Preface

Population genetics is the study of the allele frequency distribution and change under the influence of the four evolutionary forces: natural selection, genetic drift, mutation and gene flow. It also takes account of population subdivision and population structure in space. This new book presents the latest research in the field from around the globe.

Expert Commentary - A simple method is presented for estimation of genetic origin, based on ratios between allelic frequencies of 11 widely applied Y-chromosomal short tandem repeats (STRs). Evaluating this strategy on 488 men of known ethnicity, the metapopulation was correctly predicted in 77% of the tests, while Africans were correctly discerned from non-Africans in 89% of the cases.

Chapter I - The results found allow us to establish certain conclusions on the structure of the population and the origin of Ecuadorians today. The volume of information obtained and the knowledge acquired continue to be insufficient in order to understand this ethnical, geographical and genetic structure of New World settlers. The genetic studies completed represent the first formal research into Ecuadorian ethno-genetics. The cultural crossbreeding can be found in different issues like economic, social, political and religious organizations, linguistic systems, communication codes, etc. The ethnic mixture process initiated during colonialisation continues today. The dominant economic and political group conveys the culture which is imposed in its economic, social, political and linguistic models for the others groups. The dominant culture leads the development of the Western culture and tends towards uniformity. It is presented as the "cultura ideal" and is transmitted by the media and in daily interaction. Ecuador is clearly a multi-ethnic and multicultural country. Cultural assessment, respect and rescue do not mean assessing, respecting and rescuing isolated cultural elements, but rather social groups with rights to resources for subsistence, to political representation and their own social and organizational bodies, to education in their own language and culture, and to determine their own development in line with their needs. In this work, the authors analyzed samples from three different ethnic groups of Ecuador, including Mestizos, indigenous American Kichwas and Afro-American Blacks; analysis included 20 autosomic microsatellites D3S1358, HumFGA, D21S11, Penta E, Penta D, HumVWA, D8S1179, D7S820, D13S317, D5S818, D16S539, HumTH01, HumCSF1PO, HumTPOX, HumF13A01, HumF13B, HumLPL, HumHPRTB, HumFES/FPS and amelogenin; there was also capillary electrophoresis analysis of 12 microsatellites of the Y-chromosome such as

DYS19, DYS385 a/b, DYS 389I, DYS389II, DYS390, DYS391, DYS392, DYS392, DYS393, DYS437, DYS438 and DYS439. With the autosomal STR markers the authors have confirmed and quantified that Mestizos and Afro-Ecuadorians are tri-hybrid populations. Mestizos contain ~73% of autosomal chromosomes of Native Americans origin, ~19% putative European and ~8% African origin. Afro-Ecuadorians can be estimated at ~57% Africans, ~28% Europeans and ~15% Native Americans. Seven different haplotypes were shared between Kichwas and Mestizos, one between Mestizos and Afro-Ecuadorians and one between Kichwas, Afro-Ecuadorians and Mestizos. This latter one is the most frequent haplotype in Europeans and, in particular, in Spaniards. The total number of different haplotypes is 276 in the entire sample analyzed. Mestizos are quite close to Kickwas, but are clearly closer to Spaniards than Kichwas. It is also the case for respective distance to Guineans. This is consistent with a triple genetic origin for the Mestizos: Amerinds, Europeans and Africans, as shown by the STRs in the Y-chromosome. Afro-Ecuadorians are the closest to Guineans, but are closer to Kichwas and to Spaniards than Guineans. With different proportions of mixture, the triple origin model proposed for the Mestizos is also applicable to Afro-Ecuadorians. The haplotypic heredity and the large possibility of combinations which these markers offer warn of the possible existence of allelic compositions as yet unanalyzed, and of those in which the real frequency is unknown.

Chapter II - Current reconstructions of prehistory mostly reject early attempts to force artificial classifications on the Indo-Pacific region, such as the tripartite division of Melanesia, Micronesia and Polynesia historically defined by Dumont d'Urville. Instead, the modern anthropological community embraces a more fluid view of human society, in which communities change through time and space, experience internal developments, and interact with surrounding groups. However, such a pliant conception provides a poor model system for many biological, linguistic and cultural clusters observed in the Indo-Pacific region today. Natural features – acting as conduits and barriers to human mobility – have also been influential regulators of population demography, and contemporary molecular research reveals numerous biological markers that reflect this geography. Some genetic characters show signs of regulation by natural factors; others even have surprisingly accurate concordance with the western boundary of Melanesia. A reconsideration of Melanesia and its biological environs from genetic, linguistic and anthropological perspectives emphasizes the temporal and spatial complexity of the Indo-Pacific world, but also reveals a mosaic prehistory dominated by population clines and clusters, and most importantly, strong evidence for population contact.

