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Part III: Clinical Departments and Divisions -- Chapter 20: Division of Medical Genetics (pages 396-398)

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Division of Medical Genetics

LAIRD G. JACKSON, M.D.

“Rare is the tree whose every bud develops into perfect fruit, and if there be such a family tree I find no adequate evidence of it.”

—VICTOR C. VAUGHAN (1851–1929)

Genetics in the 1960s

The Division of Genetics was established within the Department of Medicine in 1969. Impetus in this field developed in 1960 with the discovery by Drs. Peter Nowell and David Hungerford¹ in Philadelphia of chromosomal changes in chronic myelocytic leukemic cells. Dr. Laird Jackson, a resident in internal medicine, who had been working with Dr. Arthur J. Weiss in 1959 and 1960 on clinical investigation of cancer chemotherapeutic agents, became interested in the cytogenetics of neoplasia. In 1961 his success with the application of cytogenetic techniques attracted requests from pediatric and obstetrical clinicians to detect cytogenetic abnormalities in their patients.

The finding of such defects, with the necessity of explaining them to patients, led to the beginning of genetic counseling.

In 1962 a National Institutes of Health research grant for the study of chromosomal nucleic acid processing, and in 1964 a March of Dimes grant for the study of chromosomal histone proteins aided the research of Dr. Jackson. Subsequent fellowship support from the Leukemia Society, expanding into a five-year Leukemia Society Scholarship in 1965, extended his tissue culture and chromosomal research.

At the creation of the Genetic Division by Dr. Robert I. Wise, Chairman of Medicine, Dr. Jackson was appointed Director. Research Fellows from the Oncology Division were provided

Robert L. Brent, Chairman of Pediatrics, provided new space for clinical activity and introduced two additional members, Drs. Gary G. Carpenter (Jefferson, 1960) and Leonard E. Reisman.

Genetics in the 1970s

training in methods used in cytogenetics and tissue culture. One of these Fellows was Dr. Carla E. Goepf.

In the late 1960s Dr. Leon A. Peris (Jefferson, 1955) of the Obstetrics Department and Dr. Irving J. Olshin of the Pediatrics Department began to combine their interest along with Dr. Jackson across departmental lines. Weekly or biweekly meetings of the three were held to share case experiences, hold informal conferences, and profit from the chromosome laboratory. In 1967, Dr. Roy B. Holly, Chairman of Obstetrics and Gynecology, supported a grant request from the March of Dimes that established a formal Genetic Counseling Clinic, which united the efforts of the Medical and Obstetric Departments. Dr.

In 1971 Dr. Peris began genetic prenatal diagnosis with Dr. Jackson and Marie Barr (Figure 20-1). Dr. Peris performed the amniocentesis, and Ms. Barr and Dr. Jackson handled the genetic counseling and the cytogenetic laboratory processing of the tissue cultures and chromosome studies of the amniotic fluid cells. With the training of Dr. Ronald J. Wapner in perinatology and genetics in the mid-1970s, the collaboration with Obstetrics became complete.

Although the Directorship of the Division of Genetics was within the Department of Medicine, Dr. Jackson held Professorships in Medicine, Pediatrics, and Obstetrics and Gynecology. Dr. Susan Z. Cowchock (Jefferson, 1968) and



FIG. 20-1. Dr. Leon A. Peris, Ms. Marie Barr, and Dr. Laird G. Jackson conduct genetic counseling.

Eugene E. Grebner augmented the Division. The teaching of genetics was done in several departments both in the preclinical and clinical years for covering the related material in several disciplines: Dr. Arthur Allen included genetics as part of the Cell and Tissue Biology block administered by the Biochemistry Department; Pediatric Genetics and Neurology were taught by Drs. Leonard J. Graziani, Jeanette C. Mason, and Gary G. Carpenter; and Maternal Fetal Medicine was taught in the Department of Obstetrics and Gynecology by Drs. Ronald J. Wapner, Benjamin Chayen, and George Davis.

Clinical service in genetics continued to grow rapidly as applied to diagnosis and counseling. This activity involved six M.D. geneticists or Fellows and four non-M.D. genetic counselors with technical personnel varying in number between six and 14. Several new genetic tools were established within the Division. In 1972 Dr. Susan Cowchock developed the first maternal serum alpha-fetoprotein method in the United States to detect fetal neural tube defects. This program provided a service for the entire Delaware Valley region as well as many areas beyond. In 1972 the Division established one of the country's first Tay-Sachs disease prevention programs. In 1974 Dr. Eugene Grebner headed this effort, became a national leader on the scientific advisory committee, and instituted biochemical genetic research now leading toward contemporary molecular genetic research. This research program was further expanded in 1986 when Dr. David A. Wenger, an internationally known researcher in lysosomal disorders, joined the staff. Dr. Wenger brought a group of three additional laboratory research workers and established a tie with the new Institute of Molecular Medicine with cross-

appointments to the Institute and the Department of Biochemistry.

In the mid-1970s Dr. Ronald Wapner established a leading prenatal diagnostic program. Collaborative work with the Division of Ultrasound led to additional innovative procedures. Fetal diagnostic work, always a major activity of the Division, was extended when Dr. Ronald J. Wapner added the fetal medicine program in the late 1970s. In collaboration with Drs. Barry B. Goldberg and Alfred B. Kurtz of the Ultrasound service, early diagnosis of fetal anatomic abnormalities was developed. The most rapidly successful accomplishment occurred when Drs. Wapner and Jackson began work on first-trimester fetal diagnosis by chorionic villus sampling in 1983.³ This led to an international leadership position in the field of first-trimester diagnosis.

Other innovative diagnostic achievements by the entire genetics team may be anticipated in this rapidly expanding field.

References

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