



American College of Medical Genetics Foundation

ACMG Foundation NEWS For Immediate Release

Contact: Kathy Beal, MBA
ACMG Media Relations Director
301-238-4582
Mobile 978-853-1810
kbeal@acmg.net

The Late Dr. Charles J. Epstein, Notable Geneticist, is Named 2011 ACMG Foundation Lifetime Achievement Award Recipient

BETHESDA, MD – March 18, 2011 | The late Dr. Charles J. Epstein, MD, FACMG, Founding Fellow and Past President of the American College of Medical Genetics (ACMG) is the 2011 recipient of the American College of Medical Genetics Foundation Lifetime Achievement Award.

Dr. Epstein, who lost his battle with pancreatic cancer on February 15th, was recognized for his extensive research on Down syndrome and other genetic disorders and for his part in gaining recognition for medical genetics by the American Board of Medical Specialists, which eventually led to the founding of the ACMG.

Though gravely ill and nearing the end of his fight with cancer, Dr. Epstein learned that he was the award recipient from his friend and colleague, Dr. R. Rodney Howell, MD, FACMG, President of the ACMG Foundation, who sits on the award selection committee.

“It was very clear that everybody was supportive of Charlie getting the award this year,” Dr. Howell said. “Once that was final, I called Charlie’s home,” said Dr. Howell. “I did talk to him briefly. He was very weak but very clear during our discussion. He was very pleased about the award. Later that day they made the decision to discontinue his treatment and go into hospice care. He died very soon after. It was the last conversation I had with him and (the award) was certainly one of the last things he learned about.”

After graduating first in his class at Harvard Medical School, Dr. Epstein began a career in medical genetics. Among his most notable accomplishments was the work he did with the help of his wife, Dr. Lois Epstein, on the study of Down syndrome.

Together with postdoctoral fellow David Cox, MD, PhD, and his wife Lois they developed a mouse model for the disorder. The model led researchers to begin identifying specific genes and proteins that play a role in the development of the disorder.

As a leader in medical genetics, Dr. Epstein was instrumental in establishing credibility for his field.

“The one thing that was most important occurred while he was head of the American Board of Medical Genetics. He worked very hard to get that board recognized by the American Board of Medical Specialties, which is the group that recognizes specialties such as surgery and pediatrics,” said Dr. Howell.

“That was an extremely important accomplishment and it had a significant impact on the field of medical genetics.”

-more-

American College of Medical Genetics Foundation • www.acmgfoundation.org

7220 Wisconsin Avenue, Suite 300, Bethesda, MD 20814 Telephone 301-718-2014 Fax 301-718-9604

Dr. Epstein received his MD from Harvard Medical School in 1959 and did his internship and residency at Peter Bent Brigham Hospital in Boston. He was Board Certified in Clinical Genetics in 1982.

He held positions at the National Heart Institute and a fellowship at the University of Washington School of Medicine in 1963 with Arno Motulsky, the recipient of the 2009 ACMG Foundation Lifetime Achievement Award. In 1967 he began his long tenure at the Department of Pediatrics at University of California San Francisco. During that time he trained many medical geneticists, including many recognized leaders in the field today. He was named Professor in 1972 and was appointed head of human genetics in the Pediatrics Department, a position he held until 2005.

In addition to his role at ACMG, he served on the boards of several professional societies, government panels and editorial boards.

Born in Philadelphia, Dr. Epstein attended Philadelphia Central High School, where he excelled not only in academics but also on the track and field team and as a cellist.

He attended Harvard College and graduated magna cum laude with an AB in chemistry in 1955. He and Lois who became a widely-known and talented physician and researcher both entered Harvard Medical School and were married in 1956. Charles graduated magna cum laude and at the top of his class at Harvard.

While he is perhaps best known for his work with Down syndrome, Dr. Epstein also did extensive research on cellular aging and how various enzymes affected the aging process. Because of these interests Dr. Epstein was appointed chairman of the scientific advisory board and board of trustees of the Buck Institute for Research on Aging.

He was the recipient of multiple awards, including the William Allan Memorial Award and the 2010 American Society of Human Genetics' Victor McKusick Leadership Award.

The McKusick Award was presented to Dr. Epstein by his friend and mentor, Dr. Arno Motulsky, MD, Emeritus Professor of Medicine (Medical Genetics) and Genome Sciences School of Medicine University of Washington.

In his remarks during the presentation Dr. Motulsky noted that Dr. Epstein, "fostered and enriched the development of various medical genetics disciplines and exemplified enduring leadership and farsighted vision that ensured the field of human genetics to flourish and be successfully assimilated into the broader context of science, medicine and health."

In addition to serving as president of the ACMG, Dr. Epstein also served as president and board director of the American Society of Human Genetics, and president and board director of the American Board of Medical Genetics. He was also editor of many professional journals and a member of a several government panels.

A prolific writer, Dr. Epstein produced more than 500 scholarly papers, authored and edited several books and served as a highly successful editor of The American Journal of Human Genetics for seven years.

While Dr. Epstein's life as a physician and researcher was his major focus, he made time to foster his love of art, music and travel. He played the violoncello in several orchestras, most recently the orchestra of the Bohemian Club in San Francisco. He and Lois were also world travelers and visited most of the major countries for both work and pleasure, collecting musical instruments that have been loaned to museums for special exhibitions.

-more-

Dr. Epstein's love of life and his belief in the value of every individual were well known among his colleagues and patients and he offered this advice to the parents of Down syndrome afflicted children:

"Love your child! Treat him or her as normally as possible and as a cherished member of the family...do the best that you can and try to take each day as it comes."

Dr. Epstein's career and life was disrupted severely when in June 1993, a package addressed to Dr. Epstein at his home exploded as he opened it. He sustained hearing loss, severe damage to his right hand, and internal injuries.

Dr. Howell said that he has talked with Dr. Epstein about the attack but what he remembers the most was Dr. Epstein's resilience. "The key thing was that he continued to be positive and profoundly productive well after that. He did not let it throw him off course, he simply moved ahead."

About the American College of Medical Genetics and ACMG Foundation

Founded in 1991, the American College of Medical Genetics (www.acmg.net) is the national non-profit professional organization that advances the practice of medical genetics by providing education, resources and a voice for physician geneticists, biochemical, clinical, cytogenetic, medical and molecular geneticists, genetic counselors and other healthcare professionals committed to the practice of medical genetics. ACMG's website (www.acmg.net) offers a variety of resources including Policy Statements, Practice Guidelines, Educational Resources, and a Medical Geneticist Locator. The educational and public health programs of the American College of Medical Genetics are dependent upon grants and contracts and charitable gifts from corporations, foundations, and individuals. The American College of Medical Genetics Foundation (www.acmgfoundation.org) is a 501(c)(3) not-for-profit organization dedicated to funding the College's diverse efforts to translate genes into health. The Foundation is dedicated to Better Health Through Genetics™

-end-