Chapter III - There is now a large body of scientific literature detailing the incidence of recessive genetic disorders within the British Pakistani population as well as a wealth of information describing the cultures, marriage traditions and general social structure of these communities. Too often, however, a simplistic connection between high disease incidence and marital mores is made. Indeed, appeals from high profile individuals to end the tradition of consanguineous marriages on health grounds continue to be made with the rather predictable backlash from community leaders. Although research indicates that the British Pakistani population does indeed suffer relatively high rates of infant inborn errors of metabolism, the simple genetics model relating consanguinity to an increased probability of homozygosity is by no means conclusive. Other complicating factors, such as broader

endogamous kin-networks and reproductive compensation, as well as demographic history and changing attitudes to traditional marriage customs, also require consideration. If useful advice and understanding are to benefit the communities in question, rather than offend and alienate them, an exploration of the consanguinity hypothesis is necessary. This chapter will outline some recent developments in the population genetics theory relevant to the incidence of recessive disorders within consanguineous populations and discuss how this knowledge can offer a more comprehensive approach to benefit community health; an approach which is likely to be in demand until more routine screening of recessive mutations becomes a real option.

Chapter IV - The National Geographic Society in the USA has decided to sponsor a multi-year project in population genetics research collecting 100,000 DNA samples from diverse indigenous peoples globally. The purpose of the project is to gain additional insights into the evolutionary and migration history of peoples inhabiting "the New World." Both Y-chromosome and mitochondrial DNA samples contribute evidence related to the hypothesis of a migration into the Americas from Asia via a Bering Land bridge. Many indigenous peoples object to the project with claims of biocolonialism, with competing scientific narratives of genetic history prospectively contradicting indigenous narratives about origins. Accordingly, both epistemological and ethical issues about competing perspectives on knowledge and the rights of scientific discovery versus indigenous rights are central to the dispute. This chapter explores relevant features of both positions in this dispute.

The cultural tensions looming over the Indigenous/West relations, in their historical dimension, are particularly magnified on the contested ground of knowledge production and in particular its flagship enterprise of research.

Willie Ermine, "Ethical Space: Transforming Relations"

For American Indians, the struggle of this century has been to emerge from the heavy burden of anthropological definitions that have made Indian communities at times mere laboratories for political and social experiments.

Vine Deloria, Red Earth, White Lies

Chapter V - Speciation is, according to the biological species concept, the process of decreasing gene flow between two subpopulations of one species. Population geneticists have introduced the concept of "effective migration rate" in order to quantify gene flow passing through a hybrid zone. The effective migration rate can be numerically calculated for a broad range of speciation models. The practical utility of the concept, however, has been limited by the lack of a systematic method to calculate effective migration rates analytically. Recently, we have developed a general and intuitive methodology to obtain analytical approximations of the effective migration rate for given isolating barriers of gene flow (e.g., genetic mating incompatibilities). Analytical formulae have great advantage over numerical analysis since they show how the strength of reproductive isolation is affected by genetic and ecological parameters. Here, the authors introduce this new methodology and apply it to the following genetic barriers: underdominance, two-locus incompatibility, and hybrid male inviability. These examples demonstrate that the concept of effective migration rate is a useful tool for analyzing population genetic models of speciation.

