De Vivo I, **Plenge R** and Karlson EW. *PTPN22*, *PADI-4 and CTLA-4 Genetic Polymorphisms and Risk of Rheumatoid Arthritis in Two Longitudinal Cohort Studies: Evidence of Gene-Environment Interactions with Heavy Cigarette Smoking*. Arth Res Ther. 2008;10(3):R52.
- 25. Peters AL, **Plenge RM**, Graham RR, Altshuler DM, Moser KL, Gaffney PM, and Bishop GA. *A Novel Polymorphism of the Human CD40 Receptor with Enhanced Function*. Blood 2008;112(5):1863-71.
- 26. Taylor KE, Remmers EF, Lee AT, Ortmann WA, **Plenge RM**, Tian C, Chung SA, Nititham J, Hom G, Kao AH, Demirci FY, Kamboh MI, Petri M, Manzi S, Kastner DL, Seldin MF, Gregersen PK,

Behrens TW, Criswell LA.. Specificity of the STAT4 Genetic Association for Severe Disease Manifestations of Systemic Lupus Erythematosus. PLoS Genet. 2008 May;4(5):e1000084.

- 27. Ding B, Padyukov L, Lundström E, Seielstad M, **Plenge RM**, Rioux JD, Gregersen PK, Alfredsson L, Klareskog L. *Different patterns of associations with anti-citrullinated protein antibody-positive and anti-citrullinated protein antibody-negative rheumatoid arthritis in the extended major histocompatibility complex region*. Arthritis Rheum. 2009;60(1):30-8.
- 28. Graham RR, Cotsapas C, Davies L, Hackett R, Lessard CJ, Leon JM, Burtt NP, Guiducci C, Parkin M, Gates C, **Plenge RM**, Behrens TW, Wither JE, Rioux JD, Fortin PR, Graham DC, Wong AK, Vyse TJ, Daly MJ, Altshuler D, Moser KL, Gaffney PM. *Genetic variants near* TNFAIP3 *on* 6q23 *are associated with systemic lupus erythematosus*. Nat Genet. 2009;40(9):1059-61.
- 29. Liu C, Batliwalla F, Li W, Lee A, Roubenoff R, Beckman E, Khalili H, Damle A, Kern M, **Plenge RM**, Coenen M, Behrens TW, Furie R, Carulli JP, Gregersen PK. *Genome-wide association scan identifies candidate polymorphisms associated with differential response to anti-TNF treatment in Rheumatoid Arthritis*. Mol Med. 2008;14(9-10):575-81.
- 30. Raychaudhuri S, Remmers EF, Lee AT, Hackett R, Guiducci C, Burtt NP, Gianniny L, Korman BD, Padyukov L, Kurreeman FA, Chang M, Catanese JJ, Ding B, Wong S, van der Helm-van Mil AH, Neale BM, Coblyn J, Cui J, Tak PP, Wolbink GJ, Crusius JB, van der Horst-Bruinsma IE, Criswell LA, Amos CI, Seldin MF, Kastner DL, Ardlie KG, Alfredsson L, Costenbader KH, Altshuler D, Huizinga TW, Shadick NA, Weinblatt ME, de Vries N, Worthington J, Seielstad M, Toes RE, Karlson EW, Begovich AB, Klareskog L, Gregersen PK, Daly MJ, Plenge RM. Common variants at CD40 and other loci confer risk of rheumatoid arthritis. Nat Genet. 2008;40(10):1216-23.
- 31. Choy E, Yelensky R, Bonakdar S, **Plenge RM**, Saxena R, De Jager PL, Shaw SY, Wolfish CS, Slavik JM, Cotsapas C, Rivas M, Dermitzakis ET, Cahir-McFarland E, Kieff E, Hafler D, Daly MJ, and Altshuler D. *Genetic Analysis of Human Traits In-Vitro: Drug Response and Gene Expression in Lymphoblastoid Cell Lines*. PLoS Genet. 2008 Nov;4(11):e1000287.
- 32. Korman BD, Seldin MF, Taylor KE, Le JM, Lee AT, **Plenge RM**, Amos CI, Criswell LA, Gregersen PK, Kastner DL, Remmers EF. *The chromosome 7q region association with rheumatoid arthritis in females in a British population is not replicated in a North American case-control series*. Arthritis Rheum. 2009;60(1):47-52.
- 33. Liao KP, Gunnarsson M, Källberg H, Ding B, **Plenge RM**, Padyukov L, Karlson EW, Klareskog L, Askling J, Alfredsson L. *Specific association of type 1 diabetes mellitus with anti-cyclic citrullinated peptide-positive rheumatoid arthritis*. Arthritis Rheum. 2009;60(3):653-60.
- 34. Lee YC, Raychaudhuri S, Cui J, De Vivo I, Ding B, Alfredsson L, Padyukov L, Costenbader KH, Seielstad M, Graham RR, Klareskog L, Gregersen PK, **Plenge RM**, Karlson EW. *The PRL-1149 G/T polymorphism and rheumatoid arthritis susceptibility*. Arthritis Rheum. 2009;60(5):1250-4.
- 35. Cui J, Taylor KE, Destefano AL, Criswell LA, Izmailova ES, Parker A, Roubenoff R, **Plenge RM**, Weinblatt ME, Shadick NA, Karlson EW. (2009) *Genome-wide association study of determinants of anti-cyclic citrullinated peptide antibody titer in adults with rheumatoid arthritis*. Mol Med. 2009 May-Jun;15(5-6):136-43.

