

## **National Human Genome Research Institute - HN4**

(1) Provides leadership for and formulates research goals and long-range plans to accomplish the mission of the Human Genome Project, including the study of the ethical, legal, and social implications of human genome research; (2) fosters, conducts, supports, and administers research and research training programs in human genome research by means of grants, contracts, cooperative agreements, and individual and institutional research training awards; (3) provides coordination for genome research, both nationally and internationally, and serves as a focal point within NIH and the DHHS for Federal interagency coordination, collaboration with industry and academia, and international cooperation; (4) plans, supports and administers intramural, collaborative, and field research to study human genetic disease in its own laboratories, branches, and clinics; and (5) sponsors scientific meetings and symposia and collects and disseminates educational and informational materials related to human genome research to health professionals, the scientific community, industry, and the lay public.

## **Office of the Director - HN41**

(1) Plans, directs, and coordinates the development and progress of the Institute's programs; (2) develops major policy and program decisions based on an evaluation of the status of support and accomplishments of the Institute's program areas; (3) coordinates grant review and program management activities; (4) plans and organizes conferences and workshops; and (5) communicates with the scientific community and coordinates activities with other private and government agencies.

## **Communications and Public Liaison Branch - HN417**

(1) Advises the Institute Director and senior staff on a broad range of issues regarding the dissemination of information about the agency's programs; (2) distributes information about goals, programs, and advances in genomics, NHGRI's human genetics and ELSI research; (3) helps prepare reports, pamphlets, articles, news releases and other written, audiovisual or webbased materials about NHGRI research accomplishments, program, policies and goals; (4) communicates this information to audiences including the lay public, news media, Congress, other federal agencies, universities, and professional and consumer organizations; (5) coordinates the institute's public and private web sites; and (6) assists the Institute Director and other senior staff in developing and delivering talks to public and professional audiences.

### **Office of Workforce Diversity and Health Equity - HN418**

- (1) Advises the NHGRI Director and senior staff on a broad range of issues regarding genomics workforce diversity and health equity
- (2) Works in partnership with NHGRI divisions and branches and across the NIH institutes, offices, and centers to realize the goals of NHGRI's Action Agenda for Building a Diverse Genomics Workforce
- (3) Incubates ideas, secures partner commitments, and co-creates new programs and high-impact initiatives aimed at increasing the diversity of the genomics workforce
- (4) Evaluates progress towards achieving greater diversity in the genomics workforce
- (5) Provides strategic and programmatic leadership for NHGRI's efforts to reduce health disparities and foster health equity in areas related to genomics and genomic medicine

## **Division of Intramural Research - HN45**

(1) Plans and conducts a program of laboratory and clinical research related to the application of genome research to the understanding of human genetic disease and the development of human gene therapy; (2) acts as a focal point at NIH for genome research and maintains core facilities that serve as a resource for the entire NIH intramural research community; (3) evaluates research efforts and establishes intramural program priorities; (4) allocates funds, space, and personnel ceilings to ensure maximum utilization of available resources in the attainment of NHGRI objectives and integrates new research activities into the program structure; (5) collaborates with other NIH Institutes and Centers and external research institutions, and maintains an awareness of national and international research efforts in relevant program areas; and (6) advises the NHGRI Director and staff on intramural research programs and areas of science of interest to the Institute.

## **Office of the Clinical Director - HN451**

(1) Provides support and oversight for a wide variety of clinical research and training activities, including inpatient and outpatient services, education for genetic counseling studies, and training for M.D. and Ph.D. trainees engaged in the Metropolitan Washington Medical Genetics Training Program; (2) assures appropriate credentialing of NHGRI caregivers; (3) contributes to the operation of the NIH Clinical Center by providing genetic counseling consultations and participating in the Medical Executive Committee; (4) constitutes and supports the NHGRI Institutional Review Board and the Data Safety and Monitoring Board for oversight of clinical protocols; and (5) provides an infrastructure conducive to clinical research for NHGRI physicians and scientists directing investigational studies.

### **Medical Genetics and Genomic Medicine Training Program - HN4513**

(1) Provides accredited training opportunities for Ph.D. and M.D. candidates that leads to Board Certification by the American Board of Medical Genetics; and (2) sponsors training leading to certification in Clinical Genetics, Medical Biochemical Genetics, Clinical Biochemical Genetics, Clinical Molecular Genetics, and Clinical Cytogenetics.

## **Bioethics Core – HN4516**

(1) Provides bioethics consultation, education, and administrative infrastructure for human subjects research and (2) provides administrative infrastructure, training, and consultation for investigators related to the Institutional Review Board (IRB) process; (3) coordinates annual Responsible Conduct of Research educational sessions; (4) moderates bioethics rounds at clinical genetics case conferences; (5) participates in the Clinical Center's ethics consultation service and Ethics Committee; (6) addresses emergent bioethics education and consultation needs; and (7) engages in a program of evaluation research to help ensure that recommendations are empirically well-grounded and responsive to the various stakeholders involved.



## **Cancer Genetics and Comparative Genomics Branch - HN452**

(1) Defines the genetic changes involved in the susceptibility and progression of cancer  
(2) identifies both germline risk alleles and cumulative genetic changes arising throughout life that contribute to tumor formation; (3) pursues a range of experimental studies to identify the genes and variants that increase cancer susceptibility, as well as to develop an understanding of associated functional changes; (4) identifies mutations leading to susceptibility and progression of prostate, breast and endometrial tumors; (5) utilizes naturally occurring animal systems to localize cancer susceptibility genes in a background of reduced heterogeneity; and (6) develops and applies technologies, such as cDNA and SNP arrays, together with deep sequencing for genome-wide analysis aimed at identifying genetic variants, expression changes, or haplotypes that are unique to specific cancers, response to treatment, or stages of progression.

