A Selection of Projects Related to the Workshop Themes

Project Title	Project Acronym	Project URL	URL for Data	Project Summary	Sample Types	Assays/Data Types	Funding Agencies	Status
Functional Genomics								
Encyclopedia of DNA Elements	ENCODE	https://www.genome.gov/encode	https://www.encodeproject.org	Data collection and integrative analysis of human and mouse epigenomic and trancrilptomic data, including reference maps	Human and Mouse	Epigenomics Cell lines Transcriptomics Functional genomics	NHGRI	Active
International Human Epigenome Consortium	IHEC	http://www.ihec-epigenomes.org/	https://epigenomesportal.ca/ihec/	Data collection and reference maps of human epigenomes for key cellular states relevant to health and diseases	Human	Transcriptomics Epigenomics	Consortium of projects funded by member nations	Active
Reference Epigenome Mapping Centers	REMC	http://www.roadmapepigenomics.org/	https://www.encodeproject.org/matrix/?type=Experiment&award.project=Roadmap	Data collection, integrative analysis and a resource of human epigenomic data	Human (healthy)	Transcriptomics Epigenomics	NIH Common Fund	Completed
PsychENCODE	PsychENCODE	http://grants.nih.gov/grants/guide/rfa-files/RFA-MH-14-020.html		Data collection and integrative analysis of human neural epigenomic, genomic, transcriptomic and proteomic data	Human	WES WGS Transcriptomics Epigenomics Proteomics	NIMH	Active
Functional Annotation of the Mammalian Genome	FANTOM	http://fantom.gsc.riken.jp		Data collection of CAGE transcriptomic data and data analysis to annotate human and mouse functional elements.	Human and Mouse	Transcriptomics CAGE	RIKEN	Active
4D Nucleome	4DN	https://commonfund.nih.gov/4Dnucleome	https://www.4dnucleome.org	To understand the principles behind the organization of the nucleus in space and time, the role nuclear organization plays in gene expression and cellular function, and how changes in the nuclear organization affect normal development as well as various diseases.	Human	Multi-omics Cell lines Imaging	NIH Common Fund	Active
Genomics of Gene Regulation	GGR	https://www.genome.gov/27561317/genomics-of-gene-regulation/	https://www.encodeproject.org/matrix/?type=Experiment&award.project=GGR	Determine how to develop predictive gene regulatory network models from genomic data	Human	Transcriptomics Epigenomics	NHGRI	Completed
Genotype-Tissue Expression Project	GTEx	http://www.gtexportal.org/home/		Data collection and analysis of variation in human gene expression, across individuals, and across >30 tissues from the same subjects	Human (healthy)	WGS WES Transcriptomics	NIH Common Fund	Active
Library of Integrated Network-based Cellular Signatures	LINCS	https://commonfund.nih.gov/LINCS/		Data collection and analysis of molecular signatures describing how different cell types respond to perturbing agents	Human	Transcriptomics Phosphoproteomics Cell lines Imagining Epigenomics	NIH Common Fund	Active
International Cancer Genome Consortium	ICGC	http://www.icgc.org/		Data collection and analysis of genomic, transcriptomic and epigenomic changes in 50 different tumor types (includes TCGA samples)	Human (tumor and normal)	WGS WES Transcriptomics Epigenomics	Consortium of projects funded by member nations	Active

