

## I. INTRODUCTION

# The Relevance of Genetics to Medicine

This issue of the *MCV Quarterly* focuses on the explosion of knowledge in the field of human genetics. The activities of the new Department of Human Genetics at the Medical College of Virginia encompass the traditional medical school triad of teaching, patient care, and research, and an active graduate program has been developed with curricula leading to masters and doctorate degrees. The program is supported by a recently awarded National Institutes of Health predoctoral training grant as well as State and local funds from the A. D. Williams Foundation.

Clinical services are provided in a National Foundation-March of Dimes Genetic Counseling Clinic and through an interdepartmental Antenatal Diagnosis Program. At present, however, as shown by some of the case reports in this issue, research and clinical service are inextricably intertwined.

There has, perhaps, been no recent advance in medical technology that has greater potential for improving the human condition than the development of reliable methods for the prenatal diagnosis of a growing list of specific genetic diseases. The Antenatal Diagnosis Program at MCV has been developed with support from the State Department of Health and has at its disposal the most modern equipment available for these test procedures. Neonatal screening for genetic disease and carrier detection programs are two logical complements to prenatal diagnosis.

It is becoming increasingly clear that the major causes of morbidity and mortality in Western cultures are neither entirely environmental nor genetic in etiology but result, rather, from an interaction between the host and a pathogenic agent in a constantly changing environment. The concept of genetic risk factors may well emerge as the most significant and effective new health care strategy to appear during the last quarter of this century. Experience with the treatment of genetically determined metabolic diseases suggests that when a specific genetic liability can be diagnosed, patients and their parents are in

general highly motivated to comply even with extremely elaborate treatment regimes. Wouldn't an individual who had seen a parent or close relative die of hypertension, heart disease, cancer, or pulmonary disease have a strong motivation to comply with a program of presymptomatic screening and treatment if he or she could be shown to carry a specific genetic risk factor? Improved public education will be essential for the acceptance of this approach to disease prevention. Since all normal individuals carry abnormal recessive genes, the diagnosis of a genetic disease or carrier state in a family should not be viewed as a stigma but rather as a readily identifiable risk factor that can be exploited by the prudent physician to plan a highly individualized health maintenance program for the patient. At present, relatively few alleles have been identified which can be detected in the heterozygous state that are associated with an increased disease risk; however, the number is growing, and it seems likely that this will continue to be a very active area of research in the future.

Genetic diseases are estimated to account for 15% of all pediatric hospital admissions. Recognizable chromosome abnormalities account for more than 50% of all early spontaneous first trimester abortions. A single genetic disease, Usher syndrome, accounts for more than 60% of all adults seeking rehabilitative services for the combined handicap of deafness and blindness. The lifetime cost of caring for the 3,000 to 4,000 new children who are born each year with Down syndrome is estimated to be in excess of \$60,000,000 per year. More than 2,000 genetic diseases have now been described, most within the past two decades, and at least one genetic locus has now been mapped to each of the 23 pairs of chromosomes, most within the past two years. These facts presage the increasingly important role that genetics will play in medicine and human affairs in future years.

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