## **Reproductive Cancer Genetics Section - HN4522**

Conducts research into the genetic basis of reproductive cancers including, but not limited to, endometrial cancer; and conducts ongoing investigations using high-throughput nucleotide sequencing, whole genome genotyping, and linkage and association studies to understand the genetic basis of endometrial cancer susceptibility, as well as disease progression and outcome.

## **Comparative Genetics Section - HN4523**

(1) Localizes and studies genetic variants responsible for disease susceptibility and morphologic variation; (2) analyzes high-risk families by mapping or exome sequencing to find variants that increase susceptibility to prostate cancer, particularly to aggressive forms of the disease; (3) understands the role of low penetrant but frequent variants in cancer-associated genes, particularly those that increase risk or progression of disease; and (4) uses whole genome sequencing and genome wide studies of dog breeds to identify genomic variants important in disease susceptibility and morphological variation, with a focus on traits related to cancer, body size, and skull shape.

## **Cancer Genomics Unit - HN4527**

Adopts genomic technologies to cancer susceptibility inherited disorders such as Fanconi anemia in order to understand genotype-phenotype correlations, to develop better diagnostics and to explore novel therapeutic approaches.

## **Comparative Genomics Analysis Unit - HN4528**

Develops, evaluates, and applies genomic algorithms and software tools using high-performance computers, in an effort to understand and interpret the increasing amounts of sequence and genotyping data available both from public sources and from collaborations with other investigators.

## **Genomics Core - HN4529**

Consults NHGRI investigators on their genotyping needs and provides guidance as to the type of service needed, generates the appropriate genotyping data using both the ABI sequencer for STRP and Illumina technologies for SNP based genotypes, and assists with the analysis of data. Provides services related to physical mapping, DNA sequencing, and access to DNA panels.

## **Microarray Core - HN452A**

Provides intramural investigators with full service, cost-effective, and time-efficient access to comprehensive, state-of-the-art genomics, and transcriptomics technologies; and supports analysis on all commercial microarray platforms to offer a broad range of products and services, including whole genome gene expression, genotyping (SNP), epigenetics (DNA methylation), methylation, copy number variation (CNV, LOH and CGH), non-coding and microRNA analysis, proteomic microarray, custom microarray printing, hybridization, data mining and analysis, as well as consultation on experimental design.

### **Genetic Disease Research Branch - HN453**

(1) Plans and conducts research using genomic and genetic approaches to identify and understand mechanisms regulating human genetic diseases; (2) uses functional genomic approaches to study genetic alterations involved in human disease states; (3) utilizes genetically modified model organisms and cellular models to explore signaling pathways affected in human health, development and disease; and (4) determines the functions of cloned genes involved in human genetic diseases.



## **Cell Signaling and Immunity Section - HN4532**

(1) Plans and conducts research on signal transduction pathways focusing on signaling in the immune system and its contribution to normal cellular development and abnormal disease states; (2) uses a combination of genetic, genomic, biochemical, and cellular approaches including both transgenic and gene-targeting technology in mice to examine the function of tyrosine kinases and related signaling molecules involved in lymphocyte development, function and responses to infection and immunization; and (3) uses functional genomic, cellular and systems approaches to understand mechanisms of human immunity and immune-mediated diseases, including genetic causes of primary immunodeficiencies.

## **Genomics, Development and Disease Section - HN4534**

(1) Uses genomic tools and genetic manipulation of model systems to unravel genome function and to dissect gene regulatory pathways in development and disease; (2) integrates data from basic science studies with clinical information; (3) identifies pathways that regulate mammalian development; (4) understands how alterations in these pathways lead to disease states; and (5) develops paradigms for therapeutic interventions.

### **Embryonic Stem Cell and Transgenic Mouse Core - HN4537**

(1) Provides a shared support service to generate genetically engineered mice (GEM); (2) generates GEM by utilizing embryonal stem cell (ESC) technology and conventional transgenic methods (direct microinjection); (3) consults and guides researchers in planning transgenic and embryonic stem cell projects; (4) provides support to researchers in breeding and analyzing GEM generated through transgenic and ESC technology; (5) rederives imported mice into the animal facility and cryopreserves all important mice for investigators.; (6) provides investigators with in-house breeding stock of tester strains and other useful mice that are used across various animal users' protocols.

### **Cytogenetic and Microscopy Core - HN4538**

(1) Provides state-of-the-art molecular cytogenetic and microscopy services; (2) serves as a source for experimental planning, implementation of protocols, and conveyance of results; (3) implements new cytogenetic and microscopy techniques; (4) mentors trainees; and (5) maintains and utilizes state-of-the-art equipment and techniques.

## **Genetics and Molecular Biology Branch - HN454**

Uses state-of-the-art genetic and genomic technologies to study genomes and disease mechanisms, to translate research findings into improved diagnosis, treatments, and prevention of human diseases; provides rigorous mentoring to the next generation of genetics and genomics investigators and involves all trainees in translational research projects.

## **Hematopoiesis Section - HN4544**

(1) Studies hematopoiesis in mouse models and human patients; (2) identifies novel genes involved in the differentiation of red blood cells; (3) determines the regulatory signals for red cell gene expression at the genome level; and (4) performs testing of in vivo and in vitro methods to increase red cell production.

