

Herbert A. Lubs, Jr. MD

In Memoriam

Herbert Augustus Lubs, Jr, MD died on February 10, 2023, at the age of 94. Known as “Herb” to his friends and genetics colleagues, his long and illustrious career includes recognition as a clinical investigator, educator, administrator, clinical geneticist and cytogeneticist as well as time devoted to national service. His numerous publications covered a broad range of cytogenetics and clinical genetics topics such as pediatric and cancer cytogenetics, chromosome banding techniques, characterizations of chromosomal heteromorphisms, examination of chromosomal gaps and breaks in lymphocytes of unaffected individuals, clinical description of genetic syndromes, and genetics of dyslexia. However, Herb is best known for his work in X-linked mental retardation and his 1969 elucidation and reporting of the “marker X chromosome” (later known as the “fragile X chromosome”).

While at Yale Medical School, Herb became interested in the nascent field of medical genetics. One of his earliest contributions to medical genetics and the study of chromosomes came during two years at the National Cancer Institute (NIH) as part of a team that brought about the first three advanced cancer treatments. His time there resulted in the publication of a series of studies investigating the correlation between the degree and number of chromosomal abnormalities and the progressive severity of cancers.

As the first Director of Medical Genetics at Yale Medical School, Herb conducted a comprehensive study in which chromosome analysis was performed on 4,500 consecutive newborns, resulting in a database of chromosomal disorders at birth that subsequently provided valuable information for use in prenatal diagnosis.

In 1969, Herb published his landmark discovery of an unusual "marker X chromosome" in association with intellectual disability (AJHG, 1969 PMID: 5794013) in 4 affected males in 3 generations. He reported “an unusual secondary constriction at the ends of the long arm of a group C chromosome. This gave the appearance of large satellites, but the chromosome was never seen in association with the acrocentric chromosomes.” Herb’s work resulted in his original identification of the Fragile X Syndrome, which represents the most common genetic cause of mental retardation in males as well as the first genetic marker specifically associated with dyslexia. This groundbreaking work was recognized in 1986 by the Joseph P. Kennedy Jr. Foundation, which awarded him its International Award in Mental Retardation.

Herb left New Haven to become Associate Professor of Pediatrics, and Biophysics and Genetics at the University Colorado Medical Center. In 1979, he moved to the University of Miami School of Medicine and served as Professor of Pediatrics and Genetics Division Director, Mailman Center for Child Development until retirement and Professor Emeritus status in 2004. At the Mailman Center, he headed up a busy clinical genetics service, cytogenetic and biochemical genetic testing laboratories and research into the genetics of dyslexia among other things. Late in his career he was also a Professor of Genetics, University Tromsø, Norway.

Noted for his busy lecture schedule from medical school classrooms to national and international scientific venues, in 2017 Herb was presented the Albert Nelson Marquis Lifetime Achievement Award by Marquis Who's Who for his noteworthy contributions as a genetics educator and administrator.

Herb's professional service activities include Chairmanships of the American Society of Human Genetics (ASHG) Committee on Environmental Hazards and the 1979 National Institute of Child Health and Human Development (NICHD) sponsored meeting on genetic counseling. From 1974-76 he served on the ASHG Board of Directors, and later on ASHG's Genetic Services Subcommittee on Cytogenetics Laboratory Proficiency Testing and Quality Assurance. Herb Co-chaired the National Institutes of Health (NIH) 1983 meeting on Marker X, and he was a member of the Steering Committee for the NICHD Collaborative Study on Chorionic Villus Sampling and Amniocentesis. He was also a Founding Fellow of the American College of Medical Genetics and Genomics, and as a founding member of the Southeastern Regional Genetics Group (SERGG), Herb led its Cytogenetics Workshop along with Jean Priest.

Throughout his long career, Herb was frequently called upon as an NIH site visitor and *ad hoc* review committee member, known for his broad cytogenetics expertise—from the behavior of chromosomes in the laboratory to their clinical manifestations throughout the lifecycle. Herb was a Diplomate of the American Board of Internal Medicine and the American Board of Medical Genetics and Genomics whose commitment to clinical investigation and scientific discovery never waned.

Submitted by:

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Adapted from:

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Herbert A. Lubs Presented with the Albert Nelson Marquis Lifetime Achievement Award by Marquis Who's Who. Marquis Who's Who Press Release, October 26, 2017. <https://www.24-7pressrelease.com/press-release/445774/herbert-a-lubs-presented-with-the-albert-nelson-marquis-lifetime-achievement-award-by-marquis-whos-who>