Web Table 1. List of Genome-wide Association Study Consortia (N=14) That do not Meet Analysis Definition,* as of April 1, 2011

Name of Consortium	Criteria not met*
The RNAi Consortium (TRC)	2,3
AZ Parkinson's Disease Consortium (APDC)	2
International HapMap Consortium	1,3
Autism Genetic Resource Exchange Consortium (AGRE)	1,2,3
Collaborative Programs of Excellence in Autism (CPEA)	1,3
Myocardial Infarction Genetics Consortium (MiGEN)	2
International Consortium to Identify Genes & Interactions Controlling Oral Clefts	1,2
Tobacco & Genetics Consortium (TAG)	1
NIMH Human Genetics Initiative Repository	1,3
Gene Ontology (GO)	2
GeneMSA (Multiple Sclerosis Association)	1
Central Connecticut Cancer Consortium	2,3
Starr Cancer Research Consortium	2,3
Consortium for Frontotemporal Dementia Research	2,3

^{*} For this analysis, a consortium is defined as:

¹⁾ A team of individual investigators who are associated with a study or a research institution, or a group of studies and research institutions;

²⁾ who <u>assemble</u> large data sets of genome-wide genotypic data and phenotypic information from existing resources, through recruitment of new study participants, *or* both;

³⁾ for the <u>purpose</u> of discovering associations between the genotypic variants and phenotypes, including diseases, risk factors, or other physical characteristics.

Web Table 2. List of Genome-wide Association Consortia (N=55) That Meet Analysis Definition, as of April 1, 2011

Name of Consortium	Acronym	Type of Members	Disease or trait studied	Date of first paper in NHGRI catalog	Website (Yes/No)	Guidelines Accessible (Yes/No)
Triple-Negative Breast Cancer Consortium	TNBCC	Research centers	Breast cancer	9/19/10	No	No
International Headache Genetics Consortium	IHGC	Research centers	Migraines	8/29/10	Yes	No
International Schizophrenia Consortium	ISC	Researchers	Schizophrenia	8/16/10	Yes	No
Psychiatric GWAS Consortium		Investigators	Schizophrenia, Bipolar Disorder, Depression	8/16/10	Yes	No
Cohorts for Heart & Aging Research in Genomic Epidemiology	CHARGE	Studies	Heart disease & aging	8/12/10	Yes	Yes
Autism Genome Project Consortium	AGP	Consortia	Autism	7/27/10	Yes	No
International Molecular Genetic Study of Autism Consortium	IMGSAC	Investigators	Autism	7/27/10	No	No
Autism Genetics Cooperative	AGC	Investigators	Autism	7/27/10	Yes	No
Genetic Investigation of Anthropometric Traits Consortium	GIANT	Research centers	Type 2 Diabetes; Height	6/27/10	No	No
Asia Breast Cancer Consortium		Studies	Breast Cancer	6/24/10	No	No
Genetic Factors for Osteoporosis Consortium	GEFOS	Studies	Osteoporosis	6/10/10	Yes	Yes
Study of Under-lying Genetic Determinants for Vitamin D & Highly Related Traits	SUN- LIGHT	Studies	Vitamin D deficiency	6/9/10	No	No
Type I Diabetes Genetics Consortium	T1DGC	Investigators	Type 1 Diabetes	6/9/10	Yes	Yes
Genetic Association Information Network	GAIN	Investigators	Migraine in bipolar disorder	6/7/10	No	No
NIMH Genetics Initiative for Bipolar Disorder Consortium		Research centers	Bipolar disorder	6/7/10	No	No
Canadian and North American Rheumatoid	NARAC-	Investigators	Rheumatoid	5/9/10	Yes	No