### **Flow Cytometry Core - HN454B**

(1) Provides investigators with state-of-the-art flow cytometry services; (2) serves as a source for experimental design and planning, execution of experiments and data analysis; (3) collaborates with researchers to reach scientific goals; (4) teaches trainees in the art of flow cytometry; and (5) maintains and utilizes state-of-the-art flow cytometry equipment.

## **Medical Genetics Branch - HN456**

(1) Conducts clinical and basic research into the genetic contribution to human growth and development in health and disease and the application of that knowledge to genetic diagnosis, counseling and therapy; (2) provides resources and oversight for the conduct of research; (3) investigates skeletal and connective tissue biology, disorders of human brain development and the process and effectiveness of genetic education and counseling; and (4) manages a multi-disciplinary staff including M.D.'s, Ph.D.'s, nurses, genetic counselors, and trainees dedicated to achieving these goals.



## **Human Biochemical Genetics Section - HN4564**

(1) Investigates metabolic disorders of man at the clinical, biochemical, molecular, and cell biological levels; (2) pursues clinical endeavors including patient care, controlled therapeutic trials, investigational drug use, employment of unique diagnostic tests, and description of new disorders or new presentations of known disorders; (3) determines the primary defect in rare genetic diseases; (4) studies the pathogenesis of specific clinical manifestations; (5) investigates the mechanisms of action of cellular processes whose aberrations result in metabolic disease states; (6) describes defective processes or systems which often provide the opportunity to elucidate normal function; (7) studies disorders of vesicle formation and trafficking; (8) investigates the metabolic diseases known collectively as Hermansky-Pudlak syndrome (HPS) which provide models for the genesis of three intracellular vesicles (the melanosome, lysosome, and platelet dense body); (9) describes various aspects of HPS in different ethnic groups; (10) elucidates the many molecular bases of HPS; (9) determines the primary cellular defect responsible for the distinctive phenotype of HPS; (10) studies each type of HPS to reveal critical aspects of how vesicles form or move inside cells; (11) investigates nephropathic cystinosis (a prototypic lysosomal transport disorder), sialuria (a defect in feedback inhibition of the rate-limiting step in sialic acid synthesis), and alkaptonuria (a progressive joint disorder due to deficiency of homogentisic acid oxidase, an enzyme in the tyrosine catabolic pathway); (12) studies unique and unusually instructive diseases to gain insight into normal and abnormal intracellular processes; and (13) designs therapeutic interventions to correct specific metabolic defects from knowledge acquired from these disease states.

### **Molecular Neurogenetics Section - HN4568**

(1) Conducts clinical and basic research into the factors contributing to the phenotypic variation observed in monogenic diseases, using Gaucher disease as a prototype disorder; (2) investigates the relationship between Gaucher disease and parkinsonism; and (3) explores new therapeutic approaches for Gaucher disease.

**Medical Genomics Unit - HN456C**

Develops and studies methods for conducting research on genomic and non-genomic datasets in order to perform genotypic and phenotypic analyses that aim to inform clinical and overall management and understanding of diverse aspects of human health and disease.

**Glycosphingolipid and Glycoprotein Disorders Unit - HN456D**

Conducts research and clinical trials on glycosphingolipid and glycoprotein disorders affecting the central nervous system. These uniformly fatal, poorly recognized lysosomal storage disorders affect both children and adults and there are currently no effective therapies.

## **Social and Behavioral Research Branch - HN459**

(1) applies genetic discoveries to improve interventions for preventing disease and promoting health and well-being; (2) applies social, behavioral, and communication theories to understand the essential elements of communicating genetic risk effectively; (3) develops and refines theory-based methods for communicating genetic risk to affected individuals, families, communities and populations; (4) understands how social factors (e.g., interpersonal, organizational, cultural and societal) influence genetic discoveries and research; and (5) investigates the ethical and public policy implications of genetic research and the use of genetics in clinical practice.

## **Social Network Methods Section - HN4594**

(1) Develops methods to measure and model the complexities of social systems; (2) utilizes these methods to understand the social, psychological, and communicative context of families at risk for hereditary disease; (3) translates this understanding into effective network-based interventions; (4) investigates differences in the diffusion or coping processes across families from different ethnic and racial backgrounds and across different genetic diseases; (5) investigates how formal support systems, such as health care providers (i.e. genetic counselors, general practitioners), family members who are not at-risk (e.g. spouses), as well as those who are "like family" (e.g. friends) participate in the process of decision making, communication and support for at-risk family members; (6) understands the familial culture from a network perspective that provides important information for the delivery of genetic counseling services and dissemination of genetic risk information; and (7) develops network-based interventions that facilitates sharing of risk information throughout the family system.

## **Neurobehavioral Clinical Research Section - HN4595**

(1) Conducts research at the intersection of neuroscience, basic behavioral science and social science to advance understanding of the etiology, clinical course and treatment of attention deficit hyperactivity disorder (ADHD); (2) defines the genetic bases of the abnormal neurodevelopmental trajectories that characterize childhood ADHD; (3) characterizes subtypes of the disorder based on neurobehavioral profiles; (4) develops tools to predict long term outcomes; and (5) explores the genetic, epigenetic, behavioral and interpersonal factors that influence clinical course and response to pharmacological and behavioral treatments.

### **Immersive Simulation Program - HN4597**

Develops and applies innovative simulation methods to conduct and support research to anticipate and shape genomic translation for health and healthcare.



