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July 23, 2008

FOR IMMEDIATE RELEASE

VICTOR A. McKUSICK, M.D., "FATHER OF MEDICAL GENETICS," 1921-2008

Victor Almon McKusick, M.D., University Professor of Medical Genetics at the Johns Hopkins University School of Medicine, one of the two distinguished Johns Hopkins geneticists for whom the McKusick-Nathans Institute of Genetic Medicine was named, and a towering international figure in genetics research, diagnosis and treatment, died Tuesday, July 22 at home. He was 86.

The relentlessly energetic clinician scholar, a pioneer in the pursuit of the links between inheritance and disease, died at home, of complications due to cancer.

To the world scientific community, McKusick had long been honored as a founding father of medical genetics as a specialty, and as a memorable mentor of generations of faculty, fellows, residents and students numbering in the thousands. Showered with scores of national and international prizes, honorary doctorates and accolades during a professional career spanning more than 60 years spent entirely at Johns Hopkins, he was the recipient of the 1997 Albert Lasker Award for Special Achievement in Medical Science, the 2001 National Medal of Science, and the 2008 Japan Prize in Medical Genomics and Genetics. He was the founding president of the Human Genome Organization and a member of the National Academy of Sciences.

"Today we have lost a giant," said Edward D. Miller, M.D., dean and chief executive officer of Johns Hopkins Medicine. "He spent virtually all of his incredible career at Hopkins, but his influence and legacy reach around the world."

"Victor McKusick was a superb scholar in many fields of medicine," said Richard Starr Ross, M.D., dean emeritus of the Johns Hopkins University School of Medicine. "His achievements have added great luster to Johns Hopkins."

Elevated to the rank of full professor in 1960, McKusick was elected to the National Academy in 1973, astonished countless students, patients and colleagues with his prodigious memory for clinical and scientific details, and drew praise as an avid historian of the field he helped define.

McKusick first came to Johns Hopkins as a medical student in 1943 and stayed to complete his internship and residency in internal medicine. He served as executive chief of the cardiovascular unit at Baltimore Marine Hospital from 1948 to 1950 while progressing through the ranks in the Johns Hopkins Department of Medicine. He also held joint professorships in epidemiology in the Johns Hopkins University School of Public Health and in biology at The Johns Hopkins University. In 1957, he founded the Division of Medical Genetics, which he headed until 1973, when he became the William Osler Professor and chairman of the Department of Medicine and physician in chief of The Johns Hopkins Hospital. He held these posts until 1985, when he was named University Professor of Medical Genetics.

Spry, serious, but with an almost jolly sense of humor, McKusick remained active until last year in his role as University Professor of Medical Genetics, making frequent trips to his campus office. At least once a year for decades, residents, medical students and even faculty and staff could be seen following him at a quick pace up the narrow winding stairs to the catwalk of The Johns Hopkins Hospital's iconic dome. It was there that he invited them to share the view and concluded his famous mini-history of the institution he loved.

Drawn initially to cardiology, McKusick detoured early in his career into a deepening study of a relatively rare inherited disorder known as Marfan syndrome, which is marked by heart defects, unusually tall stature and other abnormalities. His fascination with this disorder started with one tall patient, with a dangerous weakening of the aorta and a detached retina.

Soon after making that first diagnostic recognition of the complex group of symptoms linked to a single inherited gene, he was sought out by other Marfan patients and began to keep his hallmark meticulous records of the inheritance patterns and clinical features of the syndrome as well as other diseases that ran in families.

That path would capture his full-time devotion, as he went on to examine populations with relatively isolated gene pools, such as the Old Order Amish of Pennsylvania, sensitively but formidably using their medical and genetic histories as a way to quickly identify the genes responsible for their inherited physical abnormalities and disorders.

"Dr. McKusick was passionate about all things medical and genetic; his enormous abilities and energies in these areas created an entire field and served and will continue to serve as a model for all of us interested in genetics and medicine," said David Valle, M.D. the Henry J. Knott professor and director of the McKusick-Nathans Institute of Genetic Medicine. "He was a force in our field and a great colleague and friend. We will miss him tremendously but will carry on as he would want us to do."

Throughout the next several decades, McKusick led the world in searching for, mapping and identifying genes responsible for thousands of inherited conditions including Duchenne muscular dystrophy, achondroplasia and many other forms of dwarfism. He showed, by example, through careful record-keeping and an unmatched enthusiasm in following inheritance patterns, that understanding the genetics behind so-called Mendelian or single-gene diseases could lead to new methods of classifying disease and to their diagnosis and treatment. His pioneering work in Marfan syndrome set the foundation for others at Hopkins and elsewhere, who have identified not only the molecular players in those genes, but also now successful treatments.

"While I will miss Dr. McKusick deeply, I feel his tangible presence in my clinic and in my lab on a daily basis," said Harry C. (Hal) Dietz III, M.D., the Victor A. McKusick Professor of Genetics and Medicine and director of the William S. Smilow Center for Marfan Syndrome Research at Johns Hopkins. "His legacy to medicine is so pervasive, even fundamental, that it will be difficult to pinpoint but impossible to avoid."

An early proponent of completely mapping the human genome, in 1966 McKusick created the first edition of his now classic reference *Mendelian Inheritance in Man*, an ever-enlarging compilation of inherited disease genes. Now in its 12th edition in print, and consisting of three volumes, the seminal reference also exists as *OMIM, Online Mendelian Inheritance in Man*, a continuously updated version on the Internet, providing a searchable database of disease genes' locations and characteristics.