Chapter VI – The authors analyzed autosomal (Actine, CAT, CHRNA, GBA, IFN and Lactoalbumine) and Y-chromosome (DY8S, SmyS7, DY7S, and UBEY7) intron sequences from *Inia boliviensis* (Bolivia) and *Inia geoffrenis* (Peru and Colombia) captured from the Amazon and Orinoco watersheds of South America. The low level of genetic diversity they report, considering the number of base pairs sequenced and supported by as few as six nucleotide substitutions in some samples, is in disagreement with mitochondrial (control region and cyt-b) studies of these same populations. Although there are several possible explanations for this low diversity the authors suggest it may be due a homogenous aquatic habitat and the affect of constrictive natural selection on the *Inia*'s nuclear genome or to a more recent temporal divergence (Pleistocene) between the pink river dolphin populations than had been considered previously. Nevertheless, relative genetic differentiation statistics combined with phylogenetic procedures with the polymorphic sequence introns revealed almost complete isolation of the Bolivian and Amazon-Orinoco populations. Furthermore, most of the G_{ST}, F_{ST} and N_{ST} values were equal to 1 and respective gene flow estimates were equal to 0, supporting complete genetic isolation. Therefore, these molecular data provide additional evidence that the Bolivian *Inia* population is a different and significant evolutionary unit.

Chapter VII - With the advent of genomic analysis, increasingly sophisticated statistical methods have been devised to investigate and characterize human population structure. Coalescent theory allows the allelic states of individuals to be generated by assigning an allelic state to their most common recent ancestor (MRCA), with their lines of descent either followed forward in time or backwards to estimate the time to the most recent common ancestor (TMRCA). Alternatively, a full Bayesian analysis can be utilized, with the incorporation of previously ascertained information into the estimation process, and this approach now forms the basis of biostatistical analysis in many major genetic studies, including the HapMap project. The theoretical basis of one such Bayesian method, STRUCTURE, is described with its practical application initially portrayed in terms of global patterns of population structure and genomic diversity. Genomic diversity in nine ethnic populations in PR China is then explored via more traditional analytical techniques and Bayesian clustering inference. The study illustrates how STRUCTURE analysis can be successfully applied to smaller scale molecular anthropology studies based on a limited number of autosomal microsatellites, with information provided on both inter-population genetic differences and intra-population substructure.

Chapter VIII - The 8-10 million European Roma, also known as Gypsies, are a trans-national founder population, comprising a mosaic of socially and culturally divergent groups separated by traditional rules of endogamy. The Gypsies defy the conventional definition of a population: they have no nation-state, speak different languages and belong to many religions. Unlike other founder populations whose genealogy has been comprehensively documented, the demographic history of the Gypsies is sparse and, given the lack of written records, has been inferred from current genetic data. The authore studies of ancient maternal and paternal lineages across endogamous Gypsy groups in different countries point to common origins from a small group of related Asian founders. Sharing of disease mutations and high carrier rates support a strong founder effect, and the identity of some founder mutations in the Gypsies with those in Indian/Pakistani chromosomes provide the best

evidence yet of the Indian origins of the Gypsies. The history and demography of disease mutations in the Gypsies is also concordant with their migrational history in Europe. Disease haplotype coalescence times indicate that the entire Gypsy population was founded ~800 to 1000 years ago, with subsequent secondary founder events occurring ~200 to 700 years ago. Their exodus from India resulted in an initial profound demographic bottleneck, followed by gradual fragmentation into multiple genetically differentiated sub-isolates due to social and economic pressures within Europe. Together with the effects of random genetic drift and differential admixture, the string of population bottlenecks and founder effects have played a major role in shaping the current genetic architecture and consequently the genetic epidemiology of the Gypsies. The unique genetic profile of the Gypsies provides a considerable potential for genetic research.

Chapter IX - The role of 18 polymorphisms in 10 metabolizing genes (*CYP1A1*, *CYP2D6*, *GSTT1*, *GSTM1*, *MTHFR*, *MTRR*, *NQO1*, *CYP2C9*, *CYP2C19* and *NAT2*) in the risk of developing B-CLL and T-cell NHL in adults and the risk of developing leukemia in children has been analyzed. All patients and healthy controls were residents of the European part of Russian Federation and belonged to East Slavic group. For adult patients, the data obtained revealed the significance of specific genotypes for *CYP1A1, GSTM1, CYP2C9* genes in case of B-CLL and for *GSTT1, GSTM1* genes in case of T-cell NHL. Also it was demonstrated that null *GSTT1* genotype, null *GSTM1* genotype, *MTRR 66A/-* and *NAT2 341T/T,481C/C,590G/G* genotypes were risk factors to develop leukemia in childhood. The frequencies of metabolizing gene alleles in Russian population regarding other world populations have been discussed. The detailed analysis of the *NAT*-gene polymorphisms using NAT-biochip in about hundred healthy unrelated individuals allowed defining *NAT* alleles/genotypes occurred. The authors' data demonstrated that the frequencies of metabolizing gene variants in the European part of Russia are close to those described in other European populations.