**Health Disparities Unit - HN4598**

Conducts research that evaluates approaches to integrating new genomic knowledge into clinical settings without exacerbating inequities in healthcare delivery.

**Social Epidemiology Research Unit - HN459B**

Conducts research related to the effects of social determinants on cardiovascular disease morbidity and mortality as well as the effects of social determinants on gene expression among disproportionately affected sub-populations.

**Engagement Methods Unit - HN459C**

Develops novel tools and modes of scientific communication and public engagement; evaluates impact and effectiveness of dissemination of genomic knowledge, with a focus on behavioral conditions; and collaborates with practitioners to create an evidence base for genomic applications.

**Health Communication and Behavior Unit - HN459D**

Investigates and develops innovative communication approaches for translating genomics discovery in the context of common, complex health conditions to improve individual and public health.

## **Systems Biology and Genome Engineering Section- HN4539**

The Systems Biology and Genome Engineering Section's goal is to systematically understand the effects of genetic differences. Researchers in this section study genetic diversity across an expansive range, such as natural variation between individuals or between species, or novel genetic changes that they create. By studying the functional effects of these variants, section researchers work to learn principles of genome function.

Section researchers use genome engineering to direct activities such as genome editing or recombination, using budding yeast as a model organism. As the space of potential genetic variants is immense, the section's researchers study large numbers of genetic differences in parallel. They do this by using large-scale oligonucleotide synthesis to generate a diversity of instructions targeting desired modifications, in order to generate pools of cells containing thousands of unique genomic changes. In addition to existing genome engineering methods, the section is continually developing new approaches to further expand the genetic space that can be studied.

### **Intramural Training Office - HN45A**

(1) Supports NHGRI DIR trainees' professional needs through mentoring, training programs and outreach activities to enhance institutional experiences while enabling achievement of career goals; and (2) trains and retains a highly skilled and diverse genomic and genetics workforce, providing future leaders for the global community.

### **Office of Laboratory Animal Medicine - HN45C**

(1) Provides veterinary medical support to research animal models; (2) trains NHGRI investigators in all aspects humane care and use of animals in biomedical and behavioral research, teaching, and testing; (3) provides research support to investigators using animal models; (4) aides in development/refinement of research biomethodologies; (5) assists in Animal Study Proposal development; and (6) orders, receives, and ships research models.

## **Technology Transfer Office - HN45D**

(1) Educates administrative and scientific personnel about patent, licensing, CRADA- and other technology transfer-related procedures and policies.; (2) facilitates the exchange of materials and information between NHGRI scientists and their collaborators and fosters productive interactions between intramural NHGRI labs and outside collaborators by effectively negotiating the legal, financial and scientific terms for these joint research projects; and 3) contributes to the successful commercialization of promising NHGRI research discoveries by facilitating interactions and providing advice and oversight to NHGRI scientists and potential collaborators and/or licensees.



## **NIH Intramural Sequencing Center - HN45E**

(1) Performs large-scale DNA sequencing for NIH intramural investigators; (2) provides expert advice for setting up new genomic sequencing projects; and (3) serves as the focal point at NIH for genomic sequencing technologies, chemistries, and instrumentation as well as innovative sequence analysis software.

## **Computational and Statistical Genomics Branch - HN45H**

(1) Develops and applies methods in statistics, bioinformatics, genetics, genomics, and computer science to identify genes and regulatory elements that are responsible for disease expression and trait variation in humans; (2) uses computationally intensive approaches to analyze large-scale genomic data from population-based and family-based studies of humans, as well as from studies of model organisms, to accomplish this mission; (3) employs comparative genomic approaches to understand the evolution and function of protein families and their ultimate role in human disease; (4) develops and disseminates new software tools and bioinformatic approaches; and (5) serves as a focal point for research and training in statistical genetics, genetic epidemiology, and bioinformatics.

### **Statistical Genetics Section - HN45H3**

(1) Performs research in both theoretical and applied statistical genetics as applied to human traits and disease; (2) performs data collection; power analysis, linkage, association and interaction analysis of human data; (3) analyzes large scale sequence and other genomic datasets in humans; (4) investigates the properties of various methods of statistical genetic analysis through computer simulation; and (5) emphasizes studies on complex diseases such as human cancers, eye diseases, and malformations.

## **Computational Genomics Unit - HN45H5**

Uses phylogenetic and comparative genomic techniques to study developmental proteins that play a fundamental role in the specification of body plan, pattern formation, and cell fate determination during metazoan development; and employs a variety of computational approaches to understand the evolution and function of these proteins and their ultimate role in human disease.

## **Bioinformatics and Scientific Programming Core - HN45H6**

(1) Supports the research being performed by NHGRI investigators by providing expertise and assistance in bioinformatics and computational analysis; (2) facilitates access to specialized software and hardware; (3) develops generalized software solutions that can address a variety of questions in genomic research; (4) develops database solutions for the efficient archiving and retrieval of experimental and clinical data; (5) disseminates new software and database solutions to the genome community at-large; (6) collaborates with NHGRI researchers on computationally intensive projects; and (7) provides educational opportunities in bioinformatics to investigators and trainees.

## **Genome Informatics Section- HN45H7**

(1) Designs and applies efficient algorithms for the analysis of large-scale genomic data; (2) develops fundamental software tools for the assembly, comparison, and exploration of genomes and metagenomes; (3) produces high-quality reference genomes that enable the discovery of novel genomic structures and variation; (4) actively promotes open source and open data initiatives in genomics; (5) fosters interdisciplinary collaboration between the computational, biological, and medical sciences to advance global health.