In 1969, McKusick was one of the first to propose the human genome map, whose sequence was published in February 2001.

Today, the importance of recognizing and understanding the links between various genes and disease almost goes without saying in many circles, including among non-scientists. Indeed, finding a gene, and frequently even linking it to a disease, no longer makes headlines, reflecting the widespread acceptance of McKusick's fundamental approach to studying disease.

"Dr McKusick has had as great an impact on medicine and Johns Hopkins," said Myron L. Weisfeldt, M.D., the William Osler Professor of Medicine and director of the Department of Medicine at Johns Hopkins. "His life-long creative genius in his early days advanced cardiovascular medicine. He then discovered numerous genetic diseases and formulated information that led to identification of their genetic cause and, for many, their treatment. I doubt that anyone would have conceived of the human genome project if he had not shined the light on the value of genetics in so many human conditions. He was an undying advocate of the Johns Hopkins spirit, principles and history. I was one of his many mentees who valued his support."

Inevitably, his growing interest in genetics led McKusick to explore using experimental scientists' favorite human substitute - the mouse - in studies that could not be done in people. And in 1960, McKusick co-founded the highly regarded Short Course in Medical and Experimental Mammalian Genetics, held in conjunction with The Jackson Laboratory in Bar Harbor, Maine, an internationally recognized center of excellence in mouse genetics. Because mice and humans share similar development and physiology, and because of the power of mouse genetics, McKusick was among the earliest human geneticists to recognize the value of the mouse as a model for understanding human disease. He championed joint studies between scientists in both fields, and he was a co-director of the "Short Course" for 49 years and was busy planning the 50th at the time of death.

With a faculty from Johns Hopkins, Jackson Lab and institutions around the world, the Short Course has, over the years, played a key role in training more than 4,000 aspiring medical geneticists. Additionally, hundreds of medical journalists have participated in another of his innovations, "Press Week," at the annual Short Course, resulting in more informed stories on the latest developments in the understanding genetics in health and disease.

"Victor McKusick's seminal contributions in genetics, medicine and education have simply become synonymous with excellence in biomedicine," said Richard P. Woychik, Ph.D., president and chief executive officer of The Jackson Laboratory. "His involvement with The Jackson Laboratory over the past 50 years in co-organizing the Short Course in Experimental and Mammalian Genetics is a reflection of his intense commitment to help thousands of students, scientists and physicians learn and put into practice the remarkable power of genetics for understanding human disease."

Born Oct. 21, 1921, in Parkman, Maine, McKusick and his identical twin, Vincent, grew up on a dairy farm in Maine. Both of McKusick's parents had been teachers and made education a priority for their five children. McKusick attended Tufts University from 1940 to 1943, when he entered the Johns Hopkins University School of Medicine without completing his bachelor's degree. In 1949, he married fellow physician Anne Bishop, who remains a part-time assistant professor in Johns Hopkins' Division of Rheumatology.

Among McKusick's many honors are the John Phillips Award of the American College of Physicians for distinguished contributions in internal medicine (1972), the Gaidner International Award (1977), the William A. Allan Award of the American Society of Human Genetics (1977), the James Murray Luck Award from the National Academy of Sciences (1982), and the Sanremo International Prize for Genetic Research (1983). He was inducted into the International Pediatrics Hall of Fame (1987) and received the Passano Award (1989) and the American Association of Physicians' George M. Kober Medal (1990). More recently, McKusick was honored with the Ellen Browning Scripps Medal and the

John P. McGovern Compleat Physician Award. McKusick also served in distinguished positions on numerous advisory boards and editorial boards and within professional organizations.

In addition to his wife, Anne, and their children Ms. Carol Anne McKusick of Urbana, Ill., Mr. Kenneth Andrew McKusick of Ruxton, Md., and Reverend Victor Wayne McKusick of Herkimer, NY, McKusick is survived by his twin brother, Vincent, a retired chief justice of the Supreme Court of Maine.

Visitation is scheduled for Friday, Aug. 1, from 3 p.m. to 5 p.m., and from 7 p.m. to 9 p.m. at the Ruck Towson Funeral Home, in Towson, Maryland. A memorial service will be held Saturday, Aug. 2, at 2 p.m. at the Second Presbyterian Church of Baltimore. Interment will be Friday, Aug. 8, at 11 a.m. at Pingree Cemetery in Parkman, Maine.

On the Web:

European Genetics Foundation:

<http://www.ronzano.org/index.php?l=it&p=mcks>

Lasker Award 1997:

<http://www.the-scientist.com/article/display/17766/>

http://www.jhu.edu/news_info/news/univ97/sep97/lasker2.html

National Academies of the Sciences

http://www.nasonline.org/site/PageServer?pagename=AWARDS_scirev

Oral History of Human Genetics Project

<http://www.socgen.ucla.edu/hgp/mckusick.html>

University of New England

CHP and CAS Honorary Degree Recipients 2008

<http://www.une.edu/studentlife/graduation/honorary.asp>

Memorial University of Newfoundland

World's best known geneticist visits Memorial

<http://www.med.mun.ca/munmed/104/genetics.htm>

The New York Academy of Medicine 2006

<http://www.nyam.org/news/2663.html>

Other Links:

<http://astro4.ast.vill.edu/mendel/mckusick.htm>

<http://www.jhu.edu/~jhumag/0400web/38.html>

http://www.sunjournal.com/story/247918-3/MaineNews/Maine_scholar_receives_prize_for_genetics/

<http://www.ncbi.nlm.nih.gov/omim/>