

# Anthropological and Primate Genetics

Southwest Foundation for Biomedical Research &  
American Association of Anthropological Genetics

18-20 November 2004  
SFBR, San Antonio, TX



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## *Thursday, 18 November*

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8:00-8:30a	CONTINENTAL BREAKFAST, Packet Pick-up (Breakfast sponsored by M&A Technology)	
8:30-8:45a	Welcome & Introduction	Lorena M. Havill, Ph.D.
8:45-9:30a	General Concepts: Family Studies	Harald Göring, Ph.D.
9:30-10:15a	Quantitative Genetics	Laura Almasy, Ph.D.
10:15-10:30a	BREAK	
10:30-11:15a	Linkage	Harald Göring, Ph.D.
11:15a-12:00p	Association	Laura Almasy, Ph.D.
12:00-1:30p	LUNCH	
1:30-2:15p	Pleiotropy	Michael C. Mahaney, Ph.D.
2:15-3:00p	Genotype-by-Environment Interaction	Michael C. Mahaney, Ph.D.
3:00-3:15p	BREAK	
3:15-5:30p	Genetic Analyses in <i>SOLAR</i> : Heritability, Linkage, and Bivariate Analyses	Tom D. Dyer, Ph.D. Charles Peterson
5:30p	Group Picture	
6:00-9:00p	WELCOME RECEPTION (Reception sponsored in part by Applied Biosystems)	

## *Friday, 19 November*

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8:00-8:30a	CONTINENTAL BREAKFAST (Breakfast sponsored by Purina Mills LabDiet)	
8:30-8:45a	Announcements	
8:45-9:30a	Genetics Core Lab & the Logistics of Genotyping	Shelley Cole, Ph.D.
9:30-10:15a	Primate Gene Mapping	Jeff Rogers, Ph.D.
10:15-10:30a	BREAK	
10:30-11:15a	Statistical Considerations and Pedigree Reconstruction with Forensic Quality Samples	Amanda Vinson, Ph.D. Candidate
11:15a-12:00p	Genetics of Baboon Skeletal Biology	Lorena M. Havill, Ph.D.
12:00-1:30p	LUNCH	
1:30-2:15p	Genetics of Complex Traits in the Baboon Cranium	Richard Sherwood, Ph.D.
2:15-3:00p	Genetics of Bone Accrual During Childhood (FELS)	Dana Duren, Ph.D.
3:00-3:15p	BREAK	
3:15-4:00p	Genetics of Susceptibility to Helminthic Infection in the Jirils of Nepal	Sarah Williams-Blangero, Ph.D.
4:00-4:45p	GOCADAN: Practicalities of Fieldwork and Data Collection	Sandy Laston, Ph.D.
4:45-5:30p	Mexican-American Program Project: Incorporation of Household Effects	Diane Warren, Ph.D.

*Saturday, 20 November*

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| 8:00-8:30a    | CONTINENTAL BREAKFAST<br>(Breakfast sponsored by Sigma Solutions)    |
| 8:30-10:30a   | Tour of Core Lab and Genomics Computing Center                       |
| 10:30a-12:00p | “How to” Question & Answer Session with Experienced<br>Investigators |

## *Workshop Speakers*

Laura Almasy, Ph.D.

*Dr. Almasy is an Associate Scientist in the Department of Genetics, SFBR.*

The central theme of my research is development and application of statistical genetic methods for the localization, characterization and identification of genes influencing common complex diseases and related quantitative risk factors. In collaboration with others at SFBR, I work on variance component methods for linkage and association studies, implemented in our SOLAR software package. On the applied side, I am involved in studies related to thrombosis, heart disease, diabetes, and obesity as well as studies of schizophrenia, alcoholism, and other drug addiction. These projects include quantitative risk factors ranging from simple anthropometrics such as waist circumference to measures of brain wave activity and cognition. In most of these studies, we are currently using genome-wide linkage analyses to localize genes of interest. In a few cases, we have moved beyond linkage to the identification of specific genes and the functional variants in them.

Shelley Cole, Ph.D.

*Dr. Cole is an Assistant Scientist in the Department of Genetics, SFBR and is the Director of the Genetics Core Laboratory.*

The Department of Genetics has initiated several collaborative genome-wide searches to identify new genes that influence common diseases such as atherosclerosis, diabetes, and obesity. The Genetics Core Laboratory is responsible for the generation of the high-quality, high-throughput microsatellite genotypes used for over eight of these genome searches. As a goal towards identifying the new genes responsible for these diseases, we also perform more extensive molecular genetic analyses including high-throughput resequencing and SNP discovery in the chromosomal regions where linkages to disease-related phenotypes are uncovered. In addition, we are using gene expression as a novel phenotype in a genome scan in baboons to identify novel genes involved in obesity, diabetes, and metabolic syndrome phenotypes.