## **Metabolic, Cardiovascular and Inflammatory Disease Genomics Branch - HN45L**

(1)Plans and conducts research into the genomic and social determinants of complex diseases with particular attention to diseases (e.g., diabetes, hypertension, heart and kidney disease, scleroderma, and podoconiosis) that are disproportionately distributed by ethnicity and geography; (2)designs and analyzes genomic data to document and interpret the non-random distribution of human genetic variation with the goal of understanding human history and how these histories inform disease distribution and etiology; and (3) facilitates the development of large genomic initiatives in populations currently under-represented in the genomic revolution.

## **Center for Research on Genomics and Global Health - HN45L2**

Advances research in the role of culture, lifestyle, genetics and genomics in disease etiology, differential susceptibilities to disease and variable drug response at the individual and population levels; and develops genetic epidemiology models that explore the patterns and determinants of common complex diseases in populations in the US and other human populations around the world.



### **Inflammatory Disease Section - HN45L3**

Utilizes genetic and genomic strategies to elucidate the molecular basis, pathophysiology, and treatment of human inflammatory diseases. Major areas of concentration include (1) the genetics of Mendelian inflammatory diseases, including known disorders such as familial Mediterranean fever and currently unexplained phenotypes; (2) analysis of genetically complex inflammatory disorders, such as Behçet's disease and scleroderma; (3) studies of disease pathogenesis, using both patient samples and animal models; (4) natural history and targeted therapeutic studies in patients; and (5) integrated studies of patients at the NIH Clinical Center with laboratory investigations.

## **Cardiovascular Disease Section - HN45L4**

(1) Specializes in multi-disciplinary translational research, in concordance with the traditional genetic-to-phenotype imputations; (2) utilizes a combination of noninvasive cardiovascular imaging, biomedical informatics, systems biology, and genomic epidemiology to conduct very large scale data analyses; and (3) pursues understanding of the relationships and interdependencies between clinical phenotypes, human behavior, biosurveillance, molecular, genomic and social determinants on gene expression and their contribution to health disparities in cardiovascular disease.

## **Genomics of Autoimmune Rheumatic Disease Section - HN45L6**

Advance precision health and medicine approaches to the prevention, diagnosis and treatment of autoimmune rheumatic disease through application of genomic, epidemiologic and computational approaches

## **Medical Genomics and Metabolic Genetics Branch - HN45M**

(1) Plans and conducts research investigating the relationship of genetic variation to health and disease; (2) uses genetic, genomic, and metabolic approaches to understand normal and abnormal physiology; (3) develops and tests innovative approaches to the diagnosis and management of disease; and (4) facilitates the development of early career physician-scientists.

## **Precision Genomics Section - HN45M2**

1) Develop and evaluate cutting edge genomic tools for early diagnosis of diseases to improve management and treatment of genetic diseases 2) Develop novel approaches to the diagnosis and treatment of mosaic genetic disorders.

#### **Molecular Genetics Section - HN45M4**

(1) Conducts research to uncover the genetic basis of monogenic and polygenic diseases; (2) develops better diagnostic and therapeutic approaches for these diseases; and (3) conducts research studies on progeria and type 2 diabetes mellitus, including the use of genomic technologies to identify disease-associated variations and elucidate their functional consequences.

**Genetic Counseling Training Program - HN45M7**

**Train future leaders in genetic counseling research, policy, and practice.**

**Precision Health Informatics Section - HN45M9**

Advance precision health through the development and evaluation of genomic, phenomic, and informatics tools to improve the diagnosis and management of disease.



**Cohort Data Analytics Core - HN45MA**

Support IRP investigators in answering biologic and clinical questions using large clinical and biobank datasets.

**Reverse Phenotyping Core - HN45MB**

1) Develop a cohort of sequenced individuals who are available for post-hoc phenotyping. 2) Support intramural investigators in using this cohort and perform selected phenotyping projects.

### **Prenatal Genomics and Therapy Section - HN45MC**

(1) Uses genomic tools and pathways analyses to identify structural and functional differences in embryonic development resulting from aneuploidy; (2) performs preclinical studies on cells and animal models to evaluate effects of prenatal therapeutic interventions with an emphasis on brain development and cognition; (3) integrates pre-and post-natal genomic data to understand underlying biologic mechanisms for unusual prenatal test results; and (4) considers ethical dilemmas arising from prenatal genomic testing such as maternal incidental findings.

## **Childhood Complex Disease Genomics Section - HN45MD**

Designs and implements genomic research studies to better understand the pathophysiology of multifactorial diseases of childhood. Leverages population genetics to understand the impact of genetic variation on disease in populations of diverse ancestries.

## **Genomic Medicine Implementation Unit - HN45ME**

Conducts research to evaluate the effectiveness and impact of various methods for using new genomic knowledge to improve clinical care.

## **Translational and Functional Genomics Branch - HN45N**

(1) Plans and conducts research to explore the full functional potential of the human genome; (2) investigates the genetic, epigenetic and metagenomic basis of human disorders; (3) applies genetic and genomic approaches to model organisms to understand the molecular etiology of human disorders; (4) catalyzes technology development in genetics and computational genomics; and (5) translates laboratory findings to improved diagnoses and therapeutics for human disorders.