Arthritis Consortium-III Gene Environment Association Studies	III GENEVA	Research	arthritis Gene-	5/2/10	Yes	No	
Consortium	GENEVII	centers	environment associations	3/2/10	105	110	
Leukocyte Telomere Length Consortium	LTL	Studies	Telomere length	4/26/10	No	No	
International MS Genetics Consortium	IMSGC	Investigators	Multiple sclerosis	6/14/09	Yes	Yes	
Chronic Kidney Disease Genetics Consortium	CKDGen	Investigators	Renal disease	4/11/10	No	No	
Inflammatory Bowel Disease Genetics Consortium	IBDGC	Research centers	Ulcerative colitis	3/14/10	Yes	Yes	
Global Blood Pressure Genetics Consortium	Global BPGen	Studies	Blood Pressure, Atrial fibrillation	2/21/10	No	No	
International Consortium for Brain Mapping	ICBM	Research centers	Brain structure	2/17/10	No	No	
Frontotemporal Lobar Degeneration Consortium	FTLD	Investigators	Frontotemporal lobar degeneration	2/14/10	Yes	No	
Hematology Genetics Consortium	HaemGen	Investigators	Blood and biochemical traits	2/7/10	No	No	
Bipolar Disorder Genome Study Consortium	BiGS	Investigators	Major mood disorders/ Bipolar Disorder	1/17/10	No	No	
Genetic Markers for Osteoporosis Consortium	GEN- OMOS	Research centers	Osteoporosis	10/4/09	Yes	Yes	
EchoGen Consortium		Studies	Cardiac structure	7/8/09	No	No	
Pritzker Neuropsychiatric Disorders Research Consortium	PNDRC	Research centers	Mood disorders, schizophrenia	5/5/09	Yes	No	
Q-T Sudden Cardiac Death Consortium	QTSCD	Investigators	QT interval	3/22/09	No	No	
Q-T Genetics Consortium	QTGEN	Studies	QT interval	3/22/09	No	No	
							3

International Colorectal Cancer Genetic Association Consortium	COGENT	Research centers	Colorectal cancer	11/16/08	No	No
National Institute on Aging Longevity Consortium		Investigators	C-reactive protein	4/24/08	No	No
Finland-United States Investigation of Non- Insulin-Dependent Diabetes Mellitus Genetics	FUSION	Investigators	Type 2 Diabetes	4/26/07	Yes	No
The Australo-Anglo-American Spondyloarthritis Consortium	TASC	Investigators	Ankylosing spondylitis	1/10/10	No	No
R-R Genetics Consortium	RRGEN	Studies	Resting heart rate	7/16/10	No	No
Consortium of Investigators of Modifiers of BRCA 1/2	CIMBA	Research centers	Breast cancer	9/19/10	Yes	Yes
Ovarian Cancer Association Consortium	OCAC	Investigators	Ovarian cancer	8/2/09	Yes	Yes
Chronic Lymphocytic Leukemia Research Consortium Also called GenEpi of CLL Consortium	CRC	Investigators	Lymphoma	7/18/10	Yes	No
International Consortium on Lymphoma Epidemiologic Studies	Inter- Lymph	Investigators	Lymphoma	7/18/10	Yes	No
Breast Cancer Association Consortium	BCAC	Studies	Breast cancer	5/9/10	Yes	Yes
Pancreatic Cancer Cohort Consortium		Studies	Pancreatic cancer	8/2/09	Yes	No
Pancreatic Cancer Case-Control Consortium	PanC4	Investigators	Pancreatic cancer	8/2/09	Yes	Yes
Melanoma Genetics Consortium	GenoMEL	Research centers	Melanoma	7/5/09	Yes	Yes
Brain Tumor Epidemiology Consortium	BTEC	Investigators	Brain tumor	7/5/09	Yes	No
Electronic Medical Records and Genomics	eMERGE	Research centers	1 st paper on red blood cell traits	1/26/11	Yes	Yes
Alzheimer's Disease Genetics Consortium	ADGC	Investigators	AB1-42 Alzheimer's Disease	10/8/10	Yes	Yes
Consortium on Genetics of Alcoholism	COGA	Research centers	Event-related brain	11/2/10	No	No