Chapter X - Two different questions can be addressed by using molecular markers for making inferences on the effects of positive natural selection. First, when positive selection is proposed but direct evidence is lacking, genetic inference can reveal its existence. Alternatively, when positive selection is evident (fitness advantage is apparent), genetic or genomic tools can be used to identify specific genes or genomic regions that underwent selection. In this review the author discusses the methods and progress in detecting the signatures of selection by single-locus, multi-locus and genome-wide analyses. The author emphasizes 1) the differences between the two approaches, and 2) importance of verification of putative evidences of selection obtained with molecular markers by direct tests of fitness. Testing for adaptation with molecular markers engages primarily two types of positive natural selection that both increase frequency of a favorable allele, either to fixation (directional selection) or to an intermedium equilibrium (balancing selection). Therefore, the author discusses how signatures of balancing vs. directional selection can be detected by different statistical tests.

Chapter XI - Hungarians are unique among the other European populations with no nations but the Finns with a language belonging to the same (Finno-Ugric) language family. According to history, the ancient Magyars have come from the eastern side of the Ural Mountains and have settled down in the Carpathian basin in the 9th century AD. The origin of

the Hungarian-speaking Seklers, or as they call themselves the Székely, today an isolated minority in Transylvania, Rumania is still debated, though all sources agree on the Hungarian origin of this population.

Since variations in the maternally inherited human mitochondrial genome (mtDNA) are routinely used to explore the histories of different populations, the authors examined the distribution of restriction fragment length polymorphism (RFLP) sites of the mtDNA in apparently healthy, unrelated Sekler subjects in order to determine the genetic origin of this population. Among the 64 samples analyzed, the majority belonged to haplogroups common in other European populations examined. However, five samples fulfilled the criteria of haplogroup M classified as a haplogroup characteristic mainly for Asian populations. Thus, the presence of haplogroup M suggests that an Asian matrilineal ancestry can be detected among modern Seklers. The genetic structure of the Seklers investigated did not significantly differ from that of the Hungarians investigated earlier, therefore, it is most probable that an intense connection and genetic material interchange existed between the two populations during the past centuries. Photographs of each participating subject have also been taken and submitted for anthropological analysis which may reveal phenotypic variances and/or relationship between genotypes and phenotypes in the investigated Sekler population.

Chapter XII - With recent technological advances that can be applied to genetic tools for understanding ecological patterns and processes, the authors have seen a corresponding increase in the utility and application of molecular techniques to address important questions regarding species spread and persistence in novel environments. Applying molecular approaches to inform us about the invasion process has been tested in a number of recent cases, with varying degree of success. Success may depend upon the focal species or community in question, the marker system employed, the time since invasion, the biology of the species in question, and numerous other factors. There are many advantages to combining molecular approaches with field research to understand invasion processes. First, molecular markers can inform us as to the originating population or continent from which non-native or weedy species have arrived. They may provide insight to the numbers of invasion events and the time since invasion. This provides an opportunity to understand potential agents which can be utilised for biological control. Second, molecular markers may be the best means through which we can understand the precise movement patterns or dispersal of species within and across landscapes. Understanding movement pathways and dispersal trajectories is critical for management purposes. Third, because invasions frequently involve rapid evolution, molecular approaches can inform us as to gene expression that may contribute to invasion success. Hopefully, combining molecular marker tools with timely detailed information as to where in space and time invasions are occurring will permit more effective and efficient success in eradication or containment strategies while providing information that can be applied to a predictive framework in which to understand potential species and habitats which may be particularly susceptible to invasion.

This chapter focuses specifically on plant invasions, particularly those occurring within tropical ecosystems. However, the concepts described are applicable to a variety of taxa and landscapes and most research to date has taken place in other non-tropical ecosystems. Molecular approaches to understanding invasion and movement within and across landscapes are discussed with a focus on the importance of dispersal processes in tropical ecosystems.

The potential role of allee effects in impacting invasive species biology is considered and examples of recently published works are highlighted with a focus on innovative science. Marker utility in addressing ecologically and evolutionarily important aspects of invasion ecology are mentioned. Exciting new applications of technology are described, and some areas in which further study are urgently needed are mentioned.