## **Microbial Genomics Section - HN45N2**

(1) Performs foundational studies to characterize the diversity of microbes (bacteria, fungi, other small micro-organisms) that reside on human skin; (2) characterizes changes in the human microbiome associated with skin disorders and transition through life stages; (3) examines functional host-microbial (commensal and pathogenic) interactions; (4) develops and utilizes genomic sequence technology to identify emerging hospital bacterial pathogens; and (5) tracks transmission and evolution of hospital-associated bacterial pathogens incorporating epidemiologic and genomic information.

### **Oncogenesis and Development Section - HN45N3**

(1) Identifies and studies genetic, genomic, and epigenetic alterations involved in the pathogenesis of leukemias and other human blood diseases; (2) investigates genetic, genomic, and epigenetic involvements, including chromosome rearrangements, point mutations, and altered expressions, in the initiation and progression of these human diseases; and (3) uses animal and cell-based models to study these disease-related genes and their encoded proteins in diverse biological processes and disease pathogenesis, and to develop novel therapeutics including small chemical and biological treatments.



## **Developmental Genomics Section - HN45N4**

(1) Utilizes a variety of modern molecular methods to determine the functions of genes and how they relate to human genetic disease; (2) emphasizes zebrafish as a model organism and systematic mutagenesis to identify the function of new genes important in human disease; (3) focuses on isolating embryonic mutations of candidate human disease genes and mutations that disrupt tissue regeneration; (4) develops large-scale, high-throughput gene inactivation technology and analyzes gene functions on a genome-wide scale; and (5) annotates large scale functional regulatory regions of the zebrafish genome.

## **Genomic Functional Analysis Section - HN45N5**

(1) Plans and conducts research investigating functional entities in the human genome; (2) characterizes the relationship of the aforementioned elements to those in non-human organisms with respect to sequence alignment and phylogeny; (3) aims to analyze the relationship between mutations in functional elements and correlations with human genetic diseases; (4) integrates epigenomic and genomic data with transcriptomic outcomes; and (5) provides genomics analysis tools to the community at large for the betterment of computational genomics as a field of study.

## **Zebrafish Core - HN45N6**

(1) Provides resources, training and services to analyze gene function and generate disease models using zebrafish for research; (2) provides microinjection services to perform transient knockdown and over-expression of desired genes; (3) performs microinjections and founder screening service to generate transgenic zebrafish lines; (4) provides services to generate genetic mutants using ZFNs, TALENs and/or CRISPR-cas9 nucleases; (5) consults with researchers in planning zebrafish experiments; (6) provides training in breeding and imaging to analyze phenotypes of mutant, morphant and transgenic zebrafish; and (7) provides chemical libraries and training to perform drug screening using zebrafish.

## **NIH Undiagnosed Diseases Program - HN45P**

(1) Reviews medical records of applicants with undiagnosed disorders who seek a definitive diagnoses; (2) investigates mysterious human diseases by intensive and extensive phenotyping through week-long Clinical Center admissions, single nucleotide polymorphism array and exome sequencing analyses, and functional studies of candidate gene variants; (3) develops new methods for exome sequence analysis, such as the diploid aligner and variant filtering programs; (4) serves as an integral part of the newly established Undiagnosed Diseases Network and its Data Coordinating Center, providing experience and expertise on best practices for an undiagnosed diseases program; (5) develops algorithms for diagnostic evaluations, data submission to dbGaP, and other databases, and privacy protection for patients with undiagnosed diseases; (6) develops and conducts one-week training sessions and year-long fellowships in the analysis of Next Generation sequencing data of patients with rare and new diseases; and (7) defines and discovers new human disorders and disease mechanisms.

## **Metabolic Medicine Branch - HN45Q**

(1) Plans and conducts research investigating the relationship of heritable disorders in intermediary metabolism to health and disease; (2) plans and conducts research on gene therapy for disorders of intermediary metabolism; (3) uses metabolic, metabolomic, cell biologic, genetic, and genomic approaches to understand normal and abnormal physiology; (3) develops and tests innovative approaches to the diagnosis and management of metabolic disease; and (4) facilitates the development of early career physician-scientists.

## **Metabolic, Infection and Immunity Section - HN45Q2**

**Designs and conducts clinical and laboratory research in order to: (1) understand metabolic perturbations brought about by immune activation; and (2) demonstrate the role of intermediary metabolism in the differentiation, activation and function of immune cells.**

### **Organic Acid Research Section- HN45Q3**

**(1) Plans and conducts research to diagnose and study patients with organic acidemias; (2) develops animal models of organic acidemias; (3) develops new therapies for organic acidemias; and (4) develops new assays for diagnosing and for evaluating the effectiveness of therapies for organic acidemias.**

## **Skeletal Genomics Unit - HN45Q4**

**The goals of the unit are to use a translational research approach to understand the etiology, pathophysiology, diagnosis, and management of individuals with heritable disorders of skeletal biology. Specifically, to conduct clinical research to uncover the clinical spectrum and genomic basis of selected skeletal dysplasias, develop animal and cell models to understand the pathomechanism of these disorders, and develop targeted treatment approaches.**



## **Division of Genome Sciences - HN47**

(1) Plans, directs, and facilitates multi-disciplinary research to understand the structure and function of genomes and their implications for biology and disease etiology, through research and training grants, research center grants, and contracts; (2) assesses the need for research and research training in genome sciences and related areas; (3) determines program priorities in genome sciences and related areas and recommends funding levels; (4) prepares reports and analyses to assist Institute staff and advisory groups in carrying out their responsibilities; (5) collaborates with the other NHGRI extramural research Divisions to establish a balance of resources, personnel, research and training budgets to achieve NHGRI goals; (6) provides expert advice to the Director, NHGRI, on various aspects of genome sciences; and (7) collaborates with the other NHGRI Divisions, other NIH ICs, and other agencies and entities, nationally and internationally, and maintains an awareness of research efforts in relevant program areas.