Dana L. Duren, Ph.D.

*Dr. Duren is an Assistant Professor at the Lifespan Health Research Center, Wright State University School of Medicine.*

My current research focuses on human bone and joint health, including precursors to osteoarthritis and the genetic epidemiology of skeletal maturation and childhood cortical bone accrual, conducted in collaboration with Drs. Bradford Towne and Roger Siervogel. Because peak bone mass attained by early adulthood is a strong predictor of osteoporosis risk, we are investigating the genetic underpinnings of bone accrual throughout childhood in participants from the Fels Longitudinal Study. Approaches utilized in this research include a variance components-based method (SOLAR) to estimate the heritability of bone traits, genetic and environmental correlations between traits, and, in collaboration with Drs. Blangero, Dyer, and Cole, linkage analyses to identify regions of the genome that are involved in childhood cortical bone mass.

Thomas D. Dyer, Ph.D.

*Dr. Dyer is a Staff Scientist in the Department of Genetics, SFBR.*

The major emphasis of my work is in computational genetics. My duties include writing software for genetic analysis, developing procedures to facilitate the use of the Genomics Computing Center compute ranch, overseeing departmental system administration activities, and providing technical support in the areas of numerical analysis and statistical genetics. As a member of the SOLAR development team, my focus has been on identity-by-descent (IBD) calculations, allele frequency estimation, simulation, analysis of SNP data, and power calculations. In addition, I have assisted in the organization and presentation of several SOLAR courses and have been involved in the preparation of simulated data for several past Genetic Analysis Workshops.

Harald H.H. Göring, Ph.D.

*Dr. Goring is an Assistant Scientist in the Department of Genetics, SFBR.*

My research focuses on the statistical localization, identification and characterization of human disease genes. I seek to develop and implement novel or improved statistical approaches for the detection of genotype-phenotype correlations in related individuals. I am also interested in the design of genetic studies and its impact on the power to detect and ability to characterize genetic risk factors. In parallel to this methodological work, I am involved in a number of applied gene mapping studies in collaboration with investigators at SFBR and other research institutions. These studies are focused on a large variety of human traits, including rare “mendelian” diseases and risk factors for common “complex” traits. I am particularly interested in understanding the genetic factors that determine an individual’s susceptibility to infectious diseases.

Lorena M. Havill, Ph.D.

*Dr. Havill is a Staff Scientist in the Department of Genetics, SFBR.*

I study the genetics of osteoporosis, a complex disease resulting from a number of genetic and environmental factors affecting bone mass and architecture. My research, conducted in collaboration with Drs. Mahaney and Rogers, is concentrated on osteoporosis-related phenotypes that have been linked to osteoporosis risk in humans, including bone mineral density, serum measures of bone formation, and microscopic bone remodeling processes. I employ maximum-likelihood-based variance decomposition methods to assess heritability, test for pleiotropic effects on related phenotypes, and test for genotype-by-“environment” interaction. In this case, “environment” may refer to the individual’s age, sex, or other covariate (e.g. reproductive history). The ultimate goal of this research is to improve our understanding of factors contributing to variation in the maintenance and repair of bone at the microstructural level and, by extension, susceptibility to osteoporosis and other age-related pathologies in bone.

Sandra Laston, Ph.D.

*Dr. Laston is a Staff Scientist in the Department of Genetics, SFBR.*

My research at SFBR includes projects that examine the relationship of genetics and environment to CVD (cardiovascular disease) in Alaskan Eskimos and Native Americans. CVD is increasing rapidly in Eskimos and other Alaska Natives as their traditional diets and lifestyle become Westernized. The GOCADAN project is investigating CVD in this relatively isolated, homogeneous population which provides an excellent opportunity to identify genetic factors that interact with lifestyle and diet to increase susceptibility to CVD. My specific interests include field logistics, household surveys and collection of family information, construction and cleaning of pedigrees, and genotype cleaning. My background in Medical Anthropology and fieldwork in developing countries have been valuable for our household visits in Eskimo villages.

Michael C. Mahaney, Ph.D.

*Dr. Mahaney is a Scientist in the Department of Genetics, SFBR.*

Analyzing pedigree data both from humans and nonhuman primates, my primary research activities focus on statistical genetic analyses of normal quantitative variation in phenotypes associated with 1) risk of cardiovascular disease, particularly biomarkers of oxidative stress and inflammation; 2) age-related changes and pathology in bone; and 3) dental development and evolution. The objectives of these activities are to detect, identify, and characterize the action of genes that contribute to normal variation in the complex phenotypes under study. I am most interested in gene-by-environment interactions that influence patterns of variation in traits and detecting pleiotropic interactions among traits. These activities are predominantly NIH-funded and entail collaborations with colleagues both within and without SFBR. Additionally, I am involved in collaborative efforts with colleagues at the Southwest National Primate Research Center to develop and deploy a web-based Primate Genomics Database that will collect and share nonhuman primate genomics data in support of comparative genomics research.

