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Book Review: Genetic Variation and Disorders In Peoples of African Origin

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Books

Genetic Variation and Disorders in Peoples of African Origin

By James E. Bowman and Robert F. Murray, Jr.
The Johns Hopkins Press, Baltimore, Md., 472 pp.

Reviewed by Melvin E. Jenkins

This unique and informative volume brings together descriptions, comparative frequencies and commentary on scores of genetic variations in peoples of African descent. Implications of major disorders, such as sickle cell disease, glucose-6-phosphate dehydrogenase deficiency and hypertension, are explored in considerable detail. Lactose intolerance, which deserves special attention, is the subject of a separate chapter.

At the outset, it is important for the reader to remember that much documented evidence points to the continent of Africa as the site where mankind first evolved and later appeared on other land masses. From this broad perspective, all human life is genetically linked to Africa, with wide variations orchestrated by mutations and environmental forces.

Another possible factor for variation was determined by the indigenous characteristics of geographic areas where homosapiens lived and reproduced for thousands of years. Certainly, hundreds of thousands of years of genetic adaptation have altered many components of the human genome. Influences of the environment is readily observed. Peoples who live in intensely hot climates have darker skin as a consequence of the solar effects on melanin synthesis.

Genetic variation in male and female phenotypes, chromosome karyotypes and blood hormone levels is essential for the reproductive role of humans and most animals and plants. Thus, this variation is essential to human life and ultimately to the survival of the species. In this connection, knowledge of genetic variation is critical to a fuller understanding of most congenital and acquired diseases. Careful utilization of genetic information has led and will continue to lead to better medical diagnostic skills, treatment strategies and long-term health care for people as individuals, as well as defined population groups.

> The appaling health status of Africans and African Americans is related to poverty, lack of access to good health care. . . .

While Bowman and Murray emphasize genetic studies in Africans and their descendants, considerable data is included in the book from studies of Europeans, Asians, Amerindians and non-African blacks, such as the Australian Aborigines and South Sea Island Negritos. This book, indeed, can be quite useful both as a reference and for professional enhancement. This is so because of the numerous epidemiologic and demographic research studies by Bowman, a specialist in internal medicine and a pathologist at the University of Chicago, and the broad medical experiences of Murray, a geneticist at the Howard University College of Medicine.

The first chapter considers the migration of African peoples, both within the continent and to distant lands. Early migrations of the western Zonda and Bantu to the east, for example, carried gene pools with them. Obviously, the slave trade resulted in the transplantation of millions of Africans to western continents where gene pool admixtures with Europeans, Amerindians and others diluted the African genes. Before the forced migrations of slaves, the voluntary migration of Africans to the Americas, long before Columbus, is scientifically supported by documented scholarly literature.

The second chapter covers anthropometry and skeletal variation. Numerous studies by anthropologists delineate population variations in facial and skeletal characteristics which prove useful in health maintenance and disease detection. For example, the significantly higher frequency of hip fractures in elderly white women, as compared to elderly Black women, may be explained by genetic differences in hip bone thickness, among other factors. While a few racist scientists have attempted to distort anthropometric differences to denigrate ethnic groups, particularly Blacks, the 50 pages on anthropometry in this book spans substantial literatures by respected investigators. The chapter provides a wealth of consolidated and tabulated information, as well as individual references for unbiased and objective use. I am unaware of a similar single resource which has presented so well the important skeletal variations found in African peoples.

Skin pigmentation is dealt with in the third chapter, which highlights albinism not only in Africans but also in other major racial groups. Dermatoglyphics, the study of hand and finger print patterns, is considered in the fourth chapter. In general, this topic is somewhat difficult for the non-specialist, but finger and palm prints provide important clues for the physician and geneticist as clinical supporting evidence for many congenital disorders.