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The Cancer Genome Atlas	TCGA	http://cancergenome.nih. gov/		Data collection and analysis of genomic, transcriptomic, and epigenomic changes in ~30 different tumor types, and repository for DNA and RNA sequence data	Human (tumor and normal)	WGS WES Proteomics Transcriptomics Epigenomics	NHGRI, NCI	Completed
Non-Coding Variants Program	NoVa	https://www.genome.gov/27564944/noncoding-variants-program-nova/		Development of computational approaches to interpret sequence variation in non-coding regions, and assessment of approaches through targeted data collection	Various	Functional assays	NHGRI, NCI, NIDA	Active
Knockout Mouse Phenotyping Program	KOMP2	https://commonfund.nih.gov/KOMP2/		Data collection for standardized phenotyping of a genome-wide collection of mouse knockouts; member of International Mouse Phenotyping Consortium (IMPC)	Mice	Phenotypic	NIH Common Fund	Active
HubMap	HubMap	https://commonfund.nih.gov/hubmap		to facilitate research on single cells within tissues by supporting data generation and technology development to explore the relationship between cellular organization and function, as well as variability in normal tissue organization at the level of individual cells	Human (Healthy)	Transcriptomics Phosphoproteomics Imaging Epigenomics	NIH Common Fund	Active
Human Cell Atlas	HCA	https://www.humancellatlas.org/		To create comprehensive reference maps of all human cells—the fundamental units of life—as a basis for both understanding human health and diagnosing, monitoring, and treating	Human	Multiple	Investigator-orgainized effort	Active
Toxicant Exposures and Responses by Genomic and Epigenomic Regulators of	TaRGET II	https://targetepigenomics.org	https://dcc.targetepigenomics.org	Multiple -omics measures of cellular response to toxicants.	Human	Multiple	NIEHS	Active
Extracellular RNA Communication	ERC	https://commonfund.nih.gov/exrna		to establish fundamental biological principles of extracellular RNA secretion, delivery, and impact on recipient cells; to describe exRNAs in human biofluids and the extent to which non-human exRNAs are present; to test clinical utility of exRNAs; and to provide a data and a resource repository for the community at-large	Human		NIH Common Fund	Active
Sequencing for Variant Discovery and Association								
NHGRI Genome Sequencing Program (including multiple co-funding sources)	GSP	http://gsp-hg.org/	https://www.ncbi.nlm.nih.gov/gap	i. Exomes in Mendelian disease for gene/variant discovery-- resolve as many Mendelain diseaes as possible; families; ii. Well-powered exome and genome studies in common, multiple complex diseases, multiple designs (case/control, family, etc.). Understand genomic architecture of common disease	Human	WES WGS Analysis	NHGRI, NHLBI, NEI, NIMH	Active
Trans-Omics for Precision Medicine	TopMED	https://www.nhlbiwgs.org/	https://www.ncbi.nlm.nih.gov/gap	Genomes in case/control designs related to cardiovascular phenotypes. Additional -omics data added.	Human	WES WGS Proteomics Metabolomics Analysis	NHLBI	Active
Alzheimer's Disease Sequecing Project	ADSP	https://www.niagads.org/adsp/content/home	https://www.niagads.org/adsp/content/home	Exomes (case/control) and genomes (families) in AD	Human	WES WGS Analysis	NIA	Active

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Type 2 Diabetes Genes	T2DGenes	www.type2diabetesgenetics.org/	www.type2diabetesgenetics.org/	Exomes and genomes in T2D	Human	WES WGS Analysis	NIDDK	Completed
Population Architecture using Genomics and Epidemiology	PAGE	https://www.pagestudy.org/	https://www.pagestudy.org/	Genotyping disease risk variants in diverse non-European populations	Human	Genotype	NHGRI	Completed
Electronic Medical Records and Genomics	eMERGE	https://www.genome.gov/27540473/electronic-medical-records-and-genomics-emerge-network/		Type disease-assoiated variants in patients at scale; integrate with electronic medical records, assess utility of varaint data.	Human	Genotype Targeted sequence EHR	NHGRI	Active
Gabriella Miller Kid's First	GMKF	https://commonfund.nih.gov/kidsfirst	https://kidsfirstdrc.org/	Childhood cancers and structural birth defects; families and small cohorts	Human	WGS Analysis	NIH Common Fund	Active
Biobank/ Large Cohort Sequencing								
All of Us	AoU/PMI	https://allofus.nih.gov	https://www.researchallofus.org/	To create a national resource of over a million Americans' health information. AoU aims to oversample in underrepresented communities (racial/ethnic minorities, women, etc.). The program will sequence whole genomes and generate genotype data; collect health/lifestyle/environmentla information. The program is part of the U.S.'s Precision Medicine Initiative	Human	WGS Genotype Transcriptomics EMR	NIH Common Fund	Active
UK BioBank	UKBB	https://www.ukbiobank.ac.uk/		Aims to improve prevention, diagnosis, and treatment of a wide range of serious and life-threatening illnesses. Follows the health/well-being of over 500,000 volunteer participants and provides updaed health information to qualified researchers.	Human	Samples EMR Imaging Genotyping WES Environmental Wearables	Wellcome Trust (with funding from Welsh, Scottish, British governments and other non-profits)	Active
Saudi Human Genome Project	SHG	https://www.saudigenomeprogram.org/en/	http://genomics.saudigenomeprogram.org/en/researchers/database-access/	An effort to solve genetic diseases and apply personalized medicine in Saudi Arabia by sequencing more than 100,000 individuals. The program offers free DNA sequencing and pre-marital genetic screening to Saudi residents.	Human	WES Genotyping	Saudi Arabian government	Active
NIMH Repository and Genomics Resource	NIMH-RGR	https://www.nimhgenetics.org/		Provides a collection of over 150,000 well-characterized, high quality patient and control samples from a range of mental disorders (i.e. autism, epilepsy, schizophrenia, etc.). Centers receive, process and store biomaterials, distirbute data to quaified researchers	Human	Stem cells Phenotypic data Cell lines Biopsies WGS WES	NIMH	Active

