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The research interests of Dr. Boerwinkle encompass the genetic analysis of the common chronic diseases in humans, including coronary artery disease, hypertension, and non-insulin dependent (type II) diabetes.

Dr. Boerwinkle received his B.S. in Biology from the University of Cincinnati in 1980, an M.A. in Statistics (1984), and M.S. and Ph.D. in Human Genetics (1985) from the University of Michigan, Ann Arbor where he served as Senior Research Associate in the Department of Human Genetics from 1985-1986. He joined the University of Texas-Houston Center for Demographic/ Population Genetics in 1986 as a Research Assistant and became Assistant Professor in the same year. In 1991 he joined the Department of Human Genetics at the School of Public Health, University of Texas-Houston Health Science Center as Associate Professor, in 1996 was promoted to Professor, and in 1997, Director of the Human Genetics Center. He became a faculty member of the Institute of Molecular Medicine in 1996 and became Professor and Director of the Research Center for Human Genetics.

Dr. Boerwinkle is a member of the American Diabetes Association and the American Society of Human Genetics. The research interests of Dr. Boerwinkle encompass the genetic analysis of common chronic diseases in humans, including coronary artery disease, hypertension, and non-insulin dependent (type II) diabetes. This work includes localizing genes which contribute to disease risk, identification of potentially functional mutations within these genes, testing these candidate functional mutations in experimental systems, defining the impact of gene variation on the epidemiology of disease, and determining the extent to which these genes interact with environmental factors to contribute to disease risk. Activities include both statistical analysis and laboratory work. A large part of Dr. Boerwinkle's current research effort consist of localizing genes contributing to disease risk using modern genome-wide mapping methods. Success depends on keeping up with the latest genomic technical advances. The laboratory is set-up and operating as a high through-put sequencing and genotyping facility in which speed, accuracy and efficiency are monitored continuously. However, we are constantly seeking out more efficient methods to collect and manage genetic information.

Dr. Boerwinkle and colleagues have completed the world's first genome-wide analyses for a variety of CAD risk factors, including diabetes and hypertension. These investigations have lead to the identification of novel susceptibility genes in both cases. Dr. Boerwinkle is particularly interested in methods for identifying potentially functional mutations within a gene region. This seemingly simple objective is made difficult because the functional mutations are expected to have small effects and are imbedded in a sea of silent genetic variation. Once nearly all of the variation is catalogued directly by DNA sequencing, individuals are genotyped for each variable site. Both novel and traditional statistical methods are applied to relate the array of genetic information to a wealth of phenotypic data. This algorithm generates "candidate functional mutations" that are then tested in an in vitro or mouse model system. Once a functional mutation has been identified, Dr. Boerwinkle's group evaluates the ability of the variable site to predict the onset of disease (e.g. myocardial infarction or stroke) above and beyond traditional risk factors. This work is carried out as part of multiple prospective studies of cardiovascular disease and its risk factors in tens of thousands of individuals representing the major American ethnic groups.

Finally, he is working on experimental designs for studying genotype by environment interaction in humans. In particular, we are working on the extent to which interindividual variation in lipid lowering and anti-hypertensive medications are influenced by genetic factors. The practical objective of the research is to use genetic information to identify individuals at increase risk of disease and to design more efficacious interventions. Genetic studies are defining, at the molecular level, novel mechanisms of disease risk, onset and progression. Dr. Boerwinkle and collaborators address the localization of genes which contribute to disease risk in cardiovascular diseases, hypertension and diabetes. The methodology used involves screening of families having the disease and linking the presence of disease with known markers of the human genome. In this manner, the genomic region in which relevant mutations are located can be mapped and the relevant DNA sequenced. By assessing the structural change the mutation may have caused in the gene product (protein), it is possible to infer how it may affect biological function. In order to determine experimentally whether a mutation is functional, it is necessary to introduce the mutated gene into an animal, usually a mouse, and assess its biological effects on the animal's phenotype.

Dr. Boerwinkle has participated in multiple notable discoveries since joining the Institute. Only two will be highlighted here. First, Dr. Boerwinkle's group has completed the first ever genome-wide search for genes contributing to inter-individual blood pressure levels. This initial effort has lead to the identification of an important gene (an adrenergic receptor) which influences blood pressure levels and the risk to hypertension. This is the first time that such a genome-wide approach has led to the identification of a susceptibility gene to a major cardiovascular disease risk factor. Second, Dr. Boerwinkle has participated in similar efforts to identify genes contributing to the risk of developing non-insulin dependent (type II) diabetes. In this case, however, there were no genes in the region that were suspects for the disease. A team of collaborating investigators have painstakingly characterized the genetic region and identified the mutated gene (in this case a protease). This is the first time that anyone has ever positionally cloned a gene contributing to any common chronic disease. This work is of obvious potential clinical importance. It may lead to improved prediction of those at increased risk of disease and the design of more efficacious intervention strategies. The technologies and information from the human genome project provide new tools for lessening the burden of ill-health. Dr. Boerwinkle's accomplishments in developing an internationally recognized team of investigators targeting the genetics of cardiovascular disease and its risk factors ensures a productive future and further discoveries.