The chapter on polymorphisms is an important one. It focuses on many of the 73 protein, peptide and enzyme systems which have demonstrated genetic variation in specific populations and among different populations. Population geneticists explain polymorphism by two contrasting concepts. The first is based on natural selection. Here, heterozygosity, for example, can confer a selective advantage for affected individuals, thereby increasing their survival chances. However, homozygosity or a double dose of the allelic gene may cause debilitating or even fatal disease. For example, sickle

cell anemia in its heterozygous or carrier form, appears to have a protective effect against severe falciparum malaria, but in homozygous form, sickle cell disease can be life threatening.

The second theory explains the occurrence of heterozygous traits by chance alone. Also, random genetic drift has been introduced as an explanation for non-selective genetic variation.

The identification of the major blood types was the first indication of significant genetic polymorphism in humans. The blood groups A—B—O vary widely throughout the world. The highest frequency of the O-group is found in Central and South America. In African Americans, group-O is found in 71 percent. Northwest Europeans show a similar rate, 68 percent. The gene for A is common in Europe and among the Australian Aborigines.

It is now known that polymorphisms are found in serum proteins, red cell enzymes, hemoglobins, HLA, chromosomes and DNA fragments, both associated and unassociated with disease. The polymorphisms provide useful clues to the past and perhaps to the future of the human species.

The G-6-PD deficiencies, which can cause red cell destruction and anemia when affected individuals are exposed to certain drugs, are relatively common in peoples of African origin and those from the Mediterranean region.

Sickle cell disease and the thalassemias are quite frequent in Africans and their descendants. These variants are found in most peoples who are exposed to high concentrations of falciparum malaria. Some evidence against a role for malaria in the hemoglobinopathies is included in the book. However, the authors' arguments are not very compelling for this reviewer.

A chapter on lactose intolerance stresses that problems related to milk sugar absorption are common in the peoples of Africa, Southeast and East Asia, New Guinea and the Americas. This intolerance tends to be more frequent in old age in populations with a history of low milk intake, and lower in peoples with a long history of drinking lactose rich milk. This may mean that milk intake induces the lactose absorbing gene in most people, which helps maintain milk tolerance.

Congenital malformations are discussed in the eleventh chapter. Birth defects are often genetically determined and are quite common in humans. African Americans have a higher frequency of minor congenital anomalies such as polydactyly and preauricular fistulae. Life threatening anomalies at birth occur at similar rates in Blacks and whites. Whites, however, have significantly more multiple birth defects than African Americans.

On balance, this book adds a much needed resource for population genetics.

One of the highest incidences of dizygotic twinning is found in Nigeria with frequencies as high as 1 in 20 births, compared to 1 in 80 births in the United States. Factors other than genetics, maternal age for example, may contribute to twinning.

Hypertension is more common in African Americans than in the white population. However, the relative causative roles of genetics and stress are not clear. It is noteworthy that some drug therapy for high blood pressure, such as diuretics and calcium channel blockers, are more effective in African Americans than in whites. On the other hand, the widely used beta-blockers work better in whites.

The section on diabetes mellitus is disappointingly superficial. However, it does correctly emphasize that maturity onset diabetes with its insulin resistance is much more frequent in African American youth than in white teenagers. This observation deserves much more study. Many of the known differences in diabetes in African Americans, such as HLA types, are not addressed. The reviewer is aware of an enormous frequency of diabetes in the Australian Aborigines rivaling that of the Pima Indians of the Southwestern United States. The second edition of this book must contain an expanded chapter on diabetes, the genetics of which is still unclear.

Genetic Variation and Disorders in Peoples of African Origin concludes with three chapters of immediate urgency and relevance. Chapter 13 concerns the current health status of Africans and African Americans, followed by chapters on genetic counseling and ethics and public policy.

The appalling health status of Africans and African Americans is related to poverty, lack of access to good heath care and improper health education. In Africa, inadequate food, non-potable water, poor housing and lack of educational opportunities must be corrected to effectively improve the general health in all age groups.

On balance, this book adds a much needed resource for population genetics. And it is a must reading for the geneticist and anthropologist. It provides a ready reference for health related professionals, sociologists and intellectually inquiring minds of any persuasion. □

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