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Australian Genomics Health Futures Mission		http://www.health.gov.au/internet/budget/publishing.nsf/Content/budget2018-factsheet65.htm		An 10-year national approach to addressing clinical genomics in Australia using a cohort of 200,000 individuals. The program will invest in: new and expanded studies for rare diseases, cancers, and complex disease; clinical trials and technology development; supported collaboration opportunities and career pathways; ELSI	Human	Multiple	Australian government	Active
Estonia Personalized Medicine Programme (100,000 genomes)	PMP	https://www.geenivaramu.ee/en/about-us/estonian-biocentre		Recruit 100,000 aditional participants for the Estonian Biobank and genotype them (genome-wide) and provide personalized reports in the national e-health portal. People of other nationalities (ex. Russian, Ukrainian, etc.) are represented in the initial cohorts. Secondary research includes: ancient DNA, population genetics, mitochondrial/Y-chromosome, and cellular stress	Human	WGS Genotype EMR	Estonian government (Estonia Ministry of Social Affairs/National Institute for Health Development/Estonian Genome Center at University of Tartu)	Active
Japan Initiative on Rare and Undiagnosed Diseases	IRUD	https://www.amed.go.jp/en/program/IRUD/		National research consortium that connects over 400 hospitals with 34 IRUD clinical and analysis centers. Clinical and genetic data is gathered for each case and finidngs are shared with committees, patient-matching sites, and with qualified investigators within and outside	Human	WES WGS EMR Clinical trials Pharmacogenomics	Japan Agency for Medical Research and Development	Active
France Medecine Genomique 2025	NA	https://solidarites-sante.gouv.fr/IMG/pdf/genomic_medicine_france_2025.pdf		Ten year plan to construct a medical and industrial system to introduce precision medicine into the French healthcare system.	Human	WGS WES Transcriptomics ELSI	Alliance Aviesan	Active
BGI Million Chinese Genomes	NA	https://www.bgi.com/us/company/news/bgi-publishes-largest-ever-genomic-study-chinese-population-discoveries-140000-genomes-throughout	https://db.cng.org/cmdb	To sequence the genes of 1 million Chinese residents, including individuals across the country and of all ethnicities. The main goal is to understand how Chinese people "transform from health to disease", how the environment and interactions between genes and external factors influence the country's health.	Human	WGS WES NIPT Transcriptomics	China's Ministry of Science and Technology	Active
Miscellaneous Related								
Human Genome Reference Program	HGRP	https://www.genome.gov/pages/about/nachgr/september2018agendadocuments/sept2018council_hg_reference_program.pdf		The next iteration of NHGRI's support for the human genome reference assembly (formally GRC) is an upcoming program called the Human Genome Reference Program. The HGRP will sequence additional high-quality haplotype-resolved genomes from diverse populations; improve and implement the next generation of reference representations; develop new technology and bioinformatic tools to use on the	Human (and outside-supported mice, zebrafish, chicken)	Genomic transcripts Bioinformatic tools	NHGRI (previously included Wellcome-Sanger)	Active

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Brain Research through Advancing Innovative Neurotechnologies Initiative	BRAIN	https://www.braininitiative.nih.gov/		Aims to revolutionize our understanding of the human brain by developing new technology that images, tracks, and visualizes brain cells, circuits, neural activity, and other integrated neurological approaches.	Human	Imaging Transcriptomics Epigenomics Proteomics Metabolomics Physiological Measurements ELSI	NIH	Active
Clinical Genome Resource	ClinGen	https://www.clinicalgenome.org/		A genomic knowledge base (resource) that defines clinical relevance of genes and variants for use in precision medicine and research. Consortium shares genomic and phenotypic data, standardizes clinical annotations, develops machine-learning approaches to improve calling; and disseminates the knowledge and resources	Human	EMR Phenotypic WES WGS	NHGRI, NICHD	Active