International Schizophrenia Consortium	ISC	Investigators	oscillations Bipolar disorder, schizophrenia	2/2/11	Yes	No
Reproductive Genetics Consortium	ReproGen	Studies	Menarche	11/21/10	No	No
GWASNEO Consortium (NEO is psychological test)	•	Studies	Personality dimensions	12/2/10	No	No
International Serious Adverse Events Consortium	SAEC	Research centers	Stevens- Johnson syndrome	1/1/11	Yes	Yes
Marker of Density Consortium	MODE	Studies	Mammographic density	1/30/11	No	No
International Parkinson Disease Genomics Consortium		Studies	Parkinson's Disease	2/1/11	No	No
Candidate-gene Association Resources Consortium	CARe	Studies	PR Interval	2/10/11	Yes	No

Web Table 3. List of Diseases, Risk Factors and Traits Studied in the 55 Genome-wide Association Study Consortia That Meet the Analysis Definition by International Classification of Disease, Tenth Edition (ICD-10) codes (13)

ICD-10 Codes	Diseases	Related Risk Factors and Traits
C00-D48 Neoplasms	Colorectal Cancer, Pancreatic Cancer, Melanoma, Breast Cancer, Ovarian Cancer, Brain Tumor, Lymphoma	Mammographic Density
D50-D89 Diseases of blood		Red blood cell traits
E00-E90 Metabolic diseases	Type 1 Diabetes, Type 2 Diabetes, Vitamin D Deficiency	Waist-Hip Ratio
F00-F99 Mental and behavioral disorders	Schizophrenia, Bipolar Disorder, Depression, Asperger Disorder, Autism	Amyloid β-Peptide 1-42 (marker of Alzheimer's Disease), Event-related brain oscillations (study of alcohol consumption), Serum Urate
G00-G99 Diseases of nervous system	Frontotemporal lobar degeneration, Parkinson Disease, Multiple Sclerosis, Migraine	consumption), serum orace
I00-I99 Diseases of circulatory system	Heart Disease, Atrioventricular conduction, Atrial fibrillation, Hypertension	Cardiac structure, C-Reactive Protein, QT Interval, PR interval, Resting heart rate
J00-J99 Diseases of the respiratory system	Allergy (Atopy)	
K00-K99 Diseases of digestive system	Ulcerative colitis	
M00-M99 Diseases of musculoskeletal system	Rheumatoid arthritis, Ankylosing spondylitis, Osteoporosis	

N00-N99 Diseases of genitourinary system Renal Disease

V01-Y98 External causes Other Traits not listed in ICD-10 Drug-related Adverse Events

Brain structure, Gene-Environment Interaction, Height, Menarche, Personality dimensions, Telomere length

Web Table 4: Analysis of Genome-wide Association Study Consortia Publicly Accessible Guidelines Accessible From Websites, as of April 1, 2011

Name of Consortium	Acronym	Components of Guidelines					
		Organization / Governance	Research Protocols / Methodology	Institutional Review Board (IRB) Approval	Data Sharing/ Publication/ Authorship/ Intellectual Property		
Breast Cancer Association Consortium (25)	BCAC	Study groups	Genotyping and quality assurance		List of publications		
Consortium of Investigators of Modifiers of BRCA1/2 (20)	CIMBA	Study groups	Genotyping and quality assurance		Authorship guidelines and credit; List of publications		
Ovarian Cancer Association Consortium (14)	OCAC	Steering committee to guide activities and study groups	Genotyping and quality assurance		List of publications		
Pancreatic Cancer Case Control Consortium (15)	PANC4	Steering Committee	Data pooling requirements; Genome-wide scan options				
Genetic Factors for Osteoporosis Consortium (26)	GEFOS	Research Steering Committee and Advisory Board; Program Evaluation and Review Technique (Pert) diagram	Meta-analyses of ongoing and already funded GWA studies		Publishing data synopses at regular intervals		
Genetic Markers for Osteoporosis (21)	GENOM OS	Same as GEFOS	Applies novel tools to establish osteoporosis risk alleles				
Melanoma Genetics Consortium (22)	GenoMEL	Analysis Group reviews proposals; circulated to GenoMEL membership	Application procedure listing 10 steps for data sharing proposals	Evidence of IRB approval, plans for preserving subject confidentiality and	Researchers required to supply their data to GenoMEL after publication and/or 12		