- 36. Gregersen PK, Amos CI, Lee AT, Lu Y, Remmers EF, Kastner DL, Seldin MF, Criswell LA, **Plenge RM**, Holers VM, Mikuls TR, Sokka T, Moreland LW, Bridges SL Jr, Xie G, Begovich AB, Siminovitch KA. *REL, encoding a member of the NF-kappaB family of transcription factors, is a newly defined risk locus for rheumatoid arthritis.* Nat Genet. 2009;41(7):820-3.
- Raychaudhuri S, Plenge RM, Rossin EJ, Ng AC; International Schizophrenia Consortium, Purcell SM, Sklar P, Scolnick EM, Xavier RJ, Altshuler D, Daly MJ (2009) *Identifying Relationships Among Genomic Disease Regions: Predicting Genes at Pathogenic SNP Associations and Rare Deletions*. PLoS Genetics. 2009 Jun;5(6):e1000534.
- 38. Greenway SC, Pereira AC, Lin JC, DePalma SR, Israel SJ, Mesquita SM, Ergul E, Conta JH, Korn JM, McCarroll SA, Gorham JM, Gabriel S, Altshuler DM, Quintanilla-Dieck Mde L, Artunduaga MA, Eavey RD, Plenge RM, Shadick NA, Weinblatt ME, De Jager PL, Hafler DA, Breitbart RE, Seidman JG, Seidman CE. (2009) *De novo copy number variants identify new genes and loci in isolated sporadic tetralogy of Fallot*. Nat Genet. 2009;41(8):931-5.
- 39. Raychaudhuri S, Thomson BP, Remmers EF, Eyre S, Hinks A, Guiducci C, Catanese JJ, Xie G, Stahl EA, Chen R, Alfredsson L, Amos CI, Ardlie KG; BIRAC Consortium, Barton A, Bowes J, Burtt NP, Chang M, Coblyn J, Costenbader KH, Criswell LA, Crusius JB, Cui J, De Jager PL, Ding B, Emery P, Flynn E, Harrison P, Hocking LJ, Huizinga TW, Kastner DL, Ke X, Kurreeman FA, Lee AT, Liu X, Li Y, Martin P, Morgan AW, Padyukov L, Reid DM, Seielstad M, Seldin MF, Shadick NA, Steer S, Tak PP, Thomson W, van der Helm-van Mil AH, van der Horst-Bruinsma IE, Weinblatt ME, Wilson AG, Wolbink GJ, Wordsworth P; YEAR Consortium, Altshuler D, Karlson EW, Toes RE, de Vries N, Begovich AB, Siminovitch KA, Worthington J, Klareskog L, Gregersen PK, Daly MJ, Plenge RM (2009) *Genetic variants at CD28, PRDM1, and CD2/CD58 are associated with rheumatoid arthritis risk.* Nat Genet. 2009;41(12):1313-8.
- 40. Stahl EA, Raychaudhuri S, Remmers EF, Xie G, Eyre S, Thomson BP, Li Y, Kurreeman FA, Zhernakova A, Hinks A, Guiducci C, Chen R, Alfredsson L, Amos CI, Ardlie KG; BIRAC Consortium, Barton A, Bowes J, Brouwer E, Burtt NP, Catanese JJ, Coblyn J, Coenen MJ, Costenbader KH, Criswell LA, Crusius JB, Cui J, de Bakker PI, De Jager PL, Ding B, Emery P, Flynn E, Harrison P, Hocking LJ, Huizinga TW, Kastner DL, Ke X, Lee AT, Liu X, Martin P, Morgan AW, Padyukov L, Posthumus MD, Radstake TR, Reid DM, Seielstad M, Seldin MF, Shadick NA, Steer S, Tak PP, Thomson W, van der Helm-van Mil AH, van der Horst-Bruinsma IE, van der Schoot CE, van Riel PL, Weinblatt ME, Wilson AG, Wolbink GJ, Wordsworth BP; YEAR Consortium, Wijmenga C, Karlson EW, Toes RE, de Vries N, Begovich AB, Worthington J, Siminovitch KA, Gregersen PK, Klareskog L, Plenge RM. Genome-wide association study meta-analysis identifies seven new rheumatoid arthritis risk loci. Nat Genet. 2010 May 9. PMID: 20453842
- 41. Cui J, Saevarsdottir S, Thomson B, Padyukov L, van der Helm-van Mil AH, Nititham J, Hughes LB, de Vries N, Raychaudhuri S, Alfredsson L, Askling J, Wedrén S, Ding B, Guiducci C, Wolbink GJ, Crusius JB, van der Horst-Bruinsma IE, Herenius M, Weinblatt ME, Shadick NA, Worthington J, Batliwalla F, Kern M, Morgan AW, Wilson AG, Isaacs JD, Hyrich K, Seldin MF, Moreland LW, Behrens TW, Allaart CF, Criswell LA, Huizinga TW, Tak PP, Bridges SL Jr, Toes RE, Barton A, Klareskog L, Gregersen PK, Karlson EW, Plenge RM. PTPRC rheumatoid arthritis risk allele is also associated with response to anti-TNF therapy. Arthritis Rheum. Mar 22 PMID: 20309874