Charles P. Peterson

*Mr. Peterson is a Senior Systems Analyst at SFBR.*

I enhance, maintain, document, and support the SOLAR program used in genetics research. I designed the software architecture and interface of the first public release of SOLAR (downloadable since 1998), which permits interactive exploratory analysis and Tcl-based scripting, and continue extending it to facilitate and automate ever more complex genetic and environmental modeling. I integrate the work of others into this program. I also develop parallelized applications of SOLAR which run on the Genomics Computing Center compute ranch, especially for Quantitative Trait Nucleotide analysis.

Jeffrey Rogers, Ph.D.

*Dr. Rogers is a Scientist in the Department of Genetics, SFBR, and the Group Leader for Genetics within the Southwest National Primate Research Center.*

There are three different but related research activities currently underway in my laboratory. First, we are constructing genetic linkage maps of the genomes of nonhuman primates. In collaboration with other researchers at SFBR, we continue to add new polymorphic loci to the genetic linkage map of the baboon genome. In addition, my laboratory recently completed a 10 centimorgan linkage map of the rhesus macaque genome, using DNA and pedigree information from animals housed at SNPRC and at the Oregon National Primate Research Center. Our second major focus is the genetic analysis of individual variation in primate behavior and neurobiology. In this area, we are working with collaborators at SFBR and other institutions to conduct genome scans to locate quantitative trait loci that influence temperament, personality and neurotransmitter metabolism in both the baboons at SFBR and the rhesus macaques in Oregon. We are now adding neuroimaging to this project. Third, we perform paternity testing and pedigree analysis for a number of breeding colonies of nonhuman primates, in order to assist those colonies in building large multigeneration pedigrees that will be valuable tools for the genetic analysis of complex traits in those populations.

Richard J. Sherwood, Ph.D.

*Dr. Sherwood is an Adjunct Associate Professor of Community Health, Lifespan Health Research Center, Wright State University.*

My interests include comparative anatomy of the craniofacial complex in humans and non-

human primates. My current research projects include examination of the genetic architecture of complex craniofacial traits in humans and baboons. For these projects, I am collaborating with personnel from the LHRC, including Drs. Brad Towne and Dana Duren, and with Drs. Michael Mahaney and John Blangero of the SFBR. Maximum-likelihood variance decomposition methodology is used to estimate heritability for each trait and genetic correlation between traits. The ultimate goal of this work is to better characterize the genetic underpinnings of normal craniofacial variation.

### Amanda Vinson

*Ms. Vinson is a Senior Research Assistant in the Department of Genetics, SFBR and a Ph.D. Candidate in the Department of Ecology, Evolution, and Behavior at the University of Minnesota.*

My dissertation research focuses on using neutral genetic data to test hypotheses of relatedness patterns and classic population genetic expectations in species that exhibit complex social structure. As a model for these processes, I am using savannah baboon populations at Gombe National Park and Mikumi National Park, Tanzania. In general, I am interested in exploring the interface between the molecular genetic basis of traits under selection and mathematical population genetic theory.

### Diane M. Warren, Ph.D.

*Dr. Warren is a Postdoctoral Scientist in the Department of Genetics, SFBR.*

My research focuses on the characterization of blood coagulation phenotypes identified as risk factors for cardiovascular disease. As with cardiovascular disease itself, these are complex phenotypes influenced by genetic and environmental factors, and interactions among these factors. My investigations, conducted in collaboration with Dr. Almasy and others, make use of data primarily obtained from Mexican American families living in the San Antonio area. My research interests also include the biological outcomes of contact between human and animal populations, particularly as they relate to the skeletal biology and paleopathology of domesticated species management.

### Sarah Williams-Blangero, Ph.D.

*Dr. Williams-Blangero is a Scientist at SFBR, and is Chair of the Department of Genetics.*

My research efforts are focused on the genetics of susceptibility to infectious diseases. Utilizing statistical genetic approaches and genome scanning techniques, we are assessing the genetic components of a number of parasitic diseases and of immune function. The genetic determinants of normal immune function are being studied in the chimpanzee, an important model for infectious disease research. The genetic components of susceptibility to parasitic diseases are being investigated in two large-scale human population studies. The intestinal worm infections that affect a quarter of the world's population (hookworm, roundworm, and whipworm) are the focus of a genetic epidemiological study of the Jirels of Nepal. Two thousand members of a single family are being studied to assess the genetic determinants of levels of helminthic infection in this population. The second human family study is based in rural Brazil and focuses on the genetic determinants of susceptibility to and progression of *T. cruzi* infection, the cause of Chagas' disease. I also collaborate on a study of the genetic determinants of schistosomiasis in a population from northeastern Brazil.

*This workshop was made possible by generous contributions from the following sponsors:*

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