International Multiple Sclerosis Genetics Consortium (16)	IMSGC	Governance structure: Governance Group, Strategy Group, Working Groups, Regular members; Project decisions made by Strategy and Governance Group members	Proposed projects using either repository samples or unpublished consortium data formally proposed to the Strategy Group	Use of samples is an opt-in process that requires written permission of the samples' designated custodian	All project proposals must have specific publishing/authorship plan; All data generated must be submitted to the Consortium Data Repository
Inflammatory Bowel Disease Genetics Consortium (23)	IBDGC	Six research Centers and a data coordinating center; Major decision made by Steering Committee, subcommittees for analysis, genotyping, phenotyping, publication and recruitment	Recruitment of cases and controls to generate a repository of biospecimens for use by consortium members and the external scientific community; Access to DNA and serum samples available to scientific community	Must obtain IRB clearance or waiver before samples will be shipped	Subcommittee for publications
Type 1 Diabetes Genetics Consortium (17)	T1DGC	Steering Committee, Coordinating Center, Regional networks; Member rights and responsibilities described in detail; Consortium responsibilities listed	Specific policies for access to repository samples and data with timeline and approval process	Assurance that the project has been reviewed for human subject protection by an IRB as part of application for access to data and samples	Confidentiality Certification required for all individuals with access to data; Detailed publication and presentation policies; Required TIDGC acknowledgement statement on all presentations and publications

honoring HIPAA months after completion required for data sharing proposals

Alzheimer's Disease Genetics Consortium (18) ADGC

Organization chart: Executive Committee, External Advisory committee, Senior PI committee, and subcommittees Collect blood samples from patients who are diagnosed with possible or probable AD as well as blood from cognitively normal individuals. Upon receipt of an eligible blood sample, will extract DNA for the ADGC and provide DNA back.

Memorandum of Understanding form and Analysis Proposal Guidelines available for ADGC members Specific publication guidelines for inclusion a main author list at top of manuscript; qualifications of authors; required acknowledgement wording; Memorandum of Understanding and Analysis proposal guidelines require approval to acquire data sets for analysis

Cohorts for Heart and Aging Research in Genomic Epidemiology (27) (Benefited from guidelines from the International Lung Cancer Consortium) CHARGE

Research Collaboration Committee; Analysis Committee; Primary work takes place in the Working Group (WG) Steering Committees, assisted by the Analysis Committee WG members share results for all successfully genotyped markers in the primary GWA scan; specific list of data to be shared WG members agree not to share whole genome genotype-phenotype findings with any outside groups without permission of the WG members; WG members share results for all successfully genotyped markers in the primary GWA scan; Specific publication and authorship guidelines; Upon publication, shared results will be posted on a public website; If intellectual property related pursued, the consortium needs to be made aware of claims

Electronic Medical Records and Genomics (24)	eMERGE	Five member sites, Network Coordinating Center, External Scientific Panel	Three workgroups focused on: 1) Consent and Community Consultation; 2) Electronic Medical Records & Informatics; 3) Genomics	Consent & Community Consultation workgroup focused on IRB issues including model language for consent forms	Published manuscripts listed
International Serious Adverse Events Consortium (19)	SAEC	Scientific Management Committee	Research data made available to qualified biomedical researchers, at no charge, in a uniform manner	Only those research data which are made anonymous and approved by the Scientific Management Committee shall be released to qualified researchers	Adopted a policy of limiting the intellectual policy constraints on genetic markers discovered by or arising from the research program activities conducted or funded by the Consortium; proactively make intellectual property filing to reduce the likelihood that use of its public data will be encumbered by "follow-on" patents