- 42. Liao KP, Cai T, Gainer V, Goryachev S, Zeng-Treitler Q, Raychaudhuri S, Szolovits P, Churchill S, Murphy S, Kohane I, Karlson EW, **Plenge RM.** *Electronic medical records for discovery research in rheumatoid arthritis*. Arthritis Care Res. 2010 Mar 16. PMID: 20235204
- 43. Karlson EW, Chibnik LB, Kraft P, Cui J, Keenan BT, Ding B, Raychaudhuri S, Klareskog L, Alfredsson L, **Plenge RM.** *Cumulative association of 22 genetic variants with seropositive rheumatoid arthritis risk.* Ann Rheum Dis. 2010 Jun;69(6):1077-85. PMID: 20233754
- 44. Iannaccone CK, Lee YC, Cui J, Frits ML, Glass RJ, Plenge RM, Solomon DH, Weinblatt ME, Shadick NA (2010) Using genetic and clinical data to understand response to disease-modifying anti-rheumatic drug therapy: data from the Brigham and Women's Hospital Rheumatoid Arthritis Sequential Study. Rheumatology 2010;50(1):40-6. PMID: 20847201
- 45. Henrich TJ, Li JZ, Felsenstein D, Kotton CN, **Plenge RM**, Pereyra F, Marty FM, Lin NH, Grazioso P, Crochiere DM, Eggers D, Kuritzkes DR, Tsibris AM. *Xenotropic murine leukemia virus-related virus prevalence in patients with chronic fatigue syndrome or chronic immunomodulatory conditions*. J Infect Dis. 2010;202(10):1478-81. PMID: 20936980
- 46. International HIV Controllers Study. *The major genetic determinants of HIV-1 control affect HLA class I peptide presentation*. Science 2010;330(6010):1551-7. PMID: 21051598
- 47. Hughes LB, Reynolds RJ, Brown EE, Kelley JM, Thomson B, Conn DL, Jonas BL, Westfall AO, Padilla MA, Callahan LF, Smith EA, Brasington RD, Edberg JC, Kimberly RP, Moreland LW, **Plenge RM**, Bridges SL Jr. *Most common single-nucleotide polymorphisms associated with rheumatoid arthritis in persons of European ancestry confer risk of rheumatoid arthritis in African Americans*. Arthritis Rheum. 2010;62(12):3547-53. PMID: 21120996
- 48. Kurreeman F, Liao K, Chibnik L, Hickey B, Stahl E, Gainer V, Li G, Bry L, Mahan S, Ardlie K, Thomson B, Szolovits P, Churchill S, Murphy SN, Cai T, Raychaudhuri S, Kohane I, Karlson E, Plenge RM. Genetic Basis of Autoantibody Positive and Negative Rheumatoid Arthritis Risk in a Multi-ethnic Cohort Derived from Electronic Health Records. Am J Hum Genet. 2011;88(1):57-69. PMID: 21211616
- 49. Chen R, Stahl EA, Kurreeman FA, Gregersen PK, Siminovitch KA, Worthington J, Padyukov L, Raychaudhuri S, **Plenge RM.** *Fine mapping the TAGAP risk locus in rheumatoid arthritis*. Genes Immun. 2011 Jun;12(4):314-8. PMID: 21390051
- 50. Zhernakova A, Stahl EA, Trynka G, Raychaudhuri S, Festen EA, Franke L, Westra HJ, Fehrmann RS, Kurreeman FA, Thomson B, Gupta N, Romanos J, McManus R, Ryan AW, Turner G, Brouwer E, Posthumus MD, Remmers EF, Tucci F, Toes R, Grandone E, Mazzilli MC, Rybak A, Cukrowska B, Coenen MJ, Radstake TR, van Riel PL, Li Y, de Bakker PI, Gregersen PK, Worthington J, Siminovitch KA, Klareskog L, Huizinga TW, Wijmenga C, Plenge RM. Meta-Analysis of Genome-Wide Association Studies in Celiac Disease and Rheumatoid Arthritis Identifies Fourteen Non-HLA Shared Loci. PLoS Genet. 2011 Feb;7(2):e1002004. Epub 2011 Feb 24
- 51. Chibnik LB, Keenan BT, Cui J, Liao KP, Costenbader KH, **Plenge RM**, Karlson EW. *Genetic risk score predicting risk of rheumatoid arthritis phenotypes and age of symptom onset*. PLoS One.