## **Division of Genomic Medicine - HN48**

(1) Plans, directs, and facilitates multi-disciplinary research to identify genetic contributions to human health and disease and use them to improve diagnosis, treatment, and prevention through research and training grants, research center grants, and contracts; (2) determines program priorities in genomic medicine and related areas and recommends funding levels; (3) assesses the need for research and research training in genomic medicine and related areas; (4) prepares reports and analyses to assist Institute staff and advisory groups in carrying out their responsibilities; (5) collaborates with the other NHGRI extramural research Divisions to establish a balance of resources, personnel, research and training budgets, to achieve goals; (6) provides expert advice to the Director, NHGRI, on various aspects of genomic medicine; and (7) collaborates with the other NHGRI Divisions, other NIH ICs, and other agencies and entities nationally and internationally, and maintains an awareness of research efforts in relevant program areas.

## **Division of Genomics and Society - HN49**

(1) Serves as a nexus for activities related to society that arise from genetics and genomics research; (2) collaborates widely with other NHGRI research divisions, other ICs, and other agencies to establish and achieve NHGRI goals related to genomics and society; (3) provides expert advice to the NHGRI Director on various aspects of the societal, educational, and policy issues raised by genomics; (4) maintains an awareness of research efforts in relevant program areas; and (5) produces scholarly work and informational products related to the issues relevant to the division.

## **Ethical, Legal, and Social Implications Research Branch – HN492**

(1) Plans, directs, and facilitates multi-disciplinary research to understand the ethical, legal, and societal implications of genomic advances through research and training grants, research center grants, and contracts; (2) manages a portfolio of research programs focused on anticipating the societal impact of genomics on disease etiology, prevention, treatment, and management; (3) assesses the need for research and research training in societal issues raised by genomics and related areas, determines program priorities in societal issues raised by genomics and related areas, and recommends funding levels for such research; (4) prepares reports and analyses to assist Institute staff and advisory groups in carrying out their responsibilities; and (5) provides input and expertise about a wide range of activities carried out by the Division of Genomics and Society.

## **Education and Community Involvement Branch - HN493**

(1) Advises the Institute Director and senior staff on a broad range of issues regarding public education and community involvement matters; (2) develops, implements, and evaluates public education programs; (3) conducts and coordinates public education initiatives; (4) develops community involvement programs to engage a broad range of the public in understanding genomics and accompanying ELSI issues; (5) conducts, coordinates, and evaluates community involvement initiatives; and (6) acts as the liaison between the Institute and community-based organizations.

## **Policy and Program Analysis Branch - HN494**

(1) Advises the Institute Director and senior staff on a broad range of policy and program matters; (2) develops, implements, and reports analyses of Institute programs; (3) analyzes and tracks legislation relevant to the mission of the Institute and makes recommendations for legislative proposals; and (4) conducts and coordinates policy analysis related to the ethical, legal and social implications of human genomic and genetic research and develops programs to address these issues.

## **Division of Extramural Operations - HN4A**

(1) Provides leadership and advice to the Director on implementing and coordinating extramural research grant, contract, and training program operations and policies; (2) provides a full range of activities that empower the scientific divisions in implementing and coordinating cross-cutting, multi-disciplinary activities in the mission areas of NHGRI; (3) represents NHGRI on overall NIH extramural and collaborative program policy committees and coordinates such policies within the institute; (4) manages the activities of the National Advisory Council for Human Genome Research; (5) develops and provides extramural staff training and enhances communication across the Institute regarding standardized approaches policies, methods, and procedures; (6) manages institute-wide extramural services and activities; (7) provides oversight and direction for the initial scientific merit review of research, resource, and training applications and proposals assigned to the Institute; (8) provides oversight and direction for grants management activities, ensuring that all awards are made in accordance with applicable statutes, regulations, and policies; (9) provides data, reports and analyses to assist NHGRI staff and advisory groups in carrying out their responsibilities; and (10) maintains effective relationships with other NIH institutes and divisions, the research community, other federal agencies, and professional societies.

## **Scientific Review Branch - HN4A2**

(1) Plans and administers scientific review activities, including the organization and management of Initial Review Groups (IRGs), constituted to review NHGRI's extramural grants, cooperative agreements, and contract proposals; (2) performs initial scientific and administrative review of research, research center, program project, conference, and research training grant applications, cooperative agreements, and contracts; (3) plans and directs site-visits and IRG meetings; (4) establishes review criteria and standards for the IRGs and provides technical leadership to the review process; and (5) prepares summary statements and provides pertinent information concerning the review of applications to the Council.



### **Grants Administration Branch - HN4A3**

(1) Develops and implements Institute policies on the business management aspects of grant and fellowship programs; (2) develops and implements guidelines, procedures and internal controls to ensure proper and continuing implementation of NHGRI, NIH, PHS, and DHHS policies; (3) administers the Institute's grant programs including negotiation of awards, post award approvals, audits, and closeout; (4) participates in awarding functions, authorizing awards and changes to active awards; and (5) prepares and distributes grant-related materials required by the NHGRI Advisory Council.

