IV. CLINICAL PROGRAM

The Genetic Counseling Program at MCV

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The Genetic Counseling Clinic at the Medical College of Virginia, established by Drs. Peter Mamunes and R. B. Young in 1973, has been supported since its inception by a clinical service grant from the National Foundation—March of Dimes; it is one of 83 genetic counseling programs in the United States and one of three in Virginia that receive support from the Foundation. The Clinic provides counseling and diagnostic services for a variety of genetic diseases and is the focus of clinical teaching and research activities of the Department of Human Genetics. The Clinic is staffed by members of the Departments of Human Genetics, Obstetrics, and Pediatrics, as well as consultants from many other clinical disciplines.

Following referrals, patients are sent a questionnaire to initiate the collection of relevant medical and genetic data; the patient is then scheduled for a Clinic appointment. At the time of the Clinic visit, the graduate student interviews the patient and documents sufficient family history information to permit the construction of a pedigree. Following review of the collected data and physical examination of the patient by the staff physician, the patient and family are invited to continue the counseling session in a room equipped with a microphone and a two-way mirror. In this setting, students, house officers, and staff members can participate in the counseling session in an unobtrusive manner, and discuss it later. About 90% of the patients and families are willing to participate in this educational program; those who object are counseled privately.

After a year's experience in the Genetic Counseling Clinic, graduate assistants are assigned to specialty areas such as hemophilia, cystic fibrosis, and endocrine clinics, where they collect family history data in selected cases, under the supervision of the Clinic director and augment the genetic counseling the families have previously received. Although virtually all patients seen by the graduate assistant in these clinics have a clearly defined Mendelian disease, many may not have received adequate genetic counseling in the past.

Since its beginning in 1973, the Clinical Genetics Program has evaluated or counseled 698 individuals (Table); of these, 33.7% were diagnosed as having a simple inherited Mendelian trait and 45.6% a recognizable chromosomal disorder. Twenty-one dominantly inherited traits were diagnosed including achondroplasia, neurofibromatosis, Huntington chorea, Marfan syndrome, retinitis pigmentosa, hypochondroplasia, Noonan syndrome, aniridia, limb-girdle dystrophy, Treacher-Collins syndrome, and tuberous sclerosis; 28 recessive traits were diagnosed including cystic fibrosis, alpha-1-antitrypsin deficiency, recessive deafness, Tay-Sachs disease, Werdnig-Hoffman disease, galactosemia, phenylketonuria, Gaucher disease, Usher syndrome, and maple syrup urine disease. X-linked traits seen in the clinic included Duchenne muscular dystrophy, hemophilia, Norrie disease, anophthalmia and Charcot-Marie-Tooth disease. Most of the cases seen with chromosome anomalies had Down syndrome, but the

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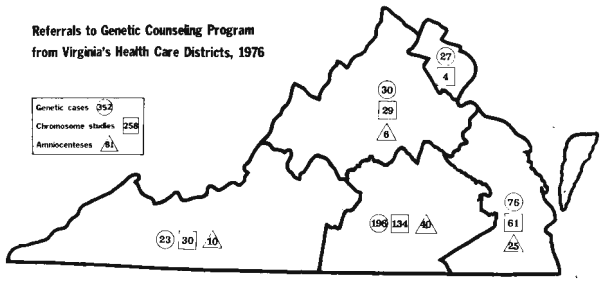
Summary of Genetic Clinic Population: 1973–1977

Classification	Year 1973–75	1976	1977 Jan-June	Total Number	Percent of total
Dominant traits	16	55	40	111	12.2
Recessive traits	46	54	61	161	17.8
Sex-linked traits	16	11	7	34	3.7
Chromosomal disorders	106	258	49	413	45.6
Multifactorial traits	16	24	10	50	5.5
Other conditions	41	55	42	138	15.2
TOTAL	241	457	209	907	100.0

sample included patients with D and E trisomy, the cri du chat syndrome, Turner syndrome, Klinefelter syndrome, the 4p-syndrome, and a variety of structural rearrangements.

Patients classified as having multifactorial diseases included individuals with midline neurologic defects, diabetes, uncomplicated cleft lips and/or palate, clubfeet, seizures, and certain patients with familial mental retardation that could not be otherwise classified. Patients falling into the other categories included individuals with multiple malformations or repeated abortions with normal chromosomes and no other definite causes, or patients with psychomotor retardation of unknown or environmental etiology.

During the year 1976 (Figure), patients came from all parts of the state either to the Genetic Counseling and Amniocentesis Clinics or for diagnostic karyotypic analysis. This pattern of referrals is largely a result of the central location of the Clinic in a large urban area.



Figure—Pattern of referrals to Genetic Counseling Program from Virginia's health care district, 1976.

Population Screening for Genetic Disease

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Recent advances in genetics and laboratory techniques have raised difficult issues for both the medical and lay communities. The desirability of initiating population screening programs is an example of one such issue that has engendered considerable confusion concerning its intent—so much so that the National Academy of Sciences recently reviewed this subject and in 1975 published a book entitled, *Genetic Screening—Programs, Principles and Research.*[1]

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