6(9):e24380. Epub 2011 Sep 12. PMID: 21931699

- 52. Raychaudhuri S, Sandor C, Stahl EA, Freudenberg J, Lee HS, Jia X, Alfredsson L, Padyukov L, Klareskog L, Worthington J, Siminovitch KA, Bae SC, **Plenge RM**, Gregersen PK, de Bakker PI. *Five amino acids in three HLA proteins explain most of the association between MHC and seropositive rheumatoid arthritis*. Nat Genet. 2012 Jan 29;44(3):291-6. PMID: 22286218
- 53. Carroll RJ, Thompson WK, Eyler AE, Mandelin AM, Cai T, Zink RM, Pacheco JA, Boomershine CS, Lasko TA, Xu H, Karlson EW, Perez RG, Gainer VS, Murphy SN, Ruderman EM, Pope RM, **Plenge RM**, Kho AN, Liao KP, Denny JC. *Portability of an algorithm to identify rheumatoid arthritis in electronic health records.* J Am Med Inform Assoc. 2012 Feb 28. PMID: 22374935
- 54. Kurreeman FA, Stahl EA, Okada Y, Liao K, Diogo D, Raychaudhuri S, Freudenberg J, Kochi Y, Patsopoulos NA, Gupta N; CLEAR investigators, Sandor C, Bang SY, Lee HS, Padyukov L, Suzuki A, Siminovitch K, Worthington J, Gregersen PK, Hughes LB, Reynolds RJ, Bridges SL Jr, Bae SC, Yamamoto K, **Plenge RM.** *Use of a Multiethnic Approach to Identify Rheumatoid-Arthritis-Susceptibility Loci, 1p36 and 17q12.* Am J Hum Genet. 2012 Mar 9;90(3):524-32. PMID: 22365150
- 55. Stahl EA, Wegmann D, Kraft P, Chen R, Kallberg H, Kurreeman FAS, Gregersen PK, Alfredsson L, Siminovitch KA, Worthington K, de Bakker PIW, Raychaudhuri S*, **Plenge RM***. *Bayesian inference reveals polygenic architecture of four common disease*. Nat Genet. 2012 Mar 25;44(5):483-9. PMID: 22446960. *contributed equally
- 56. Ananthakrishnan AN, Guzman-Perez R, Gainer V, Cai T, Churchill S, Kohane I, Plenge RM, Murphy S. Predictors of severe outcomes associated with Clostridium difficile infection in patients with inflammatory bowel disease. Aliment Pharmacol Ther. 2012 Apr;35(7):789-95. PMID: 22360370
- 57. Okada Y, Terao C, Ikari K, Kochi Y, Ohmura K, Suzuki A, Kawaguchi T, Stahl EA, Kurreman F, Nishida N, Ohmiya H, Myouzen K, Takahashi M, Sawada T, Nishioka Y, Yukioka M, Matsubara T, Wakitani S, Teshima R, Tohma S, Takasugi K, Shimada K, Murasawa A, Honjo S, Matsuo K, Tanaka H, Tajima K, Suzuki T, Iwamoto T, Kawamura Y, Tanii H, Okazaki Y, Sasaki T, Gregersen PK, Padyukov L, Worthington J, Siminovitch KA, Lathrop M, Taniguchi A, Takahashi A, Tokunaga K, Kubo M, Nakamura Y, Kamatani N, Mimori T, **Plenge RM**, Yamanaka H, Shigeki Momohara S, Yamada R, Matsuda F, and Yamamoto K. *Meta-analysis of genome-wide association studies identifies multiple novel loci associated with rheumatoid arthritis in the Japanese population*. Nat Genet. 2012;44(5):511-6. doi: 10.1038/ng.2231. PMID: 22446963.
- 58. Chung SA, Xie G, Roshandel D, Sherva R, Edberg JC, Kravitz M, Dellaripa PF, Hoffman GS, Mahr AD, Seo P, Specks U, Spiera RF, William St Clair E, Stone JH, **Plenge RM**, Siminovitch KA, Merkel PA, Monach PA. *Meta-analysis in granulomatosis with polyangiitis reveals shared susceptibility loci with rheumatoid arthritis*. Arthritis Rheum. 2012 Oct;64(10):3463-71. PMID: 22508400
- 59. Pappas DA, Oh C, **Plenge RM**, Kremer JM, Greenberg JD. *Association of Rheumatoid Arthritis Risk Alleles with Response to Anti-TNF Biologics: Results from the CORRONA Registry and Meta-analysis.* Inflammation. 2012 Sep 25. [Epub ahead of print] PMID: 23007924