**Division of Management - HN4B**

(1) Plans and directs administrative management functions including administrative management services, management analysis and evaluation, financial management, information technology and human resources; (2) advises the senior leadership on developments in administrative management and their implications and effects on program management; and (3) coordinates administrative management activities in support of their programs.

## **Administrative Services Branch - HN4B2**

1) Negotiates for and secures the resources and services needed to run the Office of the Director and the Division of Extramural Research; (2) manages the Institute's RMS Budget, monitoring and tracking obligations and expenditures to assure the availability of funds and prevent budget deficiencies; (3) manages the personnel, procurement, property, space, travel, and other administrative functions as necessary to ensure the efficient and effective implementation and operation of programs; (4) advises NHGRI staff of administrative policies and procedures; (5) performs analytical studies related to the administrative organization, processes and procedures of the Institute and establishes effective administrative controls; (6) represents the Institute on NIH committees or task groups; (7) and serves as liaison and contact point for the NHGRI with the central services areas of NIH, other agencies, and outside businesses.

### **Financial Management Branch - HN4B3**

(1) Plans, directs, coordinates the financial management activities of the Institute; (2) serves as the principal advisor to the Institute Director, Deputy Director, and Associate Director for Management on the management of the financial and personnel resources of the Institute; (3) manages the Institute budget process which includes providing guidance to Institute staff on budget preparation and management; coordinating the preparation and submission of the Institute's budget to NIH, OMB, and the President; explaining and defending the budget; and monitoring budget execution; (4) administers the Institute's budgetary, accounting, and funds control systems assuring that they are compatible with NIH and Departmental accounting systems and provide Institute management and staff with reliable accounting, financial management, and personnel resource utilization data; (5) performs analysis and provides information on the historic, current, and prospective financial status of the Institute for briefing and reporting purposes; and (6) participates in budget hearings by preparing background, briefing, and source material for the Institute Director and others appearing before committees or examiners, by attending budget hearings as part of the Institute contingent on an as-needed basis, and by coordinating responses to questions concerning the financial management of the Institute from the Congress and others.

## **Information Technology Branch - HN4B4**

(1) Provides technical leadership and advice to all levels of Institute management in order to obtain maximum utilization of current ADP resources and advancements in the field of information systems technology and communications; (2) determines requirements, designs, and implements and coordinates the Institute's management information systems which collect, maintain, and report various types of administrative information; (3) advises the NHGRI Director, Deputy Director, Associate Director for Management, and other Institute staff on the technological and policy impact and implications of developments in information systems and related fields within and outside the government; (4) coordinates staff activities with those of contractors, other components of NIH, and other Federal and non-Federal data processing agencies; (5) provides user support, including training, in LAN/information systems capabilities, programs, and procedures.

### **Intramural Administrative Management Branch - HN4B5**

Plans, directs, coordinates, and provides comprehensive administrative and management support services for the Division of Intramural Research; (2) provides technical and advisory services in financial management, human resources, procurement, facility management, travel services, property management and other administrative functions, as necessary, to ensure the efficient and effective implementation and operation of programs; and (3) develops policies, guidelines, and procedures on matters relating to administrative management and disseminates to relevant staff.

## **Acquisition and Logistics Section - HN4B52**

(1) Plans, directs and coordinates acquisition activities for the intramural staff using efficient, timely and appropriate methods; and (2) plans, directs, and coordinates logistical support for the DIR including management of DIR property activities.

## **Management Analysis and Workforce Development Branch - HN4B6**

(1) Coordinates NHGRI-wide administrative/management initiatives including organizational analysis, workflow management, delegations of authority, policy and procedure analysis, and privacy and records management; (2) develops, executes and maintains an NHGRI-wide risk management program; (3) coordinates/directs cross-functional teams and study groups in finance, IT, and general administration; and (4) coordinates workforce management and professional development activities.



## **Ethics Branch - HN4B7**

(1) Administers a comprehensive NHGRI ethics program that reflects statutory responsibilities and integrity in service to the public; (2) develops and recommends policies and procedures related to employee standards of conduct, financial interests and disclosure, outside activities, gifts administration, official duty activities, sponsored travel, and procurement integrity; (3) administers the annual public and confidential financial disclosure process including reviewing and certifying financial disclosure reports and reports of holdings in substantially affected organizations, and develops new employee ethics agreements; (4) reviews and approves requests for outside activities, official duty requests, and sponsored travel or related gifts for conformance with regulations and policies; (5) provides advice and assistance to employees regarding the application of the ethics laws, regulations, and policies; (6) develops and provides NHGRI ethics training (7) provides liaison to the HHS Office of the General Counsel, the Office of Government Ethics, the NIH Ethics Office, other agencies, and outside organizations as needed; (8) provides advice to the Office of the Director regarding conflict of interest of individuals involved in the conduct of biomedical research, including Government employees, advisory committee members, and non-Government employees such as peer reviewers, Data Safety Monitoring Board (DSMB) members, and members of working groups; (9) reviews: (a) procurements over one million dollars involving justification of other than full and open competition; (b) gifts acceptance under NHGRI statutory authorities; (c) memoranda of understanding of public private partnership proposals and co-sponsorships with non-federal entities; and (d) conflicts concerning prior employment ties to academic institutions and private entities; (10) identifies management issues requiring action by the Office of General Counsel such as copyright, intellectual property, contract, or personnel authorities; and (11) reviews clinical protocols, conducts conflict of interest (COI) analysis to confirm no COI exists between investigators' official duties on the protocol and their personal or imputed financial interests.