- 60. Eyre S, Bowes J, Diogo D, Lee A, Barton A, Martin P, Zhernakova A, Stahl E, Viatte S, McAllister K, Amos CI, Padyukov L, Toes RE, Huizinga TW, Wijmenga C, Trynka G, Franke L, Westra HJ, Alfredsson L, Hu X, Sandor C, de Bakker PI, Davila S, Khor CC, Heng KK, Andrews R, Edkins S, Hunt SE, Langford C, Symmons D; Biologics in Rheumatoid Arthritis Genetics and Genomics Study Syndicate; Wellcome Trust Case Control Consortium, Concannon P, Onengut-Gumuscu S, Rich SS, Deloukas P, Gonzalez-Gay MA, Rodriguez-Rodriguez L, Arlsetig L, Martin J, Rantapää-Dahlqvist S, Plenge RM*, Raychaudhuri S*, Klareskog L*, Gregersen PK*, Worthington J*. *High-density genetic mapping identifies new susceptibility loci for rheumatoid arthritis*. Nat Genet. 2012 Nov 11;44(12):1336-40. Epub 2012 Nov 11. PMID: 23143596 *contributed equally
- 61. Zaitlen N, Lindström S, Pasaniuc B, Cornelis M, Genovese G, Pollack S, Barton A, Bickeböller H, Bowden DW, Eyre S, Freedman BI, Friedman DJ, Field JK, Groop L, Haugen A, Heinrich J, Henderson BE, Hicks PJ, Hocking LJ, Kolonel LN, Landi MT, Langefeld CD, Le Marchand L, Meister M, Morgan AW, Raji OY, Risch A, Rosenberger A, Scherf D, Steer S, Walshaw M, Waters KM, Wilson AG, Wordsworth P, Zienolddiny S, Tchetgen ET, Haiman C, Hunter DJ, Plenge RM, Worthington J, Christiani DC, Schaumberg DA, Chasman DI, Altshuler D, Voight B, Kraft P, Patterson N, Price AL. *Informed conditioning on clinical covariates increases power in case-control association studies*. PLoS Genet. 2012 Nov;8(11):e1003032. doi: 10.1371/journal.pgen.1003032. Epub 2012 Nov 8. PMID: 23144628
- Xie G, Lu Y, Sun Y, Zhang SS, Keystone EC, Gregersen PK, Plenge RM, Amos CI, Siminovitch KA. *Identification of the NF-κB activating protein-like locus as a risk locus for rheumatoid arthritis*. Ann Rheum Dis. 2012 Dec 6. [Epub ahead of print] PMID: 23223422
- 63. Liao KP, Kurreeman F, Li G, Duclos G, Murphy S, P RG, Cai T, Gupta N, Gainer V, Schur P, Cui J, Denny JC, Szolovits P, Churchill S, Kohane I, Karlson EW, **Plenge RM**. *Autoantibodies, autoimmune risk alleles and clinical associations in rheumatoid arthritis cases and non-RA controls in the electronic medical records*. Arthritis Rheum. 2012 Dec 10. doi: 10.1002/art.37801. [Epub ahead of print] PMID: 23233247
- 64. Umicevic Mirkov M, Cui J, Vermeulen SH, Stahl EA, Toonen EJ, Makkinje RR, Lee AT, Huizinga TW, Allaart R, Barton A, Mariette X, Miceli CR, Criswell LA, Tak PP, de Vries N, Saevarsdottir S, Padyukov L, Bridges SL, van Schaardenburg DJ, Jansen TL, Dutmer EA, van de Laar MA, Barrera P, Radstake TR, van Riel PL, Scheffer H, Franke B, Brunner HG, **Plenge RM**, Gregersen PK, Guchelaar HJ, Coenen MJ. *Genome-wide association analysis of anti-TNF drug response in patients with rheumatoid arthritis*. Ann Rheum Dis. 2012 Dec 11. [Epub ahead of print] PMID: 23233654
- 65. Diogo D, Kurreeman F, Stahl EA, Liao KP, Gupta N, Greenberg JD, Rivas MA, Hickey B, Flannick J, Thomson B, Guiducci C, Ripke S, Adzhubey I, Barton A, Kremer JM, Alfredsson L; Consortium of Rheumatology Researchers of North America; Rheumatoid Arthritis Consortium International, Sunyaev S, Martin J, Zhernakova A, Bowes J, Eyre S, Siminovitch KA, Gregersen PK, Worthington J, Klareskog L, Padyukov L, Raychaudhuri S, **Plenge RM**. *Rare, Low-Frequency, and Common Variants in the Protein-Coding Sequence of Biological Candidate Genes from GWASs Contribute to Risk of Rheumatoid Arthritis*. Am J Hum Genet. 2012 Dec 19. doi:pii: S0002-9297(12)00625-8. 10.1016/j.ajhg.2012.11.012. [Epub ahead of print] PMID: 23261300
- 66. Köttgen A, Albrecht E, Teumer A, Vitart V, Krumsiek J, Hundertmark C, Pistis G, Ruggiero D, O'Seaghdha CM, Haller T, Yang Q, Tanaka T, Johnson AD, Kutalik Z, Smith AV, Shi J, Struchalin

M, Middelberg RP, Brown MJ, Gaffo AL, Pirastu N, Li G, Hayward C, Zemunik T, Huffman J, Yengo L, Zhao JH, Demirkan A, Feitosa MF, Liu X, Malerba G, Lopez LM, van der Harst P, Li X, Kleber ME, Hicks AA, Nolte IM, Johansson A, Murgia F, Wild SH, Bakker SJ, Peden JF, Dehghan A, Steri M, Tenesa A, Lagou V, Salo P, Mangino M, Rose LM, Lehtimäki T, Woodward OM, Okada Y, Tin A, Müller C, Oldmeadow C, Putku M, Czamara D, Kraft P, Frogheri L, Thun GA, Grotevendt A, Gislason GK, Harris TB, Launer LJ, McArdle P, Shuldiner AR, Boerwinkle E, Coresh J, Schmidt H, Schallert M, Martin NG, Montgomery GW, Kubo M, Nakamura Y, Tanaka T, Munroe PB, Samani NJ, Jacobs DR Jr, Liu K, D'Adamo P, Ulivi S, Rotter JI, Psaty BM, Vollenweider P, Waeber G, Campbell S, Devuyst O, Navarro P, Kolcic I, Hastie N, Balkau B, Froguel P, Esko T, Salumets A, Khaw KT, Langenberg C, Wareham NJ, Isaacs A, Kraja A, Zhang Q, Wild PS, Scott RJ, Holliday EG, Org E, Viigimaa M, Bandinelli S, Metter JE, Lupo A, Trabetti E, Sorice R, Döring A, Lattka E, Strauch K, Theis F, Waldenberger M, Wichmann HE, Davies G, Gow AJ, Bruinenberg M; LifeLines Cohort Study, Stolk RP, Kooner JS, Zhang W, Winkelmann BR, Boehm BO, Lucae S, Penninx BW, Smit JH, Curhan G, Mudgal P, Plenge RM, Portas L, Persico I, Kirin M, Wilson JF, Leach IM, van Gilst WH, Goel A, Ongen H, Hofman A, Rivadeneira F, Uitterlinden AG, Imboden M, von Eckardstein A, Cucca F, Nagaraja R, Piras MG, Nauck M, Schurmann C, Budde K, Ernst F, Farrington SM, Theodoratou E, Prokopenko I, Stumvoll M, Jula A, Perola M, Salomaa V, Shin SY, Spector TD, Sala C, Ridker PM, Kähönen M, Viikari J, Hengstenberg C, Nelson CP; CARDIoGRAM Consortium; DIAGRAM Consortium; ICBP Consortium; MAGIC Consortium, Meschia JF, Nalls MA, Sharma P, Singleton AB, Kamatani N, Zeller T, Burnier M, Attia J, Laan M, Klopp N, Hillege HL, Kloiber S, Choi H, Pirastu M, Tore S, Probst-Hensch NM, Völzke H, Gudnason V, Parsa A, Schmidt R, Whitfield JB, Fornage M, Gasparini P, Siscovick DS, Polašek O, Campbell H, Rudan I, Bouatia-Naji N, Metspalu A, Loos RJ, van Duijn CM, Borecki IB, Ferrucci L, Gambaro G, Deary IJ, Wolffenbuttel BH, Chambers JC, März W, Pramstaller PP, Snieder H, Gyllensten U, Wright AF, Navis G, Watkins H, Witteman JC, Sanna S, Schipf S, Dunlop MG, Tönjes A, Ripatti S, Soranzo N, Toniolo D, Chasman DI, Raitakari O, Kao WH, Ciullo M, Fox CS, Caulfield M, Bochud M, Gieger C. Genome-wide association analyses identify 18 new loci associated with serum urate concentrations. Nat Genet. 2012 Dec 23. doi: 10.1038/ng.2500. [Epub ahead of print] PMID: 23263486

Non-peer reviewed scientific or medical publications/materials in print or other media

Reviews, Chapters, Monographs and Editorials:

- 1. **Plenge R,** Rioux JD. *Identifying susceptibility genes for immunological disorders: patterns, power and proof.* Immunol Rev 2006 Vol. 210: 40-51. PMID: 16623763
- 2. **Plenge RM** and Criswell LA. *Genetic variants that predict response to anti-tumor necrosis factor therapy in rheumatoid arthritis: current challenges and future directions*. Current Opinion in Rheumatology, 20(2):145-52. PMID: 18349743
- 3. **Plenge RM.** *Shared Genetic Risk Factors for Type 1 Diabetes and Celiac Disease* (Editorial). New Eng J Med. 2008; Vol. 359 (26): 2837-2838. PMID: 19073966
- 4. **Plenge RM.** *Recent progress in rheumatoid arthritis genetics: one step towards improved patient care.* Curr Opin Rheumatol. 2009; May;21(3):262-71. PMID: 19365266
- 5. Yang JJ and **Plenge RM** *Genomic technology applied to pharmacological traits*. JAMA. 2009 Aug 10;306(6):652-3. PMID: 21828330

- 6. **Plenge RM.** *Rare protection against type 1 diabetes.* Genome Biol. 2009; 10(5):219. PMID: 19490597
- 7. **Plenge RM.** *Rheumatoid arthritis genetics: 2009 update*. Curr Rheumatol Rep. 2009 Oct;11(5):351-6. PMID: 19772830
- 8. **Plenge RM.** *Unlocking the pathogenesis of celiac disease*. Nat Genet. 2010 Apr;42(4):281-2. PMID: 20348961
- 9. **Plenge R.** *GWASs and the age of human as the model organism for autoimmune genetic research.* Genome Biol. 2010;11(5):212. PMID: 20441610
- 10. **Plenge RM**, Raychaudhuri S. *Leveraging human genetics to develop future therapeutic strategies in rheumatoid arthritis*. Rheum Dis Clin North Am. May;36(2):259-70. PMID: 20510233
- 11. **Plenge RM** and Bridges SL. *Personalized medicine in rheumatoid arthritis: miles to go before we sleep*. Arthritis Rheum. 2011 Mar;63(3):590-3. PMID: 21360486
- 12. **Plenge RM**, Louis Bridges S Jr, Huizinga TW, Criswell LA, Gregersen PK. *Recommendations for publication of genetic association studies in arthritis & rheumatism*. Arthritis Rheum. 2011 Oct;63(10):2839-47. PMID: 21702018
- 13. O'Shea JJ, **Plenge R.** *JAK and STAT Signaling Molecules in Immunoregulation and Immune-Mediated Disease*. Immunity. 2012 Apr 20;36(4):542-50. PMID: 22520847

Books/Textbooks for the medical or scientific community:

- 1. Gregersen PK, **Plenge RM.** *Emerging relationships: rheumatoid arthritis and the PTPN22 associated autoimmune disorders.* In: Holmadahl R, editor. Hereditary Basis of Rheumatic Diseases. Basel, Switzerland: Birkhauser Verlag, 2006, p. 61-78.
- 2. Gregersen PK, **Plenge RM**, Gulko P. *Genetic Basis of Rheumatoid Arthritis*. In: Firestein GS, editor. Rheumatoid Arthritis. Oxford, England: Oxford University Press, 2006, p. 3-14.
- 3. **Plenge RM**, Weinblatt ME. *Molecular Basis of Rheumatoid Arthritis*. In: Willard HW and Ginsburg GS, editors. <u>Handbook of Genomic Medicine</u>, 2009

Professional educational materials or reports, in print or other media

1. Contributor, Online HMS CME module for Rheumatoid Arthritis Genetics (2010)

Thesis

Genetic basis of skewed X chromosome inactivation, Case Western Reserve University School of Medicine (1998)

Narrative Report

My active research interests are directed at understanding the genetics of complex traits - those diseases

influenced by multiple genes and environmental factors. As a physician-scientist, my career goals are to apply genetic knowledge to clinical practice, thereby improving patient care for common medical conditions, such as rheumatoid arthritis (RA) and other autoimmune diseases. There are several areas of investigation that I believe are critical to my laboratory's core mission:

(1) **Discover risk alleles**. We employ and develop state-of-the-art genomics technology to identify new RA risk loci. Examples include genome-wide association studies and next-generation sequencing technology. Through international collaborations, we have identified more than 50 regions of the genome that are unambiguously associated with risk of RA. We have also demonstrated the polygenic architecture of RA and other complex traits.

(2) Understand molecular mechanisms of risk alleles. Having identified risk alleles for immunemediated traits, a next critical step is to understand how these alleles disrupt normal immune function. In this way, we hope to gain insight into fundamental mechanisms that lead to autoimmunity, which in turn could help guide the development of novel therapies. Our genetic studies have demonstrated the importance of T-cell activation by antigen presenting cells (*PTPN22, STAT4*, and *CTLA4* genes); the role of the IL6 (*IL6ST, IL6R*, and *TYK2* genes) and CD40 (*CD40*) signaling pathways; and the roles for citrullination (*PADI4*), natural killer cells (*CD244*), and chemotaxis (*CCL21*). We use primary immune cells from healthy subjects, as well as human cell lines, to study the mechanism of action of autoimmune risk alleles. We have used human genetics to guide the development of a high-throughput assay for a small molecule screen of inhibitors of the CD40 signaling pathway.

(3) **Translate genetic discoveries to improve patient care**. Finally, we are constantly striving to understand how these risk alleles can improve patient care. One approach is to determine how autoimmune risk alleles subset patients in meaningful ways (e.g., response to anti-TNF therapy, risk of cardiovascular disease). We work with an informatics team (www.i2b2.org) and as part of an international collaboration to investigate these clinical applications.

My teaching activities have focused on three areas. (1) For 2 weeks each year I serve as an Associate on the inpatient Rheumatology Consult Service at BWH. I mentor Rheumatology Fellows, as well as medical students and residents. (2) I mentor Internal Medicine Residents as part of my outpatient Rheumatology clinic (approximately 40 half-day sessions per year). (3) I serve as tutor for the first year course on human genetics at HMS.

I want to continue to be an active member of the BWH and HMS translational research community. Towards this end, I will engage in a variety of activities: cutting-edge genomics and immunology research; teaching genetics and immunology to graduate and medical students; and leadership